Curriculum Vitae

for

Professor John Christodoulou AM
MB BS PhD FRACP FFSc FRCPA CGHGSA

Director
Western Sydney Genetics Program
The Children's Hospital at Westmead
WESTMEAD NSW AUSTRALIA

&

Professor
Disciplines of Paediatrics and Child Health and Genetic Medicine
Faculty of Medicine
University of Sydney

Last updated – October 2013

John Christodoulou CV
2

Name: John CHRISTODOULOU

Street address: Western Sydney Genetics Program,
The Children's Hospital at Westmead,
Cnr of Hawkesbury Rd and Hainsworth St,
Westmead NSW 2145 Australia

Postal address: Western Sydney Genetics Program,
The Children's Hospital at Westmead,
Locked Bag 4001,
Westmead NSW 2145 Australia

Phone number: work (612) 9845 3452
fax (612) 9845 1864

email john.christodoulou@sydney.edu.au
john,christodoulou@health.nsw.gov.au

Qualifications:

1. MB BS (Honours class II) attained at the University of Sydney including:
   Distinctions in
   - physics
   - physiology
   - pharmacology
   - histology and embryology
   - paediatrics
   - biochemistry

   High distinction in - chemistry

   Year of graduation - 1981.

2. Fellow of the Royal Australian College of Physicians -admitted to Fellowship – April 1988.
3. Clinical Geneticist - certified by the Human Genetics Society of Australasia – July 1989.
4. Doctor of Philosophy - awarded by University of Melbourne Australia – March 1991.
5. Associate Membership of the Royal College of Pathologists of Australia – 2004.
6. Fellowship of the Royal College of Pathologists of Australasia (Biochemical Genetics) – 2006.
7. Founding Fellow of the Faculty of Science, Royal Collage of Pathology of Australasia – 2011.

Current scientific body and professional memberships:

American Society of Human Genetics
Australian Salaried Medical Officers' Federation
Australian Society for Biochemistry and Molecular Biology
Australian Society for Medical Research
Australasian Society for Inborn Errors of Metabolism
Genetics Society of Australia
Human Genetics Society of Australasia
Human Genome Variation Society (founding member)

Paediatric Research Society of Australia
Royal Australasian College of Physicians
Royal College of the Pathologists of Australasia
Society for Inherited Metabolic Diseases
Society for the Study of Inborn Errors of Metabolism
Human Genome Organisation
AHPRA Registration: MED0001055264 (medical practitioner with general and specialist registration)

Parent Support Group memberships:

The Association of Genetic Support of Australasia Inc
NSW PKU Parent Support Group
Rett Syndrome Association of Australia

Other courses attended:

1. 10 week introductory statistics course at the Macquarie University, NSW, Australia - 1985.
2. "The Skilled Helper" - A workshop to facilitate general counselling skills held by Mountain Psychological Associates, Mt. Dandenong, Victoria - 1988.
3. PhD thesis - "A Study of the role of malonyl CoA in normal and abnormal human metabolism" 1986 - 1990 (see below for further details).
4. ANGIS Course, New Children's Hospital, Westmead, 1998.
5. BioNavigator Course, University of Sydney, 2001.
6. Biotechnology Intellectual Property Training Course, Sprusen & Ferguson, Sydney 2001.
7. Introduction to SPSS Version 10 and Descriptive Statistics, University of Sydney, 2003

Positions held since graduation :

1981 - Intern at Westmead Hospital, Westmead, NSW, Australia.

1982 - Resident Medical Officer at the Royal Alexandra Children's Hospital, NSW, Australia.

1983 - Senior Resident Medical Officer at the Royal Alexandra Children's Hospital.

1984 - Registrar at the Royal Alexandra Children's Hospital.

1985 - Registrar at the Royal Alexandra Children's Hospital (including six months at the Prince of Wales Children's Hospital, Randwick, NSW).

1986 - Trainee Research Fellow, Birth Defects Research Institute, Royal Children's Hospital, Melbourne, Australia (1st year of PhD project - see below).

1987 - NH&MRC Postgraduate Medical Research Fellow, Murdoch Institute for Research into Birth Defects, Royal Children's Hospital, Melbourne, Australia (2nd year of PhD project).

Fellow in Clinical Genetics, Department of Genetics, Royal Children's Hospital.

1988 - NH&MRC Postgraduate Medical Research Fellow, Murdoch Institute for Research into Birth Defects, Royal Children's Hospital, Melbourne (ongoing PhD project - part-time).

Honorary Clinical Assistant, Department of Genetics, Royal Children's Hospital.

Relieving Honorary Clinical Assistant (Genetics), Royal Women's Hospital, Melbourne, Victoria, Australia.

Genetic Fellow, Foetal Diagnostic Unit, Monash Medical Centre, Melbourne, Victoria, Australia.

1989 - NH&MRC Postgraduate Medical Research Fellow, Murdoch Institute for Research into Birth Defects, Royal Children's Hospital (ongoing PhD project).

Honorary Clinical Assistant, Department of Genetics, Royal Children's Hospital.

1990 - NH&MRC Postgraduate Medical Research Fellow, Murdoch Institute for Research into Birth Defects, Royal Children's Hospital (ongoing PhD project).

Honorary Clinical Assistant, Department of Genetics, Royal Children's Hospital.

Relieving Honorary Clinical Assistant (Genetics), Royal Women's Hospital, Melbourne, Victoria, Australia.

Honorary Clinical Assistant, Foetal Diagnostic Unit, Monash Medical Centre, Melbourne, Victoria, Australia.

1990 - 1992 Fellow in Metabolic Diseases, Division of Clinical Genetics, Hospital for Sick Children, Toronto, Ontario, Canada (started July 1990).

July 1992- Senior Lecturer, Dept of Paediatrics and Child Health, University of Sydney (until March 1993).

VMO at the Children's Hospital, Camperdown NSW.

Consultant Medical Officer in Biochemical Genetics, King George V Memorial Hospital, Camperdown, NSW.

Honorary Medical Affiliate in Biochemical Genetics, Westmead Hospital, Westmead, NSW.

1993 - Honorary consultant metabolic physician, N.S.W. Biochemical Genetics Service, Oliver Latham Laboratory, North Ryde, NSW.

1995 - Honorary Specialist, Prince of Wales Children’s Hospital, Randwick NSW.

1995 - Honorary Staff Specialist (Dept of Endocrinology), Westmead Hospital, Westmead, NSW.

1996 - Honorary Staff Specialist (Prenatal Diagnosis and Genetics), Westmead Hospital, Westmead, NSW.

1997 - Director, Western Sydney Genetics Program, Royal Alexandra Hospital for Children, Westmead, NSW.

1997 - Associate Professor, Dept of Paediatrics & Child Health, Faculty of Medicine, University of Sydney.

2003 – Honorary Consultant in Clinical Genetics, Westmead Hospital

2004 – Promoted to Professor, Discipline of Paediatrics & Child Health, Faculty of Medicine, University of Sydney

2005 – Honorary Consultant Clinical Geneticist, Nepean Hospital.

2008 – Professor, Discipline of Genetic Medicine, Faculty of Medicine, University of Sydney.

2008 – Appointed as Honorary CMRI Scientist, Children's Medical Research Institute, Sydney.

Other Responsibilities: (since 1992)

University:

1. ***Teacher of Undergraduate Paediatric Training at the Royal Alexandra Children's Hospital.*** (1992-1997).
   Responsibilities included co-ordination of the teaching program for the students, regular lectures to the students, clinical tutorials, and organization of and participation in examinations in Paediatrics for the medical students of the University of Sydney

2. ***Postgraduate Student Co-ordinator, Dept of Paediatrics & Child Health, University of Sydney*** (1993-1999).

3. ***Faculty Advisor for Option Term students in Paediatrics, Royal Alexandra Children's Hospital*** (1993-1997).

4. ***Co-convenor University of Sydney Faculty of Medicine Clinical Genetics Update for the Medical Practitioner*** - Sydney, October, 1993.

5. ***Committee member, Molecules and Cells Planning Committee, Graduate Medical Degree Course, University of Sydney*** (1994).

6. ***Committee member, Human Inheritance Planning Committee, Graduate Medical Degree Course, University of Sydney*** (1994).

7. ***Case Co-ordinator, Case 1.02 (Phenylketonuria), Introductory Block, Graduate Medical Degree Course, University of Sydney*** (1995-2007).

8. ***Committee member, Interim Paediatric Undergraduate Medical Course, University of Sydney*** (1994-1997).

9. ***Committee member, Computer Based Education Committee, Faculty of Medicine, University of Sydney*** (1995-1996).

10. ***Member, Information Technology Committee, Faculty of Medicine, University of Sydney*** (1996).

11. ***Chairman, Combined Boards of Postgraduate Studies, Faculties of Dentistry, Medicine and Pharmacy, University of Sydney*** (April 2000 – 2010).

12. ***Member, PhD Awards Subcommittee, University of Sydney*** (April 2000 – 2012).
The terms of reference of this committee are: to advise the Graduate Studies Committee on resolutions, policy and procedures relating to the award of PhD and other higher degrees including professional doctorates; to take decisions in accordance with the resolutions of the Senate and Academic Board; to develop, oversee and monitor processes for the effective implementation of resolutions; to provide an annual report on its activities and to consider any matter referred to it by the Academic Board , the Vice-Chancellor, the Graduate Studies Committee or the Chair of the Academic Board. Due to come off this committee in March 2012.

13. ***Member, Graduate Studies Committee, University of Sydney*** (April 2000 – December 2006).
The terms of reference of this committee are: to advise the Academic Board on policies relating to all graduate coursework and research studies in the University, including the pattern of graduate programs and student outcomes, determining criteria for the selection for postgraduate awards, establishing effective supervisory practices, and advising about appropriate facilities for students.

14. ***Chairman, Working Party to review research candidatures in the Faculty of Medicine, University of Sydney*** (April 2000 – December 2000).

15. ***Member of Faculty of Medicine Education Committee, Sydney University*** (July 2001 – 2002).
The terms of reference of this committee are to consider matters relating to the education programs, including resource issues, in which the Faculty is involved and to provide comment or advice either on the Committee's initiative or upon reference to the Committee by the Dean, the Academic Board or other appropriate person or body.

16. ***Member, Combined Degree Program Committee, Faculty of Medicine, University of Sydney*** (November 2001 – 2002).
This committee is responsible for the development and management of the Combined Degree Program, including selection of candidates for the program.

17. ***Associate Dean (Postgraduate Studies), Faculty of Medicine*** (Jan 2002 – March 2013).

18. ***Member, Postgraduate Research Training Subcommittee*** (June 2002 – December 2006).
The terms of reference of this committee include: to advise the Graduate Studies Committee on policies relating to all graduate research studies in the University including attraction and recruitment of students, admissions, equity and access initiatives, assessment and examinations, research components of coursework programs, annual review processes, provision of facilities and services, approval of new graduate research programs and approval of changes to existing programs, establishment of effective supervisory practices.

19. ***Member Faculty of Medicine Research Committee*** (Jan 2003 – 2009)
The terms of reference of this committee include: to improve coordination of research and to promote collaborative activities within and between faculties of the College, including cross-faculty applications for major equipment; to develop and recommend policy and procedures supporting the University and College strategic objectives in relation to research, and to identify resource implications of these policies; to monitor issues relating to quality in

research and research activities, and to make recommendations; to maintain and publicise lists of research interests, expertise and equipment within the College; to review applications for Sesqui grants.

20. ***Appointment to Working Party to Review Academic Promotions Processes*** (March 2003 – December 2003)

21. ***Member, Research and Research Training Subcommittee*** (December 2006 – 2009)

22. ***Faculty of Medicine Office of Research and Research Training Committees (Go5)*** (2008 – 2009)

This committee was formed following a restructure of the various agencies within the Faculty of Medicine that are responsible for the administration of postgraduate research and coursework students. Its terms of reference are to advise the Dean of the Faculty of Medicine in relation to research strategy, and to support and promote research and research training in the Faculty of Medicine.

23. ***Member, Research Training Committee of the University of Sydney Senior Executive Group*** (2009)

24. ***Associate Dean (Research Integrity), Sydney Medical School, University of Sydney*** (April 2013 – current)

25. ***Head, Discipline of Genetic Medicine, Sydney Medical School, University of Sydney*** (April 2013 – current)

Hospital:

26. ***Member of Institutional Biosafety Committee, Royal Alexandra Children's Hospital*** (1992-current).

27. ***Co-convenor Children's Hospital Postgraduate Meeting*** - Sydney, October, 1993.

28. ***Convenor of the Paediatric Research Seminars*** (1993-1996).

These seminars were instigated as a forum where clinical and laboratory researchers could meet on a weekly basis to present ongoing research in paediatrics. Participation by individuals from the Royal Alexandra Children's Hospital, Dept of Paediatrics, Westmead Hospital, and Children's Medical Research Institute has been encouraged and developed.

29. ***Committee member, RAHC/Westmead Shared Services Committee for Genetic Services***, 1994.

30. ***Co-convenor Royal Alexandra Children's Hospital Postgraduate Meeting*** - Sydney, October, 1994.

31. ***Co-convenor Royal Alexandra Children's Hospital Paediatric Update Meeting*** - Sydney, 1994 – 2000.

32. ***Committee member, Royal Alexandra Children's Hospital Postgraduate Education Subcommittee*** (1994 – 1995).

33. ***Acting Head, Department of Genetics, Royal Alexandra Children's Hospital*** (October 1994-October 1995).

34. ***Head, PKU Clinic, Royal Alexandra Children's Hospital*** (May 1995-current).

35. ***Committee member, Medical Education Subcommittee, Royal Alexandra Hospital for Children*** (September 1995-1999).

36. ***Member, Research Policy and Procedure Committee, Royal Alexandra Hospital for Children*** (1996 - 1997).

37. ***Director, Western Sydney Genetics Program, Royal Alexandra Hospital for Children*** (1997 – current)

    This is the only fully integrated genetics service in NSW, with over 100 staff, and incorporating clinical service arms (Department of Clinical Genetics and Genetics Metabolic Diseases Service) and laboratory diagnostic arms (including Departments of Cytogenetics and Molecular Genetics, and the NSW Biochemical Genetics Service and NSW Newborn Screening Programme, the latter two being state-wide services).

38. ***Member, Research Committee, Children's Hospital at Westmead*** (January 2002 – December 2004).

    The role of this committee is to promote an inquiring approach to the problems of children and to advise the Board on the use of research funds.

39. ***Clinical Director, Departments of Cytogenetics and Molecular Genetics*** (November 2004 – current)

In addition, I am periodically asked to sit on interview panels for senior and junior medical appointments at the Children's Hospital at Westmead.

Professional Organisations:

40. ***Committee Member, Human Genetics Society of Australasia*** (NSW Branch, 1993-current).

41. ***Vice President, NSW Branch of the Human Genetics Society of Australasia*** (August 1995).

42. ***President, Human Genetics Society of Australasia*** (NSW Branch) (1997 – 1999).

43. ***Honorary Secretary, Human Genetics Society of Australasia*** (NSW Branch) (1999 – 2004).

44. ***Member of the Genetics Services Subcommittee of the Human Genetics Society of Australasia*** (1997 - current).

45. ***Co-convenor 9th International Conference on Pteridines and Related Biogenic Amines*** - Port Douglas, 1996.

46. ***Co-convenor 5th Australian ASIEM Conference*** - Canberra, September 1993.

47. ***Member of Steering Committee, International Congress of Human Genetics*** (held in Brisbane, 2006) (1999 – 2006).

48. ***Chairman, Organising Committee, Human Genetics Society of Human Genetics Annual Scientific Meeting*** – Sydney, 1999.

49. ***Member, Human Genetics Society of Australasia Annual Scientific Meetings Scientific Advisory Committee*** (2000 – current).

50. ***Member, Editorial Advisory Board, Journal of Inherited Metabolic Disease*** (January 1996-current).

51. ***Chair, Scientific Program Committee, International Congress in Inborn Errors of Metabolism, 2003***, held in Brisbane (2001 – 2003).

52. ***Vice President, Human Genetics Society of Australasia*** (2003 – 2005)

53. ***Elected to the International Scientific Advisory Committee of the Cyprus Institute of Neurology and Genetics*** (January 2005)

54. ***President Human Genetics Society of Australasia*** (2005 – 2007)

55. ***Examiner – RCPA Part 1 Examination in Genetics*** (2005)

56. ***Member – Pathology Associations Council*** (2007 – current)

57. ***Member – International Rett Syndrome Foundation Scientific Review Board*** (2008 – current)

58. ***Member – International Rett Syndrome Foundation Executive Committee*** (April 2009 – current)

59. ***Chair – Joint HGSA/RCPA Molecular Genetics Quality Assurance Program*** (August 2009 – current)

60. ***Member – Local Organising Committee of the Human Genome Meeting 2012*** (February 2010 – current)

61. ***Member – Scientific Advisory Committee, Orphanet Australia*** (July 2013 – current). Providing expertise in the development of Australian content for Orphanet.

Governmental Advisory Bodies:

62. ***Member of the DNA Working Party of the NSW Genetic Service Advisory Committee***. (1992-August 1996).

63. ***Committee member, NSW Newborn Screening Advisory Committee*** (1994-current).

64. ***Member, NSW Genetic Services Advisory Committee*** (October 1994-December 1995).

65. ***Chairman of the DNA Working Party of the NSW Genetic Service Advisory Committee*** (August 1996-current).

66. ***Leader, Health Delivery, Technology and Research Working Group of the NHMRC Human Genetics Advisory Committee*** (April 2006 – December 2006)

67. ***Member, NHMRC Project Grants Review Panel in Genetics*** (2006, 2007, 2008)

68. ***Member, NHMRC Human Genetics Advisory Committee*** (August 2009 – current)

Community Organisations:

69. ***Board Member, VCFS Foundation of Australia*** (August 1996-1997).

70. ***Member of International Rett Syndrome Association Scientific Advisory Committee*** (2001 – 2006).

71. ***Sydney University Fencing Club, Senior Vice President*** (2002)***, Technical Director*** (2003)

72. ***Member of International Rett Syndrome Foundation Scientific Review Board*** (2009 – current)

73. ***Member RettSearch Executive*** (2010 – current)

74. ***Member of the Australian Fencing Federation Medical Commission*** (June 2009 – current)

PhD Thesis:

In 1986 I commenced a PhD project studying the role of malonyl CoA in normal human metabolism. The role of malonyl coenzyme A in normal and abnormal metabolic states was studied in detail. These studies centred on one of the major enzymes involved in its regulation, namely malonyl-CoA decarboxylase (MCD).

MCD was purified to near homogeneity from beef liver. Unfortunately, inadequate amounts were purified to enable the production of antibodies to the enzyme and thereby allow molecular studies to be performed.

The kinetic characteristics of MCD were studied with the enzyme source being cultured skin fibroblasts from controls and two children with MCD deficiency of varying clinical and biochemical severity. It was found that the K_m and V_{max} of the enzyme from the two children was different from the control. The inhibitory effect of a number of CoA thioesters was studied in control fibroblasts. Of note it was found that palmitoyl-CoA and acetoacetyl-CoA were potent inhibitors of MCD, offering experimental evidence for the observed biochemical derangements in the MCD-deficient children.

A peroxisomal form of MCD was identified which has a different K_m from the mitochondrial form and which is inducible by the hypolipidaemic drug clofibrate. The two forms of the enzyme showed similar heat inactivation profiles and pH optima, suggesting that they might have very similar peptide structures.

The regulatory role of malonyl-CoA on a number of enzymes of intermediary metabolism was studied in cultured fibroblasts, with particular emphasis on those aspects seen to be abnormal in the MCD-deficient children.

These findings raise further questions about the molecular origin of MCD and the role it plays in the peroxisomal milieu and in other metabolic processes.

Subsequently, Dr David FitzPatrick has cloned the human MCD gene, and has indeed found that there are leader sequences targeting the enzyme to both the mitochondrion and the peroxisome.

Genetics Training:

From 1986 I was involved in a formal genetics training program facilitated through the Department of Genetics at the Royal Children's Hospital, Melbourne, and accredited by the Human Genetics Society of Australasia. I attended a formal genetics lecture series for second year genetics students held at the University of Melbourne. In addition, in 1988 I attended a workshop entitled "The Skilled Helper" which was devoted to developing general counselling skills.

Between 1986 and 1989 I gained practical clinical experience in basic genetic principles, dysmorphology, biochemical genetics, molecular biology and its practical applications, and the interpretation of cytogenetic results.

In 1989 I was also involved in the co-ordination of a weekly seminar series in various genetic and metabolic topics, at which I myself presented a dozen or so topics.

In July 1989 I was granted status as a medical geneticist as accredited by the Board of Censors of Human Genetics Society of Australasia.

From July 1990 to June 1992 I was the Fellow in Metabolic Diseases, in the Dept of Clinical Genetics at the Hospital for Sick Children, Toronto Canada. I was actively involved in the clinical metabolic service, and was able to obtain further laboratory experience in the field of molecular biology (see below).

Other Research Studies:

Mutation Analysis in Human Argininosuccinic Acid Lyase Deficiency: (September 1991-June 1992 Toronto)

Research on argininosuccinic acid lyase (ASAL) deficiency had been focussed on three aspects. Firstly, we were in a position to identify the range of mutations at this locus for a relatively large cohort of patients. Secondly, previous studies had demonstrated the existence of intragenic complementation in ASAL deficiency. The work I was involved in was the start of the dissection of the molecular basis for this intragenic complementation. Thirdly, as a consequence of these and other studies it is hoped that a broader understanding of the biochemical basis for the clinical heterogeneity in ASAL deficiency will be gained.

ASAL deficiency is an autosomal recessive defect in the urea cycle. It is notable for marked phenotypic variability, with the clinical presentation falling into three broad groups: neonatal, subacute, and late onset. Some of this clinical heterogeneity can be explained by residual enzyme activity, but most cannot. ASAL deficiency has been

reported in all ethnic groups, and the genetic fitness of heterozygotes is not increased. These features therefore make it likely that most affected individuals would be genetic compounds, except of course for the offspring of consanguineous matings.

Complementation studies were performed in our laboratory using 28 ASAL deficient cell lines, with two conclusions being drawn. Firstly, all of the cell lines were part of a single complementation group. In other words, only defects in the ASAL gene were responsible for the phenotype in these patients. Secondly, intragenic complementation was found, and it was possible to further divide the cell lines into 12 complementation subgroups. The similar complementation behaviour of strains in a subgroup suggests that these strains have mutations that affect the complementation event in a similar way.

One subgroup of particular interest is the so-called frequent complementers. These three cell lines take part in 30 of the 32 positive complementation events. We had been concentrating our efforts initially on defining the mutations in these cell lines. We identified the mutations in these three cell strains; the data identify the two alleles responsible for the majority of the complementation events at this locus, and provide molecular confirmation of the complementation analysis.

Vectors containing the mutations of interest were developed, and transient expression studies were performed, confirming that the mutations identified did indeed disrupt ASAL activity, and determined that complementation sometimes occurs *in vivo* in patients with ASAL deficiency.

I also identified a possible deletional "hotspot" in the ASAL gene. Five alleles of the 28 cell strains were found to have mutations that lead to skipping of one of the exons. This leads to an 84bp in-frame deletion of the cDNA, which would result in shortening of the ASAL monomer by 28 residues. In these patients genomic DNA has been analysed. Four have a 13bp deletion within the exon, which is skipped. This deletion, which is flanked by a 2bp (GC) direct repeat, ends 5bp upstream of the 3' splice site junction and does not remove any sequence known to be critical for normal splicing. The other cell strain has a 25bp deletion which begins in the exon at precisely the same position as the start of thew 13bp deletion, and continues through the splice junction. Notably, both deletions begin at the cut site for TopoII, and occur in a strong consensus sequence for the putative Topo II recognition sequence. In addition, the consensus sequence for a Pol-α mutation site overlaps with the Topo II site and the start of both deletions. It may be, therefore, that the putative Topo II and Pol-□α sites may predispose to deletions in this region. This work was presented at the 1992 American Society of Human Genetics conference in San Francisco, for which I won the award for the best postdoctoral student presentation

The above work is the subject of three papers which have been published.

Awards and Prizes:

1. Royal Children's Hospital (Melbourne) Trainee Research Fellowship -1986.
2. National Health and Medical Research Council of Australia Postgraduate Medical Research Fellowship -1987-1990.
3. Royal Australasian College of Physicians Scholarship Fund Travelling Fellowship –1990.
4. Sandoz Paediatric Travelling Fellowship – 1990.
5. Travelling Fellowship, English Speaking Union (NSW Branch) – 1990.
6. Ian Potter Foundation Travelling Scholarship – 1992.

7. American Society of Human Genetics Postdoctoral Clinical Research Student Award, 1992.
8. Winner Best Poster at the Annual Scientific Meeting of the Paediatric Research Society of Australia & Australian College of Paediatrics, Adelaide, May 1995 (Prenatal diagnosis of the carnitine transporter defect).
9. Vice-Chancellor's Award for Excellence in Postgraduate Research Higher Degree Supervision, 2004.
10. Became a Member of the Order of Australia (AM), January 2010. Awarded in recognition of my contributions to genetic practice and research, particularly in the area of genetic metabolic disorders.
11. Ippokratis Award for outstanding achievement by a medical professional. Awarded by the Australasian Hellenic Educational Progressive Association (AHEPA), March 2010 .

Current Projects:

The Biology of Rett Syndrome*: MECP2* Mutations and Beyond

Rett syndrome is the second most common (after Down syndrome) cause of severe intellectual disability in females. It is caused by mutations in the methyl CpG-binding protein 2 (*MECP2*). MeCP2 is a transcriptional repressor, although more recent studies suggest it may also have a role as a gene activator, and may be involved in the regulation of splicing.

Ours studies have focussed on:

- Identification of genotype-phenotype correlations
- Identification of MeCP2 targets using whole genome approaches such as micro-arrays and 2D gel electrophoresis
- Development of mouse models and their study (in collaboration with Professor Patrick Tam, CMRI

In addition, our group in collaboration with groups in Adelaide, Perth and Cardiff, discovered the second gene that is associated with a Rett-like phenotype, cyclin dependent kinase-like 5 (CDKL5). We are currently studying the functional role of CDKL5 and its possible association with MeCP2.

The Molecular Pathogenesis of Mitochondrial Respiratory Chain Disorders

Defects of the mitochondrial respiratory chain result in a deficiency of ATP production, and this can have severe effects on human metabolism. Mutations in one of over 200 proteins can lead to a functional defect of the respiratory chain, the genes for which are encoded in either nuclear or mitochondrial genes. We are currently studying the use of forced myogenesis of patient fibroblasts to aid in their diagnosis and prenatal diagnosis.

Genetically Engineered Probiotics for the Treatment of Human Disease: Phenylketonuria as a Model Disease

Phenylketonuria (PKU) due to phenylalanine hydroxylase deficiency, if untreated, results in profound intellectual disability, seizures and aggressive behaviour. Treatment consists of a life-long diet, restricting phenylalanine intake. The diet however is unpalatable, and for a number of reasons, compliance tends to deteriorate with age. A group led by me, in collaboration with Dr Ian Alexander of the Gene Therapy Research Unit (Children's Hospital at Westmead), is developing a novel therapy for treating PKU. This will involve genetically engineering harmless *Lactobacillus*

bacteria to produce an alternate phenylalanine-metabolising enzyme, phenylalanine ammonia-lyase. These engineered organisms, if given orally (as a "Yakhult"™ type preparation), should metabolise phenylalanine in the small intestine, preventing blood levels from rising. If successful, this novel form of treatment could be translated to other human diseases, and could have wide applicability in the animal husbandry industry.

In addition, we have been undertaking mutational analysis of the phenylalanine hydroxylase gene in our NSW cohort of PKU patients, with a particular emphasis being to identify those individuals who are likely to be responsive to the cofactor tetrahydrobiopterin.

Application of Next Generation Sequencing Technologies for Mendelian Gene Disorder Discoveries
Being located in the major paediatric centre in NSW, we see many families with presumed genetic disorders with a high likelihood of having a Mendelian inheritance. Technical improvements in the use of next generation sequencing (NGS) technologies, and a dramatic fall in the cost in NGS, has now opened up the unprecedented opportunity for gene discovery in rare Mendelian disorders. Our group is using these technologies for gene discovery in the area of mitochondrial respiratory chain disorders, and for rare apparently Mendelian disorders. Furthermore, our laboratory has the capability to undertake functional studies to confirm pathogenicity of novel presumed pathogenic gene mutations.

Competitive Research Funding:

Royal Australasian College of Physicians Epilepsy Research Grant - 1994
Molecular Analysis of Pyridoxine-dependent Epilepsy
John Christodoulou (Chief Investigator A)
Rod McInnes (Chief Investigator B) $40,000

Children's Hospital Fund Research Grant - 1994
Molecular Analysis of Pyridoxine-dependent Epilepsy
John Christodoulou (Chief Investigator A)
Rod McInnes (Chief Investigator B) $14,000

Children's Hospital Fund Small Grant - 1995
Molecular Studies of Galactosaemia
John Christodoulou (Chief Investigator A)
Veronica Wiley (Chief Investigator B)
Bridget Wilcken (Chief Investigator C) $9,976

Children's Hospital Fund Research Grant - 1995
Functional & Molecular Analysis of Defects of the Electron Transport Chain
John Christodoulou (Chief Investigator A)
John Coakley (Chief Investigator B) $15,000

Financial Markets for Children - 1995
Functional & Molecular Analysis of Defects of the Electron Transport Chain
John Christodoulou (Chief Investigator A)
John Coakley (Chief Investigator B) $15,000

NHMRC - Project Grant - 1995 - 1998
Molecular Characterisation of Birth Defects on Human Chromosome 22
Alison Colley (Chief Investigator A)
Kerry Fagan (Chief Investigator B) $43,779

Meredith Wilson (Chief Investigator C) $43,785
John Christodoulou (Chief Investigator D) $43,691

Children's Hospital Fund Research Grant - 1996
Functional & Molecular Analysis of Defects of the Electron Transport Chain
John Christodoulou (Chief Investigator A)
John Coakley (Chief Investigator B) $30,810

Apex Foundation for Research into Intellectual Disability - 1996
Rett Syndrome:Diagnostic Evaluation and Therapeutic Strategies
John Christodoulou (Chief Investigator A) $20,000

Financial Markets for Children - 1996
Molecular Studies of Galactosaemia
John Christodoulou (Chief Investigator A)
Veronica Wiley (Chief Investigator B)
Bridget Wilcken (Chief Investigator C) $47,250

Children's Hospital Fund Research Grant - 1997
Functional & Molecular Analysis of Defects of the Electron Transport Chain
John Christodoulou (Chief Investigator A)
John Coakley (Chief Investigator B) $32,080

Children's Hospital Fund Research Grant - 1997
Evaluation of a Novel Screening Procedure for Patients with Defects of the Mitochondrial Respiratory Chain
Andrew Williams (Chief Investigator A)
John Christodoulou (Chief Investigator B) $5,003

NHMRC - Project Grant - 1998 - 2000
Biochemical and Molecular Genetic Evaluation of Multiple Respiratory Chain Defects
John Christodoulou (Chief Investigator A) $49,148
Ian Trounce (Chief Investigator B) $50,618
$52,084

Ramaciotti Foundation Research Grant – 2000
Genetically Engineered Probiotics for the Treatment of Human Disease: Phenylketonuria as a Model Disease
John Christodoulou (Chief Investigator A)
Ian Alexander (Chief Investigator B)
Ted O'Loughlin (Chief Investigator C) $12,000

Children's Hospital Fund – Near Miss Funding grant - 2001
Biochemical and Molecular Genetic Evaluation of Multiple Respiratory Chain Defects
John Christodoulou (Chief Investigator A)
Andrew Williams (Chief Investigator B)
Bruce Bennetts (Chief Investigator C)
Robert Newbold (Chief Investigator D) $50,000

International Rett Syndrome Association – 2001 - 2003
Establishment of the IRSA MECP2 Mutation Database
$50,000
John Christodoulou (Chief Investigator A)
$25,000

Bruce Bennetts (Chief Investigator B)
$31,000

International Rett Syndrome Association – 2006 - ongoing
Maintenance of the IRSA *MECP2* Mutation Database
$19,500USD

John Christodoulou (Chief Investigator A)
per annum
In recognition of the importance of this locus specific database, the IRSA has agreed to provide funding on an annual basis in perpetuity to enable ongoing maintenance and enhancement of the database.

Muscular Dystrophy Association of the USA – 2001 - 2002
Molecular & Genetic Investigations of Autosomal Dominant External Ophthalmoplegia
Henrik Dahl (Chief Investigator A) $89,203US
John Christodoulou (Associate Investigator) $60,413US

Cecilia Kilkeary Foundation Grant – 2001
Molecular Pathogenesis of Mitochondrial Respiratory Chain Disorders
John Christodoulou (Chief Investigator A) $9,500
Andrew Williams (Chief Investigator B)

Rett Syndrome Research Foundation Postdoctoral Fellowship – 2001 - 2002
Assessment of Central Autonomic Nervous System in Rett syndrome
Carolyn Ellaway (Chief Investigator A) $43,850US
John Christodoulou (mentor)

NHMRC Project Grant – 2002 - 2004
Pathogenesis of Rett Syndrome: Molecular Genetic and Animal Models
John Christodoulou (Chief Investigator A) $145,000
Patrick Tam (Chief Investigator B) $145,000
Bruce Bennetts (Chief Investigator C) $145,000
Cathy Watson (Chief Investigator D)

NHMRC Project Grant – 2003 - 2005
Metabolic Complications of Obstructive Sleep Apnea During Early $105,000
Development $110,000
Karen Waters (Chief Investigator A) $90,000
Louise Baur (Chief Investigator B)
John Christodoulou (Chief Investigator C)
Dr D de la Eva (Chief Investigator D)

NHMRC Project Grant – 2003 - 2005
Evaluation of the Effectiveness of Expanded Newborn Screening $125,000
By Tandem Mass Spectrometry $115,000
Bridget Wilcken (Chief Investigator A) $115,000
John Christodoulou (Associate Investigator)

Abbott/Australian Society of Anaesthetists Research Grant – 2003
Mutation Screening of the Ryanodine Receptor Gene in Malignant Hyperthermia $35,000
Margaret Perry (Chief Investigator)
John Christodoulou (Supervisor/Mentor)

James N. Kirby Foundation – 2004 – 2005
Funds for the purchase of a quantitative PCR machine $13,000
John Christodoulou (Chief Investigator) $12,000

The University of Sydney Sesqui Research and Development Fund – 2004
$160,000
BD FACScan cell sorter upgrade (DiVa) option
awarded to: G. Farrell, T. Cunningham, R. Kefford, B. Henderson, A. Abendroth,
L. Bendall, D. Harris, L. Sedger, I. Alexander, K. North, C. Jones, J. Byrne,
J. Christodoulou, P. Gunning, S. Alexander

NHMRC Project Grant – 2004 - 2008
Genetic, Family and Social Determinants of the Burden and Outcome $154,450
in Rett Syndrome: a Population-based Investigation $164,925
H Leonard, N de Klerk, J Christodoulou, S Reilly, C Ellaway, D Ravine, $144,450
S Fyfe, S Silburn $154,925
$86,200

NHMRC Project Grant – 2005 - 2007
STK9, a second Rett syndrome gene: genetic and functional studies $154,250
John Christodoulou (Chief Investigator A) $154,250
$154,250

Rett Syndrome Research Foundation Grant – 2005
STK9, a second Rett syndrome gene: genetic and functional studies $48,500USD
John Christodoulou (Chief Investigator A)

The University of Sydney Sesqui Research and Development Fund – 2005
$107,500
"Eridan 64 Automatic Immunostaining machine from DakoCytomation"
awarded to: T. Cunningham, C. Clarke, C. Liddle, J. Fletcher, P. Harnett,
J. Christodoulou, A. DeFazio, R. Balleine, L. Bendall, G. Rangan, W. Hawthorne,
L. Sedger, A. Abendroth, S. Fleming, J. Byrne, D. Little

Rett Syndrome Research Foundation Postdoctoral Fellowship – 2005 – 2006
Investigation of the impact of regionalized *Mecp2* deficiency $50,000USD
and the manifestation of the RTT phenotype using chimera analysis
$50,000USD
Recipient Dr Greg Pelka (Mentors John Christodoulou and Patrick Tam)

Muscular Dystrophy Association of NSW – 2006 $45,000
The utility of forced myogenesis of cells in culture: unmasking

mitochondrial respiratory chain disorders.
John Christodoulou (Chief Investigator A)

Ian Thorpe Foundation for Youth Postdoctoral Fellowship **– *2006 – 2007*** $65,811
Recipient Dr Desiree Cloosterman $56,273

NHMRC Project Grant – 2007 - 2009 Grant # 457238
Functional Significance of MeCP2 Target Genes in the $152,750
Pathogenesis of Rett Syndrome
$152,750
John Christodoulou (Chief Investigator A) $152,750
Patrick Tam – Associate Investigators

Australian Rotary Health Research Fund – 2008 - 2010
Development of Novel Treatment Strategies for Phenylketonuria $54,000
$54,000
$59,400

March of Dimes Birth Defects Foundation – 2008 – 2010 Grant # 1-FY08-418
Unmasking Mitochondrial Respiratory Chain (RC) Disorders by $80,755
Forced Myogenesis of Cultured Cells $87,506
$94,447

Clive and Vera Ramaciotti Equipment Grant – *2008*
Real-time amplification system (QPCR) and high resolution melt (HRM) $30,000
- Gene Expression and Analysis Facility KN North, J Christodoulou, PW Gunning, J Byrne

NHMRC Project Grant – 2009 - 2011 Grant # 570752
Characterisation of novel CDKL5 targets: implications for $135,125
Rett Syndrome and related Neurodevelopmental disorders.
$135,125
John Christodoulou (Chief Investigator A) $135,125
Patrick Tam (Chief Investigator B)
Phillip Robinson and Gregory Pelka – Associate Investigators

International Rett Syndrome Research Foundation Grant – 2010 - 2012 Grant # 2442
Development and maintenance of RettBASE: IRSF *MECP2* $36,528 USD
Variation Database
2012 $35,165
USD

Clive and Vera Ramaciotti Equipment Grant – *2010*
Correlative Light and Electron Microscopy Suite $75,000
C Jones, G O'Neill, KN North, J Byrne, J Christodoulou, R Murray, S Cooper, L Cantrell

NHMRC Project Grant – 2011 - 2013 Grant APP1008021
In vivo gene transfer and phenotype correction of normal and urea-cycle deficient primary human hepatocytes in chimeric mouse-human livers: Towards gene therapy for metabolic liver disease
$476,706

Ian Alexander, Filip Braet, John Christodoulou, Philip Kuchel, Kevin Carpenter

NHMRC Project Grant – 2011 - 2013 Grant APP1004384
Towards evidence-based care for Rett syndrome: a research model to inform management of rare disorders
$441,972
Helen Leonard, Elizabeth Elliott, John Christodoulou, Jenny Downs, Carolyn Ellaway, Peter Jacoby, Ian Torode

University of Sydney NHMRC Equipment Grant – 2011
Ion Torrent Personal Genome Machine – DNA sequencer $75,000

ARC Linkage Grant – 2011 – 2014 Grant LP110200277
Psychosocial and economic impacts of rare diseases on Australian children, families and health professionals $316,780
Yvonne Zurynski, Elizabeth Elliott, Helen Leonard, John Christodoulou

NHMRC Project Grant – 2012 - 2014 Grant APP1026891
Gene Discovery And Functional Studies To Reveal Mechanisms Underlying Mitochondrial Respiratory Chain Disorders $368,510
John Christodoulou, Sandra Cooper

Funding Applications Pending:
NHMRC Project Grant – 2014 - 2016 Grant APP1063440 $519,838
Restoring Microtubule Acetylation in Rett Syndrome: Evaluation of an Emerging Therapeutic Agent

NHMRC Project Grant – 2014 - 2016 Grant APP1063348 $479,838
Mendelian Disease Gene Discovery Using Next Generation Sequencing Technologies

NHMRC Practitioner Fellowship – 2014 - 2018 Grant APP1059101
Implementation of Next Generation Sequencing Technologies for Improving Genetic Diagnosis and Treatment.

NHMRC CRE Grant – 2014 - 2018 Grant APP1061507 $2,457,000
A Co-operative National Centre for Research Excellence in intellectual Disability

International Rett Syndrome Research Foundation Grant – 2013 - 2015 $150,000
Preclinical Evaluation of Tubastatin A, a Novel Therapy for Rett Syndrome

Since 1994 I have held over $7,000,000 in competitive funding.

Other Funding:
Sponsored Research Project (Scientific Hospital Supplies International Ltd)
Evaluation of Dietetic Management of Phenylketonuria (PKU) due to Phenylalanine Hydroxylase Deficiency $87,000
John Christodoulou (Chief Investigator)

Sponsored Research Project (PharmaResearch Pty Ltd)
A Phase-II Open Label Clinical Study of the Efficacy and Safety of Recombinant Human N-acetylgalactosamine 4-sulphatase (rhASB) Enzyme Replacement Therapy in Patients with Mucopolysaccharidosis type VI (Maroteaux-Lamy Syndrome) $54,000USD
David Sillence (Principal Investigator)
John Christodoulou (Sub Investigator)

Sponsored Research Project (Shire Genetic Therapies Inc)

Rett Syndrome Mouse Model Tubastatin A studies	2012	$236,000
	2013*	$166,000

* subject to satisfactory progress in year 1

Donated funds to support my research into Rett syndrome

- from the Rett Syndrome Association of NSW since 1996 $130,000
- from the Country Women's Association of NSW since 1998 $65,000
- from the Rett Syndrome Australian Research Fund 2001 – 2011 $750,000

Donated funds to support PKU research

• from the Italian Chamber of Commerce	– 2005	$145,000
	– 2006	$60,000
• from the Rotary Club of Pennant Hills		$300,000
• from the PKU Association of NSW		$360,000

Donated funds to support mitochondrial research

- from private donors $800,000

Examination of Postgraduate Theses:

1998:	MD	1	University of Adelaide
	PhD	2	University of Sydney
1999:	PhD	1	University of Melbourne
2003:	PhD	1	University of Adelaide
	PhD	1	University of Sydney
2006:	MPhil	1	University of Queensland
2008:	PhD	1	University of Western Australia
2009:	PhD	1	University of Sydney
2011:	PhD	1	University of Sydney

Grant Applications Reviewed:

1995:	NHMRC	4
	RCH Research Foundation (Melbourne)	1
1996:	NHMRC	3

	Channel 7 Research Foundation (Adelaide)	2
1997:	NHMRC	5
	Channel 7 Research Foundation (Adelaide)	2
1998:	NHMRC	3
1999:	NHMRC	3
2000:	NHMRC	4
	Channel 7 Research Foundation (Adelaide)	2
2001:	Channel 7 Research Foundation (Adelaide)	3
	APEX Foundation Trust for Autism	1
	NHMRC	4
	International Rett Syndrome Association	4
2002:	NHMRC	4
2003:	NHMRC	4
2004	NHMRC	4
	National Heart Foundation	1
2005	NHMRC	2
	Telethon Institute, Italy	1
2009	NHMRC	4
2010	NHMRC	2
2011	NHMRC	2

Manuscripts Reviewed:

Journal articles reviewed on a regular basis for the following journals:

Human Genetics, Journal of Inherited Metabolic Diseases, Medical Journal of Australia, American Journal of Medical Genetics, Journal of Paediatrics, Journal of Paediatrics and Child Health, European Journal of Paediatrics, Paediatrics International, European Journal of Human Genetics, Journal of Medical Genetics, Human Mutation, Human Molecular Genetics, Annals of Neurology, J of Epigenetics.

In addition, in October 2005 I was invited to become a Communicating Editor for the Journal of Inherited Metabolic Disease, which continues.

Mentoring and Development of Staff and Students:

Honours:	**Level**	**Year Awarded**
Jim Minchenko (University of Western Sydney) – B Sc	Honours 2.1	1997
Mai Yuk Chan (Macquarie University) – B. Sc	Honours 2.1	1998
Marija Mihelec (Macquarie University) – B. Med Sci	Honours 1	2003
Alexandra Bezler (Germany) – B Sci (Biotech)		2003

Vidya Vasudevan (Macquarie University) – B. Sci	Honours 2.1	2004
Abidali Mohamedali (Macquarie University) – B. Sci	Honours 1	2005
Kirsten Reuter (Germany) – B Sci (Biotech)		2004
Vince Repaci (Macquarie University) – B. Sci		2005
Simon Hardwick (Macquarie University) – B. Sci	Honours 1	2006
Carolin Hoffman (Germany) – B Sci (Biotech)		2006
Kelly Williams (Macquarie University) – B. Sci	Honours 1	2007
Barbara Garcia (Macquarie University) – B. Sci	did not complete	2007
Marie-Theres Weil (Germany) – B Sci (Biotech)		2008

Masters of Science:	**Degree**	**Year Awarded**
Karen Setterfield (Macquarie University) – full time	M Sc (Hons)	1991
KG (Ch'ng) Sim (University of Sydney) – part time (acting as Associate Supervisor)	M Sc	2002
Michael Malandris (University of Sydney) – part time (acting as Associate Supervisor)	M Dent Sc	2005
Wendy Lou (University of Sydney) – part time (acting as Associate Supervisor)	M Sc Dent	2006

Masters of Science in Medicine:

Rose White (Sydney University) – full time	M Sc Med	2007

Masters of Philosophy:

Katrina Slater (Sydney University) – full time	M Phil	commenced 2008
Ahmand Alodaib (Sydney University) – full time	M Phil	2011
Simranpreet Kaur (Sydney University) – full time	M Phil	2011

PhD:	**Year Commenced**	**Year Awarded**
Andrew Williams (University of Sydney) – part time	1993	1999
Carolyn Ellaway (University of Sydney) – full time	1996	2001
David Croaker (University of Sydney) – full time (acting as Associate Supervisor)	1996	2003
Jim Minchenko (University of Sydney) – full time	1998	2005
Linda Weaving (University of Sydney) – full time	1999	2004
Joanne Gibson (University of Sydney) – full time	2001	2006
Gregory Pelka (University of Sydney) – full time	2001	2005
Margaret Perry (Sydney University) – part time - commenced as Master of Medicine, upgraded to PhD in 2006	2002	2011
Sarah Williamson (University of Sydney) – full time	2003	2007
Vidya Vasudevan (University of Sydney) – full time	2005	2012
Abidali Mohamedali (University of Sydney) – full time	2006	2010

Roksana Armani (University of Sydney) – full time	2007	2011
Gladys Ho (University of Sydney) – full time	2008	2013
Minal Menezes (University of Sydney – full time	2010	
Ahamd Alodaib (University of Sydney) – full time	2011	
Naz Al Hafid (University of Sydney) – full time	2011	
Stephanie Fehr (University of Western Australia – full time (acting as Associate Supervisor)	2011	
Michael Nafisina (University of Sydney – full time	2013	

I am the principal supervisor for all of these students (except where indicated).

Work Experience Students:	**Year**
Elissa Levin (from USA)	1997
Melissa Diment	1998
Matthew Vitallone	2003
Lauren Curphy (Newcastle University)	2004
Peter Willis (Charles Sturt University)	2007

Summer Students:	**Year**
Abidali Mohamedali (Macquarie University)	2005
Adrian Sung (Sydney University)	2007
Cindy Jiang (Sydney University)	2007
Angela Zhou(Sydney University)	2007
David Ho (Sydney University)	2010
Carl Tong (Sydney University)	2011
Raine Wu (Sydney University)	2012
Weng Sam Chan (Sydney University)	2012

Overseas Undergraduate Students:

Bachelor of Science in Biotechnology students from Fachhochschule Mannheim - University of Applied Sciences – 6 month research periods

Alexandra Bezler	2003 – 2004
Kirsten Reuter	2004 – 2005
Carolin Hoffmann	2006 – 2007
Marie-Theres Weil	2008
Charlotte Lot-Snidjers	2012 – 2013

Overseas Postgraduate Students:

Elisa Bettella (PhD student from the University of Padova)	2009 – 2011

Clinical Genetics Fellows (ie post FRACP or equivalent Part I examinations):

	Year
Dr Robert Ogle (Metabolic Fellow)	1994 – 1995
Dr Siak Hong Teo (Honorary Metabolic Fellow)	1996 (from Singapore)
Dr Carolyn Ellaway (Metabolic Fellow)	1996 – 2000
Dr Rowani Rawi (Honorary Metabolic Fellow)	1997 (from Malaysia)
Dr Fanny Lam (Honorary Metabolic Fellow)	1998 (from Hong Kong)

Dr Rani Sachdev (molecular genetic project)	1998
Dr Tony Roscioli (molecular genetic project)	1998 – 1999
Dr Mary-Louise Freckmann (Metabolic Fellow)	1998 – 1999
Dr Joy Lee (Metabolic Fellow)	1999 – 2001 (from the Philippines)
Dr Grace David (Metabolic Fellow)	2001 – 2005 (from the Philippines)
Dr Catherine Sy (Metabolic and Neurogenetics Fellow)	2002 – 2004 (from the Philippines)
Dr Barbra Cavan (Metabolic Fellow)	2003 – 2004 (from the Philippines)
Dr John Mitchell (Metabolic Fellow)	2003 – 2004 (from Canada)
Dr Drago Bratkovic (Metabolic Fellow)	2004 – January 2006
Dr Kaustuv Bhattacharya (Metabolic Fellow)	Jan 2005 – Jan 2006 (from the UK)
Dr Mary Anne Chiong (Metabolic Fellow)	Jan 2005 – Dec 2007 (from the Philippines)
Dr Ee Shien Tan (Metabolic Fellow)	Jan 2006 – August 2006 (from Singapore)
Dr Sunita Bijarnia (Metabolic Fellow)	June 2006 – Jan 2008 (from India)
Dr Joanne Hughes (Metabolic Fellow)	Jan 2007 – Dec 2007 (from Ireland)
Dr Shanti Balasubramaniam (Metabolic Fellow	Aug 2008 – July 2009 (from Malaysia)

FRACP Paediatric Advanced Trainee Research Projects:	**Year**
Dr Davinder Singh-Grewal	2001
Dr Sondhya Ghedia	2002 – 2008
Dr Kate Neas	2003 – 2004
Dr Drago Bratkovic	2004 – 2005
Dr Pankaj Garg	2011

Medical Students:
Informal teaching at outpatient clinics on a regular basis.
Two lectures given each year to the first year Medical Students. (until 2004)
Active participant in the Medical Student Clinical Exams during their Paediatrics and Child Health rotation, four times each year.

Dissemination and popularisation of research/scholarship:

1996 - 1998:

2 radio interviews
1 TV appearances

1999:

27th April	AIR FM (100.7)	PKU – for PKU Awareness week
11th May	AIR FM (100.7)	Prenatal Diagnosis
July	Channel 9 News	Advances in Newborn Screening
30th September	Sydney Morning Herald	Discovery of the Rett syndrome Gene
7th October	Radio National (2NUR)	Discovery of the Rett syndrome Gene
7th October	2BL (702AM)	Discovery of the Rett syndrome Gene
8th October	2LM (Lismore)	Discovery of the Rett syndrome Gene

2000:

7th February	Medical Observer	The Human Genome Project

2001:

8th May	Prime TV (Albury)	Progress with Rett syndrome Research

2002:

22nd April	Daily Telegraph	Big Success Story of the Small Screen - PKU

2003:

28th January	2SM AM radio	Flight for Rett syndrome promotion
30th January	2RRR FM radio	Flight for Rett syndrome promotion
6th June	SBS Radio	Familial Mediterranean Fever

2004:

14th June	7.30 Report (ABC)	Families confront difficulties of terminal illness
26th July	Genetic Awareness Week	chaired the launch
5th November	Radio National (ABC)	New Rett syndrome gene discovery
22nd November	Australian newspaper	Study of twins offers genetic hope

2005:

3rd August	National 9 News	Discovery assists Rett syndrome sufferers
3rd August	NBN News	Australian researchers make two key discoveries about Rett syndrome
3rd August	Channel 9 Adelaide	Australian researchers make two key discoveries about Rett syndrome

2007:

14th February	Herald Sun	Gene hope for brain disease – reversibility of RTT phenotype in RTT mice
14th February	SBS Radio interview	Gene hope for brain disease
2nd December	Health Matters interview	Reversibility of RTT (interview with Dr John D'Arcy

2008:

2nd June	ABC 702 radio	Adam Spencer discusses Lorenzo Odone and

		ALD.
20th June	Channel 9 news	New research on Rett syndrome published in the European Journal of Neuroscience
25th June	SKY News	Discussion about Rett syndrome

2010:

6th September	ABC Four Corners	Interviewed as part of a program on gene Patents ("The Body Corporate").

2011:

10th January	ABC evening news	Interviewed about Rett syndrome
19th January	Sydney Morning Herald	Interviewed about Rett syndrome
10th June	Medical Observer	Interviewed about the cost of orphan drugs

2013:

28th February	Northern Star (Lismore)	Quoted for World Rare Diseases Day
1st March	Sydney Morning Herald	Quoted about rare diseases

Lectures Given to the Lay Community:

1. Research into Velo-Cardio-Facial syndrome. Presented at the Annual General Meeting of the VCFS Foundation of Australia, Westmead, August 1996.
2. *PKU and You.* Lecture as part of a workshop held for parents of children with PKU, RAHC, September 1996.
3. Genetic Testing and Research into Velo-cardio-facial syndrome. Presented at the Second VCFS parents seminar, Turramurra, NSW, November 1996.
4. *Treatment for Genetic Disorders.* Presented at the Genetics Awareness Week, Powerhouse Museum, June 1997.
5. *Latest in VCFS Research.* Preseneted at the VCFS Foundation of Australia Annual Meeting, New Children's Hospital, August 1997.
6. *Update on PKU.* Presented at the PKU Parents' Support Group Annual General Meeting, Sydney, July 1999.
7. *PKU Research From Around the World.* Presented at the Australian PKU Conference Annual, Sydney, October 2002.
8. *What's Hot in PKU Research?* Presented at the NSW PKU Association Annual General Meeting, February 2005.
9. *PKU Research at the Children's Hospital at Westmead.* Presented at a NSW PKU Association fundraiser, May 2005.
10. *Galactosaemia Education Morning.* Presented at the Children's Hospital at Westmead, August 2005.

11. *Update on Genetic Research into Rett Syndrome.* Presented at a parent workshop, sponsored by the Australian Paediatric Surveillance Unit, Children's Hospital at Westmead, November 2005.

12. *Update on PKU Research at the Children's Hospital at Westmead.* Presented at the NSW PKU Association Annual General Meeting, February 2006.

13. *Genetic Basis and Metabolic Manifestations of Trimethylaminuria.* Presented at the Inaugural TMAU Support Group of Australia, Sydney, Children's Hospital at Westmead, November 2006.

14. *PKU Research at the Children's Hospital at Westmead.* Presented at the Rotary Club of Pennant Hills Meeting, Pennant Hills, 2006, 2007, 2008, 2009, 2010, 2011.

Other Community Activities:

Senior Vice President - Sydney University Fencing Club 2002

Consultations:

2000:

Invitation to Italy by Sigma-Tau to consult on clinical trial of acetylcarnitine in patients with Rett Syndrome.

2001:

Invitation to participate in the SOS (SIDSAustralia Focus on Stillbirth) Forum. The primary aim of this meeting was to develop consensus criteria for the investigation of and research into the causes and pathogenesis of stillbirth. This was held at Sydney University in November 2001.

2008:

Chairman of a steering committee advising Nutricia on the development of a website for patients with genetic metabolic disorders. This involved chairing a number of meetings of about a dozen content specialists in association with an IT company and representatives from Nutricia. It is anticipated that the website will go live in 2009.

Publications:

Books:

Nil

Edited Books:

Nil

Chapters in Books:

1. **Christodoulou J**, McInnes RR. A Clinical Approach to Inborn Errors of Metabolism. In Rudolph AM, Kamei R, Eds. Rudolph's Fundamentals of Pediatrics. Appleton and Lange 1994 pp171-196.
2. **Christodoulou J**, McInnes RR. A Clinical Approach to Inborn Errors of Metabolism. In Rudolph AM, Kamei R, Eds. Rudolph's Fundamentals of Paediatrics. Appleton and Lange 1998 2nd Edition, pp 181-208.

3. **Christodoulou J**. Metabolic Failure. In Macnab A, Henning R, Macrae D, Eds. Care of the Critically Ill Child. Churchill Livingstone London, 1st Ed 1999, pp125 – 128.
4. **Christodoulou J**. Genetic Metabolic Disorders. In Macnab A, Henning R, Macrae D, Eds. Care of the Critically Ill Child. Churchill Livingstone London, 1st Ed 1999, 316 – 327.
5. **Christodoulou J**: Biochemical Genetic Emergencies. In Kilham H, Isaacs D, Eds. The New Children's Hospital Handbook. 1999 Ed, 98 – 104.
6. Barlow-Stewart K, **Christodoulou J**. It's all in our genes. In Oates RK, Child Health: A Manual for General Practice. Maclennan & Petty Sydney, 1st Ed. 2001, 37 – 46.
7. **Christodoulou J**. A Clinical Approach to Inborn Errors of Metabolism. In Rudolph AM, Kamei R, Overby KJ, Eds. Rudolph's Fundamentals of Paediatrics. McGraw-Hill 2002 3rd Edition, pp221 – 252.
8. **Christodoulou J**. Genetic Metabolic Disorders and Their Psychiatric Presentations. Nunn K, Dey C, Eds. The Clinician's Guide to Psychotropics. CAMHSNET 2003 1st Edition, pp332-346.
9. **Christodoulou J**, Wilcken B. Biochemical Genetic Emergencies. In Kilham H, Isaacs D, Eds. The Children's Hospital at Westmead Handbook McGraw-Hill 2004, pp 104 – 111.
10. **Christodoulou J**, Wilcken B. Biochemical Genetic Emergencies. In Kilham H, Isaacs D, Eds. The Children's Hospital at Westmead Handbook McGraw-Hill 2008, pp xxx – yyy.

Articles/Notes/Communications in Refereed Journals:

1. **Christodoulou J**, Clarke S, Buchanan N Compliance with immunisation programmes. Aust Paediatr J 1981: 17; 213-215.
2. **Christodoulou J**, Hall RK, Menahem S, Hopkins IJ, Rogers JG A syndrome of epilepsy, dementia, and amelogenesis imperfecta: genetic and clinical features. J Med Genet 1988 25; 827-830.
3. **Christodoulou J**, Dewan PA, Tan HL, Rogers JG Priapism: A rare complication of Fabry disease. Pediatr Surg Int 1988: 4; 69-70.
4. **Christodoulou J**, McDougall PN, Sheffield LJ Choanal atresia as a feature of Ectrodactyly - Ectodermal dysplasia - Clefting (EEC) syndrome. J Med Genet 1989: 26; 586-589.
5. Hughes JL, Poulos A, Robertson E, Chow CW, Sheffield LJ, **Christodoulou J**, Carter RF Pathology of hepatic peroxisomes and mitochondria in patients with peroxisomal disorders. cc 1990: 416; 255-264.
6. Pitt JJ, Brown GK, Clift, V, **Christodoulou J** Case report: atypical pyroglutamic aciduria-possible role of paracetamol. J Inher Metab Dis 1990: 13(5); 755-756.
7. Tatuch Y, **Christodoulou J**, Feigenbaum A, Clarke JTR, Wherret J, Smith C, Rudd N, Petrova-Benedict R, Robinson BH Heteroplasmic mtDNA mutation (T □G) at 89
Leigh's disease when the percentage of abnormal mtDNA is high. Am J Hum Genet 1992: 50 (4); 852-858.

8. Jay V, **Christodoulou J**, Mercer-Connolly A, McInnes RR "Reducing body"-like inclusions in skeletal muscle in childhood-onset acid maltase deficiency. Acta Neuropath 1992: 85; 111-115.

9. Chow CW, Poulos A, Fellenberg AJ, **Christodoulou J**, Danks DM Autopsy findings in two siblings with infantile Refsum disease. Acta Neuropath 1992: 83; 190-195.

10. **Christodoulou J**, Clarke JTR, Rupar CA, Gordon BA, Kelly DP Retrospective diagnosis of medium chain acyl-CoA dehydrogenase (MCAD) deficiency. J Paediatr Child Health 1993 29(3); 237-238.

11. **Christodoulou J**, Petrova-Benedict R, Robinson B, Jay V, Clarke J An unusual patient with the neonatal Marfan syndrome and mitochondrial complex I deficiency. Eur J Paediatr 1993: 152; 428-432.

12. **Christodoulou J**, Qureshi IA, McInnes RR, Clarke JTR Ornithine transcarbamylase deficiency presenting with stroke-like episodes. J Pediatr 1993: 122(3); 423-425.

13. **Christodoulou J**, Haysaka K, Kure S, Clarke JTR Atypical Nonketotic Hyperglycinemia Confirmed by Assay of the Glycine Cleavage System in Lymphoblasts. J Paediatr 1993: 123(1); 100-102.

14. **Christodoulou J**, McInnes RR, Jay V, Wilson G, Becker LE, Lehotay DC, Platt B-A, Bridge PJ, Robinson BH, Clarke JTR Barth syndrome: clinical observations and genetic linkage studies. Am J Med Genet 1994: 50; 255-264.

15. Tein I, **Christodoulou J**, Donner E, McInnes RR Carnitine palmitoyltransferase II deficiency: A new cause for recurrent pancreatitis. J Pediatr 1994: 124(6); 938-940.

16. Ozand PT, Nyhan WL, Al Aqeel A, **Christodoulou J** Malonic aciduria. Brain Dev 1994: 16; 7-11.

17. **Christodoulou J**, Hoare J, Hammond J, Wilcken B Neonatal onset of medium chain acyl-CoA dehydrogenase (MCAD) deficiency with confusing biochemical features. J Pediatr 1995: 126(1); 65-68.

18. Smooker PM, **Christodoulou J**, McInnes RR, Cotton RGH A mutation causing DHPR deficiency results in a frameshift and a secondary splicing defect. J Med Genet 1995: 32 (3); 220-223.

19. Poulos A, **Christodoulou J**, Chow CW, Goldblatt J, Orii T, Suzuki Y, Shimozawa N Peroxisomal assembly defects: clinical, pathological, and biochemical findings in two patients belonging to a newly identified complementation group. J Paediatr 1995: 127(4); 596-599.

20. Tümer Z, Horn N, Tønnesen T, **Christodoulou J**, Clarke JTR, Sarkar B Early copper-histidine treatment for Menkes disease. Nature Genetics 1996: 12(1); 11-13.

21. Rahman S, Dahl H-HM, Blok RB, Danks DM, Kirby DM, Chow CW, **Christodoulou J**, Thorburn DR Leigh syndrome: clinical features and biochemical and DNA abnormalities. Ann Neurol 1996: 39(3); 343-351.

22. Worthington S, **Christodoulou J**, Wilcken B, Peat B Pregnancy and Argininosuccinic aciduria. J Inher Metab Dis 1996: 19 (5); 621-623.

23. **Christodoulou J**, Teo SH, Hammond J, Sim KG, Hsu BYL, Stanley CA, Watson B, Lau KC, and Wilcken B First Prenatal diagnosis of the carnitine transporter defect. Am J Med Genet 1996: 66; 21-24.

24. Ogle RF, **Christodoulou J**, Fagan E, Blok RB, Seller KL, Dahl H-H, Thorburn DR Mitochondrial myopathy with $tRNA^{Leu(UUR)}$ mutation and complex I deficiency responsive to riboflavin. J Paediatr 1997: 130(1); 138-145.

25. Williams AJ, Coakley J, **Christodoulou J** Automated quantitation of total protein in cultured skin fibroblasts. Clin Chim Acta 1997: 259; 129-136.

26. Freckman M-L, Thorburn DR, Dennett X, Hammond J, Kamath R, **Christodoulou J** Electron transport chain defect presenting as hypoglycaemia. J Paediatr 1997: 130(3); 431-436.

27. Walker DC, **Christodoulou J**, Craig HJ, Simard LR, Ploder L, Howell PL, McInnes RR Intragenic complementation at the human argininosuccinate lyase locus: identification of the major complementing alleles. J Biol Chem 1997: 272 (10); 6777-6783.

28. Ellaway C, Elliott EJ, **Christodoulou J** Myoclonus epilepsies: new genetic advances. J Paediatr Child Health 1997: 33 (2); 91-95.

29. Ellaway C, North K, Arbuckle S, **Christodoulou J** Complex I deficiency in association with structural abnormalities of the diaphragm and brain. J Inher Metab Dis 1998: 21 (1); 72-73.

30. Ellaway C, **Christodoulou J**, Kamath R, Carpenter K, Wilcken B Protein losing enteropathy in a baby with cobalamin C deficiency. J Inher Metab Dis 1998: 21 (1); 17-22.

31. **Christodoulou J**, Danks DM, Sarkar B, Clarke JTR Natural history of classical Menkes disease treated from birth: evolution to an occipital horn syndrome phenotype with copper-histidinate therapy. Am J Med Genet 1998: 76; 154-164.

32. Howell PL, Turner MA, **Christodoulou J**, Walker DC, Craig HJ, Simard L, Ploder L, McInnes RR Intragenic complementation at the argininosuccinate lyase locus. J Inher Metab Dis 1998: 21 (suppl 1); 72-85.

33. Ellaway C, Bucholtz T, Leonard H, Smith A, **Christodoulou J** Rett syndrome: phenotypic overlap with Angelman syndrome and methylation studies. J Child Neurol 1998: 13; 448-452.

34. Williams AJ, Coakley J, **Christodoulou** Automated analysis of mitochondrial enzymes in cultured skin fibroblasts. Analyt Biochem 1998: 259; 176-180.

35. Glasson EJ, Thomson MR, Fyfe S, Leonard S, Bower C, Rousham E, **Christodoulou J**, Ellaway C, Leonard H Diagnosis of Rett Syndrome: can a simple radiograph help? Dev Med Child Neurol 1998: 40; 737-742.

36. White SL, Thorburn DR, **Christodoulou J**, Dahl H-HM A novel mtDNA variant that may give a false positive diagnosis for the T8993C mutation. Molec Diagnosis 1998: 3; 113 – 117.

37. Williams GD, **Christodoulou J**, Stack J, Tobias V, Wert S, Murrell M, Nogee L Surfactant protein B deficiency: clinical, histological and molecular evaluation. J Paediatr Child Health 1999: 35; 214-220.

38. Mowat D, Thorburn DR, Kirby DM, Kamath KR, Arbuckle S, **Christodoulou J** Complex III deficiency with pruritus: a novel vitamin responsive clinical feature. J Paediatr 1999: 134 (3); 352-354.

39. Leonard H, Thomson M, Glasson E, Fyfe S, Leonard S, Ellaway C, **Christodoulou J**, Bower C Metacarpophalangeal pattern profile and bone age in Rett syndrome: further radiological clues to the diagnosis. Am J Med Genet 1999: 83; 88-95.

40. Williams AJ, Coakley J, **Christodoulou J** Flow cytometric evaluation of defects of the mitochondrial respiratory chain. J Child Neurol 1999: 14 (8); 518 – 523.

41. Leonard H, Thomson MR, Glasson EJ, Fyfe S, Leonard S, Bower C, **Christodoulou J**, Ellaway C A population based approach to the investigation of osteopenia in Rett syndrome. Dev Med Child Neurol 1999: 41 (5); 323 – 328 .

42. Ellaway CJ, Leonard H, Williams K, Higgins G, Wilcken B, **Christodoulou J** Randomised controlled trial of L-carnitine in Rett syndrome. J Child Neurol 1999: 14; 162-167.

43. Ellaway C, Sholler G, Leonard H, **Christodoulou J** Prolonged QT interval in Rett syndrome. Arch Dis Child 1999: 80; 470-472.

44. Williams AJ, Murrell M, Brammah S, **Christodoulou J** A novel system for assigning the mode of inheritance in mitochondrial disorders using cybrids and rhodamine 6G. Human Molec Genet 1999: 8 (9); 1691 – 1697.

45. Fitzpatrick DR, Hill A, Tolmie JL, Thorburn DR, **Christodoulou J** The molecular basis of malonyl-CoA decarboxylase deficiency. Am J Human Genet 1999: 65; 318 – 326.

46. McQuade L, **Christodoulou J**, Budarf M, Sachdev R, Wilson M, Emanuel B, Colley A A further case of a 22q11.2 deletion with no overlap to the minimal DiGeorge syndrome critical region (MDGCR): the involvement of the *TBX1* and *COMT* genes in the deletion and the clinical phenotype. Amer J Med Genet 1999: 86 (1); 27 – 33.

47. Ellaway C, **Christodoulou J** Rett syndrome: clinical update and review of recent genetic advances. J Paediatr Child Health 1999: 35 (5); 419 – 426.

48. Ellaway C, McQuade L, **Christodoulou J** Novel mitochondrial 16S rRNA polymorphism in a girl with Rett syndrome. J Inher Metab Dis 1999: 22; 946 – 947.

49. Kilpatrick N, Awang H, Wilcken B, **Christodoulou J,** The implications of phenylketonuria on oral health. Pediatr Dentistry 1999: 21 (7); 433 – 437.

50. Stone DL, Carey WF, **Christodoulou J**, Sillence D, Nelson P, Lau EK, Taybi N, Sidranksy E Type 2 Gaucher disease – the collodion baby phenotype revisited. Arch Dis Child Fetal Neonatal 2000: 82 (2); F163 – F166.

51. Leonard H, Fyfe S, Dye D, Hockey A, **Christodoulou J** Family data in Rett syndrome: association with other genetic disorders. J Paediatr Child Health 2000: 36; 336 – 339.

52. Carpenter KH, Wilcken B, **Christodoulou J**, Thorburn DR Holocarboxylase synthase deficiency: urinary metabolites masked by gross ketosis. J Inher Metab Dis 2000 23; 845.

53. Grattan-Smith, Healey S, Grigg JR, **Christodoulou J**, Spinocerebellar ataxia type 7 (SCA7): a distinctive form of autosomal dominant cerebellar ataxia with retinopathy and marked genetic anticipation. J Paediatr Child Health 2001: 37; 81 – 84.

54. Kerr A, Nomura Y, Armstrong A, Anvret M, Belichenko PV, Budden S, Cass H, **Christodoulou J**, Clarke A, Ellaway C, d'Esposito M, Francke U, Hulten M, Julu P, Leonard H, Naidu S, Schanen C, Webb T, Witt-Engerstrom I, Yamashita Y, Segawa M. Guidelines for the reporting of clinical features in cases with *MECP2* mutations. Brain and Development 2001: 23; 208 – 211.

55. Ellaway CJ, Badawi N, Raffaele L, **Christodoulou J**, Leonard H. a case of multiple congenital anomalies in association with Rett syndrome confirmed by MECP2 mutation screening. Clin Dysmorph 2001: 10; 185 – 188.

56. Ellaway C, Peat J, Leonard H, **Christodoulou J**, Sleep dysfunction in Rett syndrome. Brain and Development 2001: 23 (Suppl 1); S101 – S103.

57. Ellaway C, Peat J, Williams K, Leonard H, **Christodoulou J**, Medium-term open label trial of L-carnitine in Rett syndrome. Brain and Development 2001: 23 (Suppl 1); S85 – S89.

58. Setterfield K, Williams A, Donald J, Thorburn DR, Kirby DM, Trounce I, **Christodoulou J.** Flow cytometry in the study of mitochondrial respiratory chain disorders. Mitochondrion 2002: 1; 437 – 445.

59. Milledge J, Shaw PJ, Mansour A, Williamson S, Bennetts B, Roscioli A, Curtin J, **Christodoulou J**. Allogenic bone marrow transplantation: cure for Familial Mediterranean Fever. Blood 2002: 100; 774 – 777.

60. Ellaway CJ, Wilcken W, **Christodoulou J,** A clinical approach to inborn errors of metabolism presenting in the newborn period. J Pediatr Child Health 2002: 38 (5); 511 – 517.

61. Sim KG, Carpenter K, Hammond J, **Christodoulou J**, Wilcken B. Acylcarnitine profiles in fibroblasts from patients with respiratory chain defects can resemble those with mitochondrial fatty acid -oxidation disorders. Metabolism 2002: 51 (3); 366 – 371.

62. Sim KG, Carpenter K, Hammond J, **Christodoulou J**, Wilcken B. Quantitative fibroblast acylcarnitine profiles in mitochondrial fatty acid -oxidation defects: phenotype/metabolite correlations. Molec Genet Metab 2002: 76; 327 – 334.

63. **Christodoulou J**. Understanding Rett syndrome. Trends in Molec Med 2002: 8; 406 – 407.

64. Colvin L, Fyfe S, Leonard S, Schiavello T, Ellaway C, de Klerk N, **Christodoulou J**, Msall M, Leonard H. Describing the phenotype in Rett syndrome using a population database. Arch Dis Child 2003: 88 (1); 38 – 43.

65. Weaving LS, Williamson SL, Bennetts B, Davis M, Ellaway CJ, Leonard H, Thong MK, Delatycki M, Thompson EM, Laing N, **Christodoulou J,** Effects of *MECP2* mutation type, location and X-inactivation in modulating Rett syndrome phenotype. Amer J Medical Genet 2003: 118A; 103 – 114.

66. Minchenko J, Williams AJ, **Christodoulou J**. Adaptation of a mitochondrial complex III assay for automation: examination of reproducibility and precision. Clin Chem 2003: 49 (2); 330 – 332.

67. Leonard H, Colvin L, **Christodoulou J**, Schiavello T, Weaving L, Williamson S, Davis M, Ravine D, Fyfe S, de Klerk N, Matsuishi T, Kondo I, Clarke A, Hackwell S, Yamashita Y. Patients with the R133C mutation: is their phenotype different from Rett syndrome patients with other mutations? J Med Genet.2003: 40; e52.

68. **Christodoulou J**, Grimm A, Maher T, Bennetts B. RettBASE: The IRSA *MECP2* Variation Database. A new mutation database in evolution. Human Mutation 2003: 21 (5); 466 – 472.

69. Gill H, Cheadle JP, Maynard J, Fleming N, Whatley S, Cranston T, Thompson EM, Leonard H, Davis M, **Christodoulou J**, Skjeldal O, Hanefeld F, Kerr A, Tandy A, Ravine D, Clarke A. Mutation analysis in the *MECP2* gene in genetic counselling for Rett syndrome J Med Genet 2003: 40; 380 – 384.

70. **Christodoulou J**, Weaving LS. *MECP2* and Beyond: Phenotype – Genotype Correlations in Rett Syndrome. J Child Neurol 2003: 18 (10); 669 – 674.

71. Fyfe S, de Klerk N, Cream A, **Christodoulou J**, Leonard H. *MECP*2 and InterRett _ International IRSA databases for Rett Syndrome J Child Neurol 2003: 18 (10); 709 – 713.

72. **Christodoulou J**. The Human Genome Project: opportunities, challenges and consequences for population screening. Southeast Asian J Tropical Med Pub Health 2003: 34 (suppl 3); 189 – 107.

73. **Christodoulou J**. Clinical evaluation and emergency management of inborn errors of metabolism presenting in the newborn. Southeast Asian J Tropical Med Pub Health 2003: 34 (suppl 3); 234 - 238.

74. Shaw CJ, Stankiewicz P, **Christodoulou J**, Smith E, Jones K, Lupinski JR. Trisomy 17p10-p12 resulting from a duplication including the centromere. Am J Med Genet 2004: 124A (2); 173 – 178.

75. Rae C, Joy P, Harasty J, Kemp A, Kuan S, **Christodoulou J**, Cowell CT, Coltheart M. Enlarged temporal lobes in Turner syndrome. An X-chromosome effect? Cerebral Cortex 2004: 14; 156 – 164.

76. Leonard H, Weaving L, Eastaugh P, Smith L, Delatycki M, Witt-Engerström I, **Christodoulou J**. Trisomy 21 and Rett syndrome: a double burden. J Pediatr Child Health 2004: 40; 406 – 409.

77. Colvin L, Leonard H, de Klerk N, **Christodoulou J**, Davis M. Refining the phenotype of common mutations in Rett syndrome. J Med Genet 2004: 41 (1); 25 - 30.

78. **Christodoulou J**, Wilcken B. Perimortem laboratory investigation of genetic metabolic disorders. Seminars Neonatol 2004: 9; 275 – 280.

79. Tumer Z, Horn N, Tonnesen T, **Christodoulou J**, Clarke JT, Sarkar B. Gene symbol: ATP7A. Disease: Menkes disease. Hum Genet. 2004: 114(6); 606.

80. Weaving LS, **Christodoulou J**, Williamson SL, Friend KL, McKenzie OL, Archer H, Evans J, Clarke A, Pelka GJ, Tam PP, Watson C, Lahooti H, Ellaway CJ, Bennetts B, Leonard H, Gecz J. Mutations of CDKL5 Cause a Severe Neurodevelopmental Disorder with Infantile Spasms and Mental Retardation. Am J Hum Genet. 2004: 75(6); 1079 – 1093.

81. Gibson J, Williamson S, Arbuckle S, **Christodoulou J**. X chromosome inactivation patterns of the brain in Rett syndrome: implications for the disease phenotype. Brain Dev 2005: 27; 266 – 270.

82. Mancuso M, Ferraris S, Pancrudo J, Feigenbaum A, Raiman J, **Christodoulou J**, Thorburn DR, DiMauro S. New *dGK* gene mutations in the hepatocerebral form of mitochondrial DNA depletion syndrome. Arch Neurol 2005: 62; 745 – 747.

83. Biggin A, Henke R, Bennetts B, Thorburn DR, **Christodoulou J**. Mutation screening of the mitochondrial genome using denaturing high-performance liquid chromatography. Molec Genet Metab 2005: 84 (1); 61 – 74.

84. Weaving LS, Ellaway CJ, Gécz J, **Christodoulou J**. Rett syndrome: clinical review and genetic update. J Med Genet 2005: 42(1); 1 – 7. (Named one of the top 10 articles published in the Journal of Medical Genetics in 2005)

85. Moore H, Leonard L, de Klerk N, Robertson I, Fyfe S, **Christodoulou J**, Weaving LS, Davis M, Leonard S, Colvin L. Health service utilization in Rett syndrome. J Child Neurol 2005: 19; 42 – 50.

86. Neas K, Bennetts B, Carpenter K, White R, Kirk EP, Wilson M, Kelley R, Baric I, **Christodoulou J**. *OPA3* mutation screening in patients with unexplained 3-methylglutaconic aciduria. J Inher Metab Dis 2005: 28; 525 – 532.

87. Pelka GJ, Watson CM, **Christodoulou J**, Tam, P. Distinct expression profiles of *Mecp2* transcripts with different lengths of 3'UTR in the brain and visceral organs during mouse development. Genomics 2005: 85; 441 – 452. (artwork from the manuscript formed the front cover of this issue)

88. Watson CM, Pelka G, Radziewic T, Shahbazian, MD, **Christodoulou J**, Williamson SL, Tam PPL. Reduced proportion of Purkinje cells expressing paternally derived mutant Mecp2308 allele in female mouse cerebellum is not due to a skewed primary pattern of X-chromosome inactivation. Hum Molec Genet 2005: 14 (13); 1851 – 1861.

89. **Christodoulou J**, Williamson SL, Carolyn J. Ellaway CJ. Rett syndrome. Enclyclopedia of Medical Genomics and Protoemics 2005 (online publication) DOI 10.1081/E-EDGP-120025645.

90. Jian L, Archer HL, Ravine D, Kerr A, de Klerk N, **Christodoulou J**, Bailey ME, Laurvick C, Leonard H. p.R270X MECP2 mutation and mortality in Rett syndrome. Eur J Hum Genet. Eur J Hum Genet. 2005: 13(11); 1235 – 1238.

91. Evans JC, Archer HL, Colley JP, Ravn K, Nielsen JB, Kerr A, Williams E, **Christodoulou J**, Gecz J, Jardine PE, Wright MJ, Pilz DT, Lazarou L, Cooper DN, Sampson JR, Butler R, Whatley SD, Clarke AJ. Early onset seizures and Rett-like features associated with mutations in CDKL5. Eur J Hum Genet. 2005: 13(10); 1113 – 1120.

92. Leonard H, Moore H, Carey M, Fyfe S, Hall S, Robertson L, Wu XR, Bao X, Pan H, **Christodoulou J**, Williamson S, Klerk N. Genotype and early development in Rett syndrome: The value of international data. Brain Dev. 2005: 27 Suppl 1; S59 – S68.

93. Barclay AR, Sholler G, **Christodoulou J**, Shun A, Arbukle S, Dorney S, Stormon MO. Pulmonary hypertension – a new manifestation of mitochondrial disease. J Inher Metab Dis 2005: 28; 1081 – 1089.

94. Tredano M, Cooper DN, Stuhrmann M, **Christodoulou J**, Chuzhanova NA, Roudot-Thoraval F, Boëlle P-Y, Elion J, Jeanpierre M, Feingold J, Coudere R, Bahuau M. Origin of the prevalent *SFTB* indel g.1549C>GAA (121ins2) mutation causing surfactant protein B (SP-B) deficiency. Amer J Med 2006: 140A; 62 – 69.

95. Pelka GJ, Watson CM, Radziewic T, Hayward M, Lahooti H, **Christodoulou J**, Tam PPL. *Mecp2* deficiency is associated with learning and cognitive deficits and altered gene activity in the hippocampal region of mice. Brain 2006: 129(Pt 4); 887 – 898.

96. Ager S, Fyfe S, **Christodoulou** J, Jacoby P, Schmitt L, Leonard L. Predictors of Scoliosis in Rett Syndrome. J Child Neurol. 2006: 21(9); 809 – 813.

97. Saxena A, de Lagarde D, Leonard H, Williamson S, Vasudevan V, **Christodoulou J**, Thompson E, Macleod P, Ravine D. Lost in translation: Translational interference from a recurrent mutation in exon 1 of MECP2. J Med Genet. 2006: 43 (6); 470 – 477.

98. Laurvick CL, de Klerk N, Bower C, **Christodoulou J**, Ravine D, Ellaway C, Williamson S, Leonard L. Rett Syndrome in Australia: A Review of the Epidemiology. J Pediatr 2006 Mar;148(3):347 – 352.

99. Archer HL, Evans JC, Millar DS, Thompson PW, Kerr AM, Leonard H, **Christodoulou J**, Ravine D, Lazarou L, Grove, L, Verity C, Whatley SD, Pilz DT, Sampson JR, Clarke AJ. *NTNG1* mutations are a rare cause of Rett syndrome. Amer J Med Genet 2006: 140(7); 691 – 694.

100. Mitchell JJ, Wilcken B, Alexander I, Ellaway C, O'Grady H, Wiley V, Earl J, **Christodoulou J**. Tetrahydrobiopterin responsive phenylketonuria: The New South Wales experience. Mol Genet Metab. 2006: 86 (Suppl 1); S81 – S85.

101. Williamson SL, **Christodoulou J**. Rett Syndrome: New Clinical and Molecular Insights. Eur J Hum Genet 2006: 14(8); 896 – 903.

102. **Christodoulou J**, Craig HJ, Walker DC, L. Weaving, Pearson C, McInnes RR. Deletion "hotspot" in the argininosuccinate lyase gene: Association with topoisomerase II and DNA polymerase [illegible] mutation sites Hum Mutat. 2006: 27(11); 1071.

103. Kure S, Junko Kanno J, Sato K, Kojima K, Ichinohe A, Kamada F, Kato K, Aoki Y, Dinopoulos A, Gail C, TJ, **Christodoulou J**, Hakansson H, Bzduch V, Kalmanchey R, Fekete

G, Trojovsky A, Boneh A, Toyama J, Tonooka S, Takayanagi T, Kubota M, Takayanagi M,and Matsubara Y. A comprehensive mutation analysis of *GLDC*, *AMT*, and *GCSH* in nonketotic hyperglycinemia. Hum Mut 2006: 27 (4); 343 – 352.

104. Chiong M-A, Marinaki A, Duley J, Bennetts B, Ouvrier RA, **Christodoulou J**. Lesch-Nyhan disease presenting as "cerebral palsy" after general anaesthesia: a case report. J Inherit Metab Dis. 2006: 29(4); 594.

105. Kankirawatana P, Leonard H, Ellaway C, Scurlock J, Mansour A, Makris CM, Dure LS, Friez M, Lane J, Kiraly-Borri C, Fabian V, Davis M, Jackson J, **Christodoulou J**, Kaufmann W, Ravine Percy AK. Early Progressive Encephalopathy in Males and *MECP2* Mutations. Neurology 2006: 67(1); 164 – 166.

106. Jian L, Nagarajan L, de Klerk N, Ravine D, Bower C, Anderson A, Williamson S, **Christodoulou J**, Leonard H. Predictors of seizure onset in Rett syndrome. J Pediatr. 2006: 149(4); 542 – 547.

107. Archer HL, Evans J, Leonard H, Colvin L, Ravine D, **Christodoulou J**, Williamson S, Charman T, Bailey ME, Sampson J, de Klerk N, Clarke AJ. Correlation between clinical severity in Rett syndrome patients with a p.R168X or p.T158M MECP2 mutation and the direction and degree of skewing of X chromosome inactivation. J Med Genet. 2007: 44 (2); 148 – 152.

108. Chan PA, Duraisamy S, Murphy JA, Miller PJ, McBride C, Bond JP, Raevaara T, Ollila S, Nyström M, Grimm A, **Christodoulou J**, Oetting WS, Greenblatt MS. Predicting benign versus pathogenic genetic variants: validation of four methods of comparative sequence analysis in human disease genes *CDKN2A*, *MLH1*, *MECP2*, and Tyrosinase. Hum Mut 2007: 28 (7); 683 – 693.

109. Schindeler S, Ghosh–Jerath S, Thompson S, Rocca A, Joy P, Kemp A, Rae C, Green K, Wilcken B, **Christodoulou J**. The Effects of Large Neutral Amino Acid Supplements in PKU: an MRS and Neuropsychological Study. Molecular Genetics 2007: 91(1); 48 – 54.

110. Jian L, Nagarajan L, de Klerk N, Ravine D, **Christodoulou J**, Leonard H. Seizures in Rett syndrome: an overview from a one-year calendar study. Eur J Pediatr Neurol 2007: 11(5); 310-317.

111. Chiong MA, Carpenter K, **Christodoulou J**. Low citrulline may not be diagnostic of ornithine transcarbamoylase deficiency: a case report. J Inher Metab Dis 2007: (30); 405.

112. Chiong MA, Sim KG, Carpenter K, Rhead W, Ho G, Olsen RKJ, **Christodoulou J**. Transient multiple acyl-CoA dehydrogenation deficiency in a newborn female caused by maternal riboflavin deficiency. Molec Genet Metab 2007: 92(1-2); 109-114.

113. Hardwick SA, Reuter K, Williamson SL, Vasudevan V, Donald J, Slater K, Bennetts B, Bebbington A, Leonard H, Williams SR, Smith RL, Cloosterman D, **Christodoulou J**. Delineation of large deletions of the *MECP2* gene in Rett syndrome patients, including a familial case with a male proband. Eur J Hum Genet 2007: 15(12); 1218-1229.

114. de Brouwer APM, Williams KL, Duley J, van Kuilenburg ABP, Nabuurs S, Egmont-Petersen M, Lugtenberg D, Zoetekouw L, Banning MJG, Roeffen M, Hamel BCJ, Weaving L,

Ouvrier RA, Donald JA, Wevers RA, **Christodoulou J**, van Bokhoven H. Arts syndrome is caused by loss-of-function mutations in *PRPS1*. Am J Hum Genet 2007: 81(3); 507-518.

115. Singh-Grewal D, Chaitow J, Aksentijevich I, **Christodoulou J** . Coexistent *MEFV* and *CIAS1* mutations manifesting as familial Mediterranean fever plus deafness. Ann Rheum Dis 2007: 66; 1541.

116. Cotton RGH, Auerbach AD, Brown AF, Carrera P, **Christodoulou J**, Claustres M, Compton J, Cox DW, De Baere E, den Dunnen JT, Greenblatt M, Fujiwara M, Hilbert P, Jani A, Lehvaslaiho H, Nebert D, Verma I, Vihinen M: Members of the Human Genome Variation Society and the Human Variome Project Diagnostic Laboratory Working Group A Structured Simple Form for Ordering Genetic Tests is Needed to Ensure Coupling Clinical Detail (Phenotype) and Mutation (Genotype) to Ensure Utility in Publication and Databases. Hum Mutat. 2007: 28(10); 931-932.

117. Alexander IE, Cunningham SC, Logan GJ, **Christodoulou J.** Potential of AAV vectors in the treatment of metabolic disease. Gene Therapy 2008: 15; 831-839. IF 4.782

118. Kondo M, Gray LJ, Pelka GJ, **Christodoulou J**, Tam PPL, Hannan AJ. Environmental enrichment ameliorates a motor coordination deficit in a mouse model of Rett syndrome: Mecp2 gene dosage effects and BDNF expression. Eur J Neurosci 2008: 27; :3342-3350

119. Bijarnia S, Wiley V, Carpenter K, **Christodoulou J**, Ellaway CJ, Wilcken B. Glutaric aciduria type I: outcome following detection by newborn screening. J Inherit Metab Dis 2008: 31(4); 503-507.

120. Sugiana C, Pagliarini DJ, McKenzie M, Kirby DM, Salemi R, Abu-Amero KK, Dahl HH, Hutchison WM, Vascotto KA, Smith SM, Newbold RF, **Christodoulou J**, Calvo S, Mootha VK, Ryan MT, Thorburn DR. Mutation of C20orf7 disrupts complex I assembly and causes lethal neonatal mitochondrial disease. Am J Hum Genet. 2008: 83(4); 468-478.

121. Singh J, Saxena A, **Christodoulou J**, Ravine D. MECP2 genomic structure and function: insights from ENCODE. Nucleic Acids Res. 2008: 36(19); 6035-6047.

122. Ho G, Walter J, **Christodoulou J**. Costeff optic atrophy syndrome: new clinical case and novel molecular findings. J Inher Metab Dis. 2008: 31; 774 (Full text: http://dx.doi.org/10.1007/s10545-008-0981-z).

123. Ben Zeev B, Bebbington A, Ho G, Leonard H, de Klerk N, Gak E, Vecksler M, **Christodoulou J**. The common *BDNF* polymorphism may be a modifier of disease severity in Rett syndrome. Neurology. 2009: 72; 1242-1247.

124. Shanti B, Sillink M, Bhattarchaya K, Howard N, Carpenter K, Fietz M, Clayton P, **Christodoulou J**. Congenital Disorder of Glycosylation type 1a: Hyperinsulinaemic hypoglycemia as a leading symptom in two patients with phosphomannomutase deficiency. J Inher Metab Dis. 2009: 32; 450 (full text: http://dx.doi.org/10.1007/s10545-009-1180-2).

125. Wilcken B, Haas M, Joy P, Wiley V, Bowling F, Carman P, Carpenter K, **Christodoulou J**, Cowley D, Ellaway C, Fletcher J, Kirk E, Knight G, Lewis B, McGill J, Peters H, Pitt J, Ranieri E, Yaplito-Lee J, Boneh A. Newborn screening by tandem mass spectrometry: a cohort study comparing outcome in screened and clinically diagnosed patients at six years of age. Pediatr. 2009: 124 (2); e241-248.

126. Hamvas A, Nogee LM, Wegner DJ, DePass K, **Christodoulou J**, Bennetts B, McQuade LR, Gray PH, Deterding RR, Carroll TR, Kammesheidt A, Kasch LM, Kulkarni S, Cole FS. Inherited surfactant deficiency due to uniparental disomy of rare mutations in the surfactant protein-B and ATP binding cassette, subfamily A, member 3 genes. J Pediatr. 2009: 155; 854-859.

127. Bijarnia S, Shaw P, Vimpani A, Smith R, Pacey V, O'Grady H, **Christodoulou J**, Sillence DO. Combined enzyme replacement and haematopoietic stem cell transplantation in mucopolysaccharidosis type I (Hurler syndrome) and long term follow up. J Paediatr Child Health. 2009: 45; 469-472.

128. Hynes K, Tarpey P, Dibbens LM, Bayly MA, Berkovic SF, Smith R, Al Raisi Z, Turner SJ, Brown NJ, D Desai T, Haan E, Hackett A, Turner G, **Christodoulou J**, Leonard H, Gill D, Stratton M, Gecz J, Scheffer IE. Epilepsy and mental retardation limited to females with *PCDH19* mutations can present *de novo* or in single generation families. J Med Genet. 2010: 47 (3); 211-216.

129. Relf BL, Larkin EK, de Torres C, Baur LA, Christodoulou J, Waters KA. Genomewide linkage of Obstructive Sleep Apnea and High Density Lipoprotein Cholesterol in a Filipino Family. J Sleep Res. 2010: 19(2); 349-357.

130. Bebbington A, Percy A, Christodoulou J, Ravine D, Ho G, Jacoby P, Anderson A, Pineda M, Ben Zeev B, Bahi-Buisson N, Smeets E, Leonard H. Updating the profile of C-terminal *MECP2* deletions in Rett syndrome. J Med Genet. 2010: 47; 242-248.

131. Harikrishnan KN, Bayles R, Ciccotosto J, Maxwell S, Cappai R, Pelka G, Tam P, Christodoulou J, El-Osta A. Alleviating transcriptional inhibition of the norepinephrine Slc6a2 transporter gene in depolarised neurons. Journal Neurosci. 2010: 30 (4); 1495-1501.

132. Gibson JH, Slobedman B, Harikrishnan KN, Williamson S, Minchenko D, El-Osta A, Stern JL, **Christodoulou J**. Downstream Targets of MeCP2 and their Abnormal Expression in the Frontal Cortex of the Human Rett Syndrome Brain. BMC Neurosci. 2010:11; 53

133. De Brouwer APM, van Bokhoven H, Nabuurs S, Arts WF, **Christodoulou J**, Duley J. PRPS1 Mutations: Four Distinct Syndromes and Potential Treatment. Amer J Hum Genet. 2010: 86 (4); 506-518.

134. White R, Ho G, Schmidt S, Scheffer IE, Fischer A, Yendle SC, Bienvenu T, Nectoux J, Ellaway CJ, Darmanian A, TongX-Z, Cloosterman D, Bennetts B, Kalra V, Fullston T, Gecz J, Cox TC, **Christodoulou J**. Cyclin-dependent Kinase-like 5 (*CDKL5*) Mutation Screening in Rett Syndrome and Related Disorders. Twin Res Hum Genet. 2010: 13 (2); 168-178.

135. Chung S.-K., Vanbellinghen J.-F., Mullins J.G.L., Robinson A., Hantke J, Hammond C., Gilbert D.F., Freilinger M., Ryan M, Kruer M, Masri A., Gurses C. Ferrie C, Harvey K., Rita Shiang R., **Christodoulou J**, Andermann, , Thomas R.H., [a], Harvey R.J., Lynch J.W., Rees M.I. Pathophysiological mechanisms of dominant and recessive G*LRA1* mutations in human hyperekplexia. J Neurosci. 2010: 30; 9612-9620.

136. Neul JL, Kaufmann WE, Glaze DG, **Christodoulou J**, Clarke AJ, Bahi-Buisson N, Leonard H, Bailey ME, Schanen NC, Kerr AM, Renieri A, Huppke P, Percy AK, for the

RettSearch Consortium. Rett Syndrome: Revised Diagnostic Criteria and Nomenclature. Annals Neurol. 2010: 68 (6); 944-950.

137. Riley LG, Sandra Cooper S, Hickey P, Rudinger-Thirion J, McKenzie M, Compton C, Lim SC, Thorburn DR, Ryan MT, Giegé R, Bahlo M, **Christodoulou J**. A Mutation of the Mitochondrial Tyrosyl-tRNA Synthetase Gene, *YARS2*, Causes Myopathy, Lactic Acidosis, and Sideroblastic Anemia—MLASA Syndrome. Am J Hum Genet. 2010: 87; 52-59.

138. Ho G, Inui K-I, Yonezawa A, Masuda S, Sim K.G., Carpenter K, Olsen RKJ, Mitchell JJ, Rhead W, Peters G, **Christodoulou J**. Maternal Riboflavin Deficiency, Mimicking Neonatal-Onset Glutaric Aciduria Type 2, Is Caused by Mutations in the Riboflavin Transporter *GPR172B*. Human Mut. 2011: 32 (1); E1976-1984.

139. De Greef E, **Christodoulou J**, Alexander IA, Shun A, O'Loughlin EV, Thorburn DR, Jermyn V, Stormon MO. Mitochondrial respiratory chain hepatopathies: role of liver transplantation. A case series of five patients. J Inher Metab Dis Reports. 2012: 4; 5-11.

140. Alodaib A, Carpenter K, Wiley V, Sim K, **Christodoulou J**, Wilcken B. An improved UPLC-MS/MS method for the determination of allo-isoleucine and branched chain amino acids in dried blood spots. Annals of Clinical Biochemistry. 2011: 48(Pt 5); 468 – 470

141. **Christodoulou, J**. Trimethylaminuria: an under-recognised and socially debilitating metabolic disorder. J Paediatr Child Health, 2012: 48; E153-E155.

142. Armani R, Archer H, Clarke A, Vasudevan P, Zweier C, Ho G, Williamson S, Cloosterman D, Yang N, **Christodoulou J**. Transcription Factor 4 and Myocyte Enhancer Factor 2C mutations are not common causes of Rett syndrome. Amer J Med Genet 2012:158A (4); 712-719.

143. Williamson SL, Giudici L, Kilstrup-Nielsen C, Gold W, Pelka GJ, Tam PPL, Grimm A, Prodi D, Landsberger N, **Christodoulou J**. A novel transcript of cyclin-dependent kinase-like 5 (*CDKL5*) has an alternative C-terminus and is the predominant transcript in brain. Human Genet. 2012: 131(2); 187-200.

144. Tucker EJ, Hershman SG, Koehrer C, Belcher-Timme CA, Goldberger OA, **Christodoulou J**, Silberstein J, McKenzie M, Ryan MT, Compton AG, Calvo SE, Rajbhandary UL, Thorburn DR, MoothaVK. Mutations in *MTFMT* underlie a human disorder of formylation causing impaired mitochondrial translation. Cell Metab. 2011: 14; 428–434

145. Duley JA, **Christodoulou J**, de Brouwer APM. The PRPP synthetase spectrum: what does it demonstrate about nucleotide syndromes? Nucleosides, Nucleotides and Nucleic Acids 2011:30 (12); 1129–1139

146. Huppke P, Brendel C, Kahlscheuer V, Korenke GC, Marquardt I, Freisinger P, **Christodoulou J**, Pitelet G, Wilson C, Gruber-Sedlmayr U, Ullmann R, Haas S, Elpeleg O, Nürnberg G, Nürnberg P, Dad S, Møller LB, Kaler SG, Gärtner G. Mutations in *SLC33A1* cause a lethal autosomal recessive disorder with congenital cataracts and hearing loss associated with low serum copper and ceruloplasmin. Amer J Hum Genet 2012: 90(1); 61-68.

147. Calvo SE, Compton AG, Hershman SB, Lim S-C, Lieber DS, Tucker EJ, Laskowski A, Garone C, Liu S, Jaffe DB, **Christodoulou J**, Fletcher JM, Bruno DL, Goldblatt J, DiMauro S, Thorburn DR, Mootha VK. Molecular Diagnosis of Mitochondrial Disorders with Targeted

Next-Generation Sequencing. Science Transl Med 2012: 4(118); 118ra10. (featured on the NIH National Human Genome Research Institute's (NHGRI) Genomic Advance of the Month for January. See www.genome.gov)

148. Moran R, B.P. Kuilenburg ABP, Duley J, Nabuurs SB, Retno-Fitri A, **Christodoulou J**, Roelofsen J, Yntema HG, Friedman NR, van Bokhoven H, de Brouwer APM. Phosphoribosylpyrophosphate Synthetase Superactivity and Recurrent Infections Is Caused by a p.Val142Leu Mutation in PRS-I. Am J Med Genet. 2012: 158A(2); 455-460.

149. Georgiou T, Ho G, Vogazianos M, Dionysiou M, Nicolaou A, Chappa G, Nicolaides P, Stylianidou G, **Christodoulou J**, Drousiotou A. The spectrum of mutations identified in Cypriot patients with phenylalanine hydroxylase deficiency detected through neonatal screening. Clinical Biochem. 2012: 45(7-8); 588-592.

150. Alodaib A, Carpenter K, Wiley V, Wotton T, **Christodoulou J**, Wilcken B. Homocysteine measurement for neonatal in dried blood-spot detection of homocystinurias. J Inher Metab Dis Reports 2012:5; 1-6.

151. Huppke P, Brendel C, Korenke GC, Marquardt I, Donsante A, Yi L, Hicks JD, Steinbach PJ, Wilson C, Elpeleg O, Møller LB, **Christodoulou J**, Kaler SG, Gärtner J. Molecular and biochemical characterization of a unique mutation in *CCS*, the human copper chaperone to superoxide dismutase. Human Mut. 2012: 33 (8); 1207-1215.

152. Garg P, Carpenter K, Chong S, **Christodoulou J**. A Pilot Study of the Effect Of (E, E)-2, 4-undecadienal on the Offensive Odour of Trimethylamine. J Inher Metab Dis Reports. 2013:8; 11-15.

153. Fehr S, Wilson, M, Downs, J, Williams S, Murgia A, Sartori, S, Vecchi M, Ho G, Polli R, Psoni S, Bao X, de Klerk N, Leonard H, **Christodoulou J**. The CDKL5 disorder is an independent clinical entity associated with early-onset encephalopathy. Eur J Human Genet 2013: 21 (3); 266-273.

154. Ellaway CJ, Ho G, Bettella E, Knapman A, Collins C, Hackett A, McKenzie F, Darmanian A, Peters GB, Fagan K, **Christodoulou J**. 14q12 microdeletions excluding *FOXG1* give rise to a Rett syndrome-like phenotype. Eur J Hum Genet. 2013: 21; 522-527.

155. Tchan MC, Wilcken B, **Christodoulou J**. The mild form of Menkes disease: a 34-year progress report on the original case. J Inher Metab Dis Reports 2013: 9: 81-84.

156. Collins D, Angles JM, **Christodoulou J**, Spielman JD, Lindsay SA, Boyd J, Krockenberger MB. Severe Subacute Necrotising Encephalopathy (Leigh-like Syndrome) in American Staffordshire Bull Terrier Dogs. Submitted to J Compar Pathol 2012 Nov 1. doi:pii: S0021-9975(12)00155-7. 10.1016/j.jcpa.2012.08.008

157. Liu X-Z, Xie, D, Yuan, Hui Jun Y, de Brouwer APM, **Christodoulou J**, Yan, D. Hearing loss and PRPS1 mutations: wide spectrum of phenotypes and potential therapy. International Journal of Audiology. 2013: 52 (1); 23-28.

158. Ho G, Reichardt J, **Christodoulou J**. *In vitro* read-through of *PAH* nonsense mutations using aminoglycosides: a potential therapy for phenylketonuria. J Inher Metab Dis. (accepted 4th March 2013)

159. Gaignard, P, Menezes M, Schiff M, Bayot A, Rak M, de Baulny HO, Su C-H, Gillerom M, Lombes A, Abida H, Tzagoloff A, Riley L, Cooper ST, Mina K, Davis MR, Allcock RJN, Kresoje N, Laing NG, Thorburn DR, Slama A, **Christodoulou J**, Rustin P. Mutations in the cytochrome c_1 subunit of respiratory chain complex III cause insulin-responsive hyperglycemia. Accepted Am J Hum Genet 30th May 2013

160. Lim SC, Friemel M, Marum JE, Tucker EJ, Bruno DL, Riley LG, **Christodoulou J**, Kirk EP, Boneh A, deGennaro C, Springer M, Mootha VK, Rouault TA, Leimkühler S, Thorburn DR, Compton AG. Mutations in *LYRM4*, encoding iron-sulfur cluster biogenesis factor ISD11, cause deficiency of multiple respiratory chain complexes. Accepted Hum Molec Genet 20th June 2013.

161. De Brouwer APM, Duley JA, Christodoulou J. Phosphoribosylpyrophosphate Synthetase Superactivity. 2008 Sep 23 [Updated 2013 Aug 8]. In: Pagon RA, Adam MP, Bird TD, et al., editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2013. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1973/

162. Foley AR, Menezes MP, Pandraud A, Gonzalez MA, Alodaib A, Abrams AJ, K Sugano, A Yonezawa, Manzur AY, Burns J, Hughes I, McCullagh BG, Jungbluth H, Lim M, Lin J-P, Megarbane A, Urtizberea JA, Shah AH, Antony J, Webster R, Broomfield A, Ng J, Mathew AA, O'Byrne JJ, Forman E, Scoto M, Prasad M, O'Brien K, Olpin S, Oppenheim M, Hargreaves I, Land JM, Wang MX, Carpenter K, Horvath R, Straub V, Lek M, Gold W, Farrell MO, Brandner S, Phadke R, Matsubara K, McGarvey ML, Scherer SS, Baxter PS, King MD, Clayton P, Rahman S, Reilly MM, Ouvrier RA, **Christodoulou J**, Züchner S, Muntoni F, Houlden H. Treatable Childhood Neuronopathy Caused by Mutations in Riboflavin Transporter RFVT2. Accepted Brain 20th September.

Major Reviews:

1. **Christodoulou J** *Update in Genetic Metabolic Diseases.* Published in "Fellowship Affairs", 1991, produced by the Royal Australasian College of Physicians.
2. **Christodoulou J**, R.R. McInnes RR *Hereditary Metabolic Disease in Infancy: New Concepts and New Disorders.* Current Opinion Paediatr 1992 4; 228-239.
3. **Christodoulou J,** The Clinical, Biochemical and Molecular Pathology of Mitochondrial Disorders. Clinical Biochemistry Reviews 1996 17: 115-124.
4. **Christodoulou J,** The New Genetics and Mitochondrial Respiratory Chain Disorders. Clin Biochemist Rev 2000 21; 14 – 21.
5. Ellaway C, **Christodoulou J**, Rett syndrome: clinical characteristics and recent genetic advances. Disability and Rehabilitation 2001 23; 96 – 106.

6. **Christodoulou J** (updated August 2006) *MECP2*-Related Disorders in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle. 1997-2006. Available at http://www.genetests.org.

7. De Brouwer APM, Duley J, **Christodoulou J** (first posted October 2008) *Arts syndrome* In: Pagon RA, Bird TC, Dolan CR, Stephens K, editors. GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle. 1997-2008. Available at http://www.genetests.org.

8. **Christodoulou J**, Ho G (updated April 2009) *MECP2*-Related Disorders in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle. 1997-2009. Available at http://www.genetests.org.

9. de Brouwer APM, Duley JA, **Christodoulou J**: Arts Syndrome (November 2010) in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle, 1997-2010. Available at http://www.genetests.org.

10. **Christodoulou J**, Ho G (updated June 2012): *MECP2*-Related Disorders in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle. 1997-2009. Available at http://www.genetests.org.

11. de Brouwer APM, Duly JA, **Christodoulou J.** Phosphoribosylpyrophosphate Synthetase Superactivity. in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle. XXXX-XXXX. Available at http://www.genetests.org.

Other Articles/Papers:

1. Thompson GN, **Christodoulou J**, Danks DM. Metabolic stroke in methylmalonic acidaemia. J Pediatr 1989 115; 499-500 (letter to the Editor).
2. **Christodoulou J**, Bankier A, Loughnan PN. Letter to the Editor: Ring chromosome 22 karyotype in a patient with Opitz (BBBG) syndrome. Amer J Med Genet 1990 37; 422-424 (letter to the Editor).
3. **Christodoulou J**. Course Notes for Graduate Diploma in Clinical Chemistry, Pathophysiology 2.1, University of Western Sydney 1994: Principles of Human Genetics.
4. **Christodoulou J**. Course Notes for Masters of Clinical Sciences, University of Western Sydney 1998: Principles of Human Genetics.
5. **Christodoulou J**. Course Notes for Masters of Clinical Sciences, University of Western Sydney 1999: Principles of Human Genetics.
6. Ellaway C, **Christodoulou J.** Rett syndrome: genetic breakthrough. J Paediatr Child Health 1999 35 (6); 593 (letter to the Editor).
7. Shaw PJ, **Christodoulou J**, Mansour A. Bone marrow transplantation for FMF. Blood 2003 101 (3); 1025 – 1026 (response, letter to the Editor).
8. Ho G, Gold W, Williamson S, **Christodoulou J**. Pathogenicity of C-terminal mutations in CDKL5. Journal of Pediatric Epilepsy 3 (2012) 185–186 (letter to the Editor).

Patents:
Modified microorganism and uses thereof – filed October 2012; provision patent approved. Patent number - 2012904813

Refereed Full Length Conference Papers:
1. **Christodoulou J,** Genetic defects causing mitochondrial respiratory chain disorders and disease. Hum Reprod 2000: 15 (suppl. 2); 28 – 43.

Published Abstracts from Conference Proceedings:

1. Bu D-F, **Christodoulou J**, Ploder L,Gibson W, Tobin AJ, McInnes RR. Cloning of the human GAD65 and GAD67 genes, and mutation analysis of GAD65 in patients with pyridoxine (B6)-responsive seizures Amer J Human Genet 1993: 53 (3); 892.

2. Tümer Z, Horn N, **Christodoulou J**, Tønnesen T, Clarke JTR, Sarkar B. Characterization of a severe Menkes genotype resulting in an almost normal phenotype with early copper treatment.Amer J Human Genet 1995: 57 (4); A253.

3. Bu D-F, **Christodoulou J**, Murrell MJ, Ploder L, Gibson W, Tobin AJ, McInnes RR Pyridoxine-responsive epilepsy appears not to be caused by mutations in the *GAD1* or *GAD2* genes. Amer J Human Genet 1995: 57 (4); A177.

4. Adès LC, Holman KJ, Watson KC, Murrell M, Clarke JTR, **Christodoulou J**. Characterisation of an FBN1 gene mutation, G1013R, in a child with neonatal Marfan syndrome (nMFS) and mitochondrial complex I (CI) deficiency. Amer J Human Genet 1997: 61 (4); A404.

5. McQuade L, **Christodoulou J**, Sachdev R, Budarf ML, Emanuel B, Colley A. Two further cases of 22q11.2 deletions with no overlap to the DiGeorge Syndrome Critical Region. Amer J Human Genet 1998: 63 (4); A335.

6. **Christodoulou J**, Williams AJ, Murrell MJ, Brammah S, Minchenko J. Utility of rhodamine 6G (R6G) in assessing the mode of inheritance in mitochondrial respiratory chain disorders. Amer J Human Genet 1998: 63 (4); A264.

7. Nogee LM, Wert SE, Gray P, Masters B, Murrell M, Bennetts B, **Christodoulou J**. Uniparental disomy as a novel mechanism for surfactant protein B (SP-B) deficiency. Pediatr Res 1999: 45 (4); 312A.

8. **Christodoulou J**, Roscioli T, Kamath RK, McQuade L, Murrell M, Bennetts B. *MEFV* Mutations in Familial Mediterranean Fever (FMF): An Australian Experience. J Inher Metab Dis 1999: 22 (suppl 1); 128.

9. Dahl HHM, Hutchison WM, Dasvarma A, Reed K, Kirby DM, Boneh A, **Christodoulou J**, Freckmann ML, Wilcken B, McGill JJ, Fletcher JL, Van Hove J, Thorburn DR. The Clinical Spectrum of Cytochrome c Oxidase Deficiency in Leigh Syndrome Patients with and without Mutations in the *SURF1* gene. Amer J Human Genet 2000: 67 (4); 64.

10. **Christodoulou J**, Minchenko J, Williams A, Kirby DM, Thorburn DR. Somatic Cell Strategies and the Genetic Origin of Mitochondrial Respiratory Chain (RC) Defects. J Inher Metab Dis 2001: 24 (suppl. 1); 77.

11. **Christodoulou J**, Milledge J, Shaw P, Bennetts B, Williamson S, Roscioli T, Mansour A. Bone Marrow Transplantation (BMT): Cure for Familial Mediterranean Fever? . J Inher Metab Dis 2001: 24 (suppl. 1); 150.

12. Raffaele LS, Williamson SL, Bennetts B, Davis M, Ellaway CJ, Leonard H, Thong MK, Delatycki M, Thompson EM, Laing N, **Christodoulou J**. MECP2 Mutation Type, Affected Domain, X-inactivation and Phenotypic Outcome in Rett Syndrome. Amer J Human Genet 2001: 69 (4); 385.

13. **Christodoulou J**, Raffaele LS, Bennetts B, Ouvrier RA. A Syndrome of Deafness, Neurogenic Muscle Weakness and Optic Atrophy: Rosenberg-Chutorian Syndrome (RCS)? A Disease Looking for a Gene. Amer J Human Genet 2001 : 69 (4); 286.

14. Bahuau M, Stuhrmann M, **Christodoulou J**, Lorant T, Houdayer C, Elion J, Couderc R, Griese M, Tredano M. Analysis of Founder Effect in *SFTPB* 1549C→GAA (121ins2) Mutation. Amer J Human Genet 2002: 71 (4); 369.

15. **Christodoulou J,** Biggin A, Thorburn DR, Bennetts B. Mutation Screening of the Mitochondrial Genome Using Denaturing High Performance Liquid Chromatography. Amer J Human Genet 2002: 71 (4); 427.

16. **Christodoulou J,** Minchenko J, Thorburn DR, Coper S. Forced Myogenesis of Fibroblasts Can Unmask Mitochondrial Respiratory Chain Disorders. Amer J Human Genet 2003: 73 (5); 181.

17. **Christodoulou J,** Weaving LS, Williamson SL, Friend KL, McKenzie OLD, Archer H, Evans J, Clarke A, Pelka GP, Tam PPL, Watson CM, Lahooti H, Ellaway CJ, Bennetts B, Leonard H, Gécz J. Mutations of *STK9* Cause Early Onset Seizures, Mental Retardation (MR), an Autistic Disorder and a Rett Syndrome (RTT)-like Phenotype. Amer J Human Genet 2004: 73 (5); 181.

18. **Gibson JH,** Stern J, Slobedman B, Christodoulou J. Downstream Effects of *MECP2* Mutations in the Frontal Cortex of Patients with Rett Syndrome. Amer J Human Genet 2004: 73 (5); 181.

University/Dept Reports:

Produced for the Dept of Paediatrics & Child Health:

Policy in Regard to Requirements for all Higher Degree (by Research) Students in the Department of Paediatrics and Child Health (1999)

Procedure for Postgraduate Student Enrolment in the Department of Paediatrics and Child Health (1999)

Guidelines for the Supervision of Postgraduate Students in Department of Paediatrics and Child Health (1999)

Produced for the Faculty of Medicine:

Research Candidature Working Party Report (2000)

Procedure for Postgraduate Student Enrolment in the Faculty of Medicine at the University of Sydney (2000)

Guidelines for the Supervision of Postgraduate Students in the Faculty of Medicine at the University of Sydney (2000)

Policy: Postgraduate Research Student Supervisor Register (2000)

Application for Admission to the Research Degree Supervisor Register (2000)

Policy for the Resolution of Conflicts between Postgraduate Research Students and Supervisors in the Faculty of Medicine (2000)

Examiner Guidelines for PhD Theses (2002)
This was subsequently adopted as a University-wide guideline document by the Graduate Studies Committee.

Produced for the Human Genetics Society of Australasia:
Best Practice Guidelines for Clinical Genetics Services in Australia
Best Practice Guidelines for Genetic Counselling
A Clinical Approach to Inborn Errors of Metabolism Presenting in the Newborn Period

Unrefereed Conference Papers:
Nil

Conference Abstracts:
58 conference abstracts have been presented where I was first or senior author, and another 33 abstracts where I was co-author (including 27 poster and 64 oral presentations), encompassing 14 regional, 47 national and 30 international conferences. A complete list can be provided if required.

Work in Progress:

1. Saxena A, Scaife R, Croft K, **Christodoulou J**, Zhang P, Beaumont J, Pelka GJ, Tam PPL, Leonard H, Matthijs G, Kavallaris M, Ravine D. MeCP2 maintains microtubule stability by blocking HDAC6 mediated tubulin deacetylation. (submitted to Hum Molec Genet)

2. Kamalanathan P, Wilcken B, Yeo, J, Christodoulou. Haptocorrin deficiency adversely affects metabolic control in cystathionine β-synthase deficiency. Submitted to J Inher Metab Dis

3. Kondo MA, Gray LJ, Leang SK, Pelka GJ, Leang S-W, **Christodoulou J**, Tam PPL, Hannan AJ. Environment enrichment rescues affective and HPA-axis dysfunction in a mouse model of Rett syndrome. Submitted to Hum Molec Genet.

4. Lim SC, Smith KR, Stroud DA, Compton AG, Tucker EJ, Dasvarma A, Gandolfo LC, Marum JE, McKenzie M, Peters HL, Mowat D, Procopis PG, Wilcken B, **Christodoulou J**, Brown GK, Ryan MT, Bahlo M, Thorburn DR. A founder mutation in *PET100* causes isolated complex IV deficiency in Lebanese patients with Leigh Syndrome. Submitted to Am J Hum Genet

5. Trakadis YJ, Alfares A, Bodamer O, Buyukavci M, **Christodoulou J**, Connor P, Glamuzina E, Gonzalez- Fernandez F, Haim B, Echenne B, Manoli I, Mitchell J, Nordvall M, Prasad C, Scaglia F, Schiff M, Schrewe B, Touati G, Tchan MC, Varet B, Venditti C, Zafeiriou D, Rupar A, Rosenblatt DS, Watkins D, Braverman N. Update on

Transcobalamin deficiency: clinical presentation, treatment and outcome. Submitted to J Inher Metab Dis.

6. Riley LG, Menezes MJ, Rudinger-Thirion J, de Lonlay P, Rotig R, Tchan MC, Cooper ST, **Christodoulou J**. Expanding the clinical phenotype and basis for tissue-specificity in YARS2 mitochondrial myopathy, lactic acidosis and sideroblastic anaemia. Submitted to Orphanet J Rare Dis.

7. Tucker EJ, Wanschers BFJ, Szklarczyk R, Mountford HS, Wijeyeratne XW, van den Brand MAM, Leenders AM, Rodenburg R, Compton AG, Frazier AE, Bruno DL, **Christodoulou J**, Endo H, Ryan MT, Nijtmans LG, Huynen MA, Thorburn DR. Mutations in the UQCC-interacting protein, *MNF1,* cause human complex III deficiency associated with perturbed cytochrome *b* protein expression. Submitted to the Am J Hum Genet

8. Williamson SL, Ellaway C, Peters G, Pelka GJ, Tam PPL, **Christodoulou J**. Deletion of protein tyrosine phosphatase, non-receptor type 4 (PTPN4) in twins with a Rett Syndrome-Like Phenotype. To be submitted to the Eur J Hum Genet

9. Gold WA, Williamson SL, Kaur, Hargreaves IP, Land JM, Pelka GJ, Tam PPL, **Christodoulou J**. Mitochondrial Dysfunction in a Mouse Model of Rett Syndrome (RTT): Contribution to Disease Pathogenesis. Submitted to Mitochondrion

10. Menezes MJ, Riley LG, **Christodoulou J**. Mitochondrial Respiratory Chain Disorders in Childhood: Insights into Diagnosis and Management in the New Era of Genomic Medicine. Submitted to Biophys Biochim Acta – Gen Subj

11. Jenny Downs J, Ian Torode I, Ellaway C, Jacoby P, Bunting C, Wong K, Christodoulou J, Leonard L. Family satisfaction following spinal fusion in Rett syndrome. To be submitted to Dev Med Child Neurol.

12. Gladys Ho G, Alexander I, Bhattacharya K, Dennison B, Ellaway CJ, Thompson S, Wilcken B, Christodoulou J. The molecular basis of phenylketonuria (PKU) in New South Wales, Australia: Mutation profile and correlation with tetrahydrobiopterin (BH_4) responsiveness. Submitted to J Inher Metab Dis

Manuscripts in Preparation:

1. Berman Y, Wilson M, Sy C, Schollen E, Matthijs G, Fietz M, Körner C, **Christodoulou J**. Recognisable syndromes amongst the congenital disorders of glycosylation; a new case report

of CDG-Id and review of reported clinical features amongst the CDG subtypes. (in preparation).

2. De Leon EJ, Cooper S, **Christodoulou J**. Improving adenoviral-mediated transductions using the polycationic lipid polybrene. (in preparation; to be submitted to Biotechniques)

3. Gautam A, Lees R, Kirby L, Jatana V, **Christodoulou J**, Bergin M, Jacobe S. Severe Vasodilatory Shock In a Toddler: A Near Fatal Case Of Shoshin Beriberi. Submitted to ??

4. Gold W, **Christodoulou J**. Treatments for Rett Syndrome: Prospects for Targeted Therapies. Book Chapter in preparation.

Abstracts:

Since 1993, I have been a co-author in over 120 abstracts, including 65 posters (9 as first author and 30 as last [senior] author), and 59 oral presentations (12 as first author and 23 as last [senior] author). The breakdown from 1993 – 2002 is as follows:

Year	**Number of Abstracts**		
	Posters	**Orals**	**Subtotal**
1993	1	5	6
1994	2	5	7
1995	3	8	11
1996	3	7	10
1997	4	6	10
1998	6	1	7
1999	2	5	7
2000	12	3	15
2001	13	11	24
2002	19	8	27
Totals	**65**	**59**	**124**

1988:

1. Inheritance of the amelocerebrohypohydrotic syndrome.
   J. Christodoulou, R.K. Hall, S. Menahem, I.J. Hopkins, J.G. Rogers.
   Presented at the 12th Annual Scientific Meeting of the Human Genetics Society of Australasia, Brisbane, Australia (1988).

1989:

2. Load studies in the diagnosis of inborn errors of metabolism.
   J. Christodoulou.
   Presented at the First Australian Conference of Inborn Errors of Metabolism, Adelaide, Australia (1989).

3. The use of the phenylpropionate load test in the diagnosis of medium chain acyl CoA dehydrogenase deficiency.
J. Christodoulou, J. Pitt, D.M. Danks.
Presented at the 13th Annual Scientific Meeting of the HGSA, Alice Springs, Australia (1989).

1990:

4. Identification of a peroxisomal form of malonyl coenyme A decarboxylase.
J. Christodoulou, G.K. Brown, D.M. Danks.
Presented at the Vth International Congress of Inborn Errors of Metabolism (1990). (oral)

1991:

5. Isovalerylcarnitine, isovaleric acidemia, and gut bacterial metabolism.
D.C. Lehotay, J. Christodoulou, L.J. Fisher, B.A. Platt, J.T.R. Clarke.
Presented at the Society for Inherited Metabolic Diseases at Santa Fe, New Mexico, USA (1991). (oral)

6. A unique patient with the neonatal Marfan syndrome and complex I deficiency.
J. Christodoulou, R. Petrova-Benedict, B. Robinson, V. Jay, J. Clarke.
Presented at the 29th Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, London England, 1991. (poster)

1992:

7. The alleles responsible for frequent intragenic complementation at the argininosuccinate lyase (ASAL) locus: mutant monomers capable of subunit interaction.
J. Christodoulou, H.J. Craig, D.C. Walker, R.R. McInnes.
Presented at the Society for Inherited Metabolic Diseases Annual Meeting, Pine Mountain, Georgia, USA, 1992 (oral)

8. Intragenic complementation at the human argininosuccininic acid lyase locus: mutant alleles of the frequently and high activity complementing strains.
H.J. Craig, J. Christodoulou, D.C. Walker, R.R. McInnes.
Presented at the Canadian Genetic Diseases Network National Centres of Excellence Meeting, Montréal, Canada, 1992 (oral)

9. Recurrent pancreatitis in carnitinepalmitoyltransferase (CPT) 2 deficiency.
I. Tein, J. Christodoulou, R. McInnes.
Presented at the SSIEM Annual Symposium, held in September 1992. (oral)

10. A unique patient with the neonatal Marfan syndrome and complex I deficiency.
J. Christodoulou, R. Petrova-Benedict, B. Robinson, V. Jay, J. Clarke.
Presented at the Human Genetics Society of Australasia 16th Annual Scientific Meeting, Newcastle Australia, 1992. (poster)

11. Barth Syndrome: Neuromuscular and cardiovascular outcome.
J. Christodoulou, R.R. McInnes, V. Jay, G. Wilson, L. Becker, D. Lehotay, B.-A. Platt, P.J. Bridge, B.H. Robinson, J.T.R. Clarke.
Presented at the Australian Society of Inborn Errors of Metabolism 4th Scientific Conference, Newcastle Australia, 1992. (oral)

12. A possible deletion "hotspot" in the argininosuccinate lyase (ASAL) gene has topoisomerase II (Topo II) and polymerase α (Pol-α) recognition sites.
J. Christodoulou, H.J. Craig, D.C. Walker, R.R. McInnes.
Presented as a platform presentation (Winner of student competition in the category of Postdoctoral Clinical Student) at the American Society of Human Genetics 42nd Annual Meeting, San Francisco, USA 1992. (oral)

13. Identification of the frequently-complementing and high activity-complementing alleles at the human argininosuccinic acid lyase (ASAL) locus: implications for subunit interaction of mutant monomers.
H.J. Craig, J. Christodoulou, D.C. Walker, R.R. McInnes.
Presented at the American Society of Human Genetics 42nd Annual Meeting, San Francisco, USA 1992. (poster)

1993:

14. Association of a topoisomerase II (topo II) recognition site and polymerase-a (pol-a) induced mutation site with a deletion "hotspot in the argininosuccinate lyase (ASAL) gene.
J. Christodoulou, H.J. Craig, D.C. Walker, and R. R. McInnes.
Presented at the Lorne Genome Conference, Lorne Australia 1993. (oral)

15. A deletion "hotspot" in the argininosuccinate lyase (ASAL) gene has both a topo II recognition site and a DNA polymerase a (pol a) mutation site.
R. R. McInnes, J. Christodoulou, H.J. Craig, and D.C. Walker.
Presented at American Paediatric Society meeting, 1993. (oral)

16. Cloning of the human GAD65 and GAD67 genes, and mutation analysis of GAD65 in patients with pyridoxine (B6)-responsive seizures.
D.-F. Bu, J. Christodoulou, L. Ploder, W. Gibson, A.J. Tobin, R.R. McInnes.
Presented at the American Society of Human Genetics 43rd Annual Meeting, New Orleans, USA 1993 (poster # 892).

17. Genetic metabolic disorders of neurotransmitter metabolism.
J. Christodoulou.
Presented at the Fifth Australaisan Inborn Errors of Metabolism Conference, September, Canberra Australia 1993. (oral)

18. Zellweger syndrome in a premature infant with prolonged conjugated hyperbilirubinemia.
R. Jamieson, J. Christodoulou, M. Wilson, J. Hughes, A. Poulos.
Presented at the Fifth Australaisan Inborn Errors of Metabolism Conference, September, Canberra Australia 1993. (oral)

19. Lethal infantile cytochrome c oxidase deficiency.
R. Ogle, J. Christodoulou, D. Sillence, M. Latham, B. Wilcken.
Presented at the Fifth Australaisan Inborn Errors of Metabolism Conference, September, Canberra Australia 1993. (oral)

1994:

20. Mitochondrial myopathy with complex 1 deficiency responsive to riboflavin and carnitine.
R. Ogle, J. Christodoulou, E. Fagan, J. Walsh, D. Thorburn.
Presented at the VIth International Congress of Inborn Errors of Metabolism, Milan in May 1994. (oral)

21. Neonatal onset of medium chain acyl-CoA dehydrogenase (MCAD) deficiency with confusing biochemical features.
J. Hoare, J. Christodoulou, J. Hammond, B. Wilcken.
Presented at the VIth International Congress of Inborn Errors of Metabolism, Milan in May 1994. (oral)

22. Evolution of classical Menkes disease (MD) to an occipital horn syndrome (OHS) phenotype with copper-histidinate therapy.
J. Christodoulou, B. Sarkar, J.T.R. Clarke.
Presented at the VIth International Congress of Inborn Errors of Metabolism, Milan in May 1994. (oral)

23. Methylmalonic Acidemia - a 19 year review.
R. Jamieson, B. Wilcken, J. Christodoulou, J. Hammond.
Presented at the Sixth Australasian Inborn Errors of Metabolism Conference, August 1994, New Zealand. (oral)

24. Pharmacokinetics of carnitine in two carnitine uptake defect patients.
Teo SH, Earl J, Hammond J, Wilcken B, Christodoulou.
Presented at the Sixth Australasian Inborn Errors of Metabolism Conference, August 1994, New Zealand. (oral)

25. Mutation Analysis of GAD65 and GAD67 in Patients with Pyridoxine (B6)-dependent Epilepsy.
Bu D-F, Christodoulou J, Ploder L, Murrell M, Gibson W, Tobin AJ, McInnes RR.
Presented at the Inaugural University of Sydney Faculty of Medicine Research Conference, Leura Sydney, October 1994. (poster)

26. Functional and Molecular Studies of Cytochrome c Oxidase Deficiency.
Williams AJ, Coakley J, Kemp J, Smith A, Christodoulou J.
Presented at the Inaugural University of Sydney Faculty of Medicine Research Conference,
Leura Sydney, October 1994. (miniposter)

1995:

27. Mutation Analysis of GAD65 and GAD67 in Patients with Pyridoxine (B6)-dependent Epilepsy.
Bu D-F, Christodoulou J, Ploder L, Murrell M, Gibson W, Tobin AJ, McInnes RR.
Presented at the Annual Scientific Meeting of the Paediatric Research Society of Australia & Australian College of Paediatrics, Adelaide, May 1995. (oral)

28. Prenatal diagnosis of the carnitine transporter defect.
Christodoulou J, Teo SH, Hammond J, Ip W-C, Sim KG, Stanley CA, Lau KC, and Wilcken B.
Presented at the Annual Scientific Meeting of the Paediatric Research Society of Australia & Australian College of Paediatrics, Adelaide, May 1995 (winner Best Poster).

29. Nonketotic hyperglycinaemia: the NSW experience and the relevance of transient cases.
Goodwin L, Christodoulou J, Hammond J, Potter M, Wilcken B.

Presented at the Australasian Soeciety for Inborn Errors of Metabolism Annual Scientific Meeting, held in Brisbane, September 1995. (oral)

30. Prenatal diagnosis of the carnitine transporter defect.
Christodoulou J, Teo SH, Hammond J, Ip W-C, Sim KG, Stanley CA, Lau KC, Watson B, and Wilcken B.
Presented at the Australasian Society for Inborn Errors of Metabolism Annual Scientific Meeting, held in Brisbane, September 1995. (oral)

31. Mitochondrial myopathy with $tRNA^{Leu(UUR)}$ mutation and associated complex I deficiency responsive to riboflavin.
Ogle RF, Christodoulou J, Fagan E, Blok RB, Dahl H-HM, Thorburn DR.
Presented at the Australasian Society for Inborn Errors of Metabolism Annual Scientific Meeting, held in Brisbane, September 1995. (oral)

32. An unusual presentation of a respiratory chain enzyme defect relatively specific to the liver.
Freckman M-L, Thorburn DR, Hammond J, Kamath R, Christodoulou J.
Presented at the Australasian Society for Inborn Errors of Metabolism Annual Scientific Meeting, held in Brisbane, September 1995. (oral)

33. Argininsuccinic aciduria in pregnancy.
Worthington S, Wilcken B, Peat B, Stewart P, Christodoulou J.
Presented at the Australasian Society for Inborn Errors of Metabolism Annual Scientific Meeting, held in Brisbane, September 1995. (oral)

34. NSW computerised database of patients with phenylketonuria.
Thompson S, Gruca M, Christodoulou J.
Presented at the Australasian Society for Inborn Errors of Metabolism Annual Scientific Meeting, held in Brisbane, September 1995. (oral)

35. Nonketotic hyperglycinaemia in an adult with ketosis.
Sachdev RK, Wilcken B, Hammond J, Christodoulou J.
Presented at the Australasian Society for Inborn Errors of Metabolism Annual Scientific Meeting, held in Brisbane, September 1995. (oral)

36. Pyridoxine-responsive epilepsy appears not to be caused by mutations in the *GAD1* or *GAD2* genes.
Bu D-F, Christodoulou J, Murrell MJ, Ploder L, Gibson W, Tobin AJ, McInnes RR
Presented at the 45th American Society of Human Genetics Annual Scientific Meeting, Minneapolis, October 1995. (poster)

37. Characterization of a severe Menkes genotype resulting in an almost normal phenotype with early copper treatment.
Tümer Z, Horn N, Christodoulou J, Tønnesen T, Clarke JTR, Sarkar B.
Presented at the 45th American Society of Human Genetics Annual Scientific Meeting, Minneapolis, October 1995. (poster)

1996:

38. Inborn Errors of Metabolism in the Paediatric Intensive Care Unit.
Ferris PE, Schell DN, Gillis J, Christodoulou J.

Presented at the 11th World Congress of Anaesthesiologists, Sydney, April 1996. (poster)

39. Genetics of Mitochondrial Disorders.
Christodoulou J.
Invited speaker at the Annual Scientific Meeting of the Paediatric Research Society of Australasia and the Australian College of Paediatrics, Adelaide, May 1996. (oral)

40. Mitochondrial Disorders: Clinical, Biochemical and Genetic Considerations.
Christodoulou J.
Invited Plenary speaker for the AIMS/AACB Tropical Division Conference, Mackay, June 1996. (oral)

41. Protein Losing Enteropathy in a Baby with Homocystinuria and Methylmalonic Aciduria.
Ellaway C, Christodoulou J, Kamath R, Wilcken B.
Presented at the Australasian Soeciety for Inborn Errors of Metabolism Annual Scientific Meeting, Adelaide, September 1996. (oral)

42. Gaucher Disease Presenting as a Collodion Baby.
Christodoulou J, Ellaway C, Arbuckle S, Murray H, Carey W, Nelson P.
Presented at the Australasian Soeciety for Inborn Errors of Metabolism Annual Scientific Meeting, Adelaide, September, 1996. (oral)

43. Experience of Alglucerase Therapy for Gaucher Disease at the New Children's Hospital Westmead.
Sachdev RK, Wilson M, Christodoulou J, Hayden M, McLean K
Presented at the Australasian Soeciety for Inborn Errors of Metabolism Annual Scientific Meeting, Adelaide, September, 1996. (oral)

44. Velocardiofacial Syndrome (VCFS): A Molecular Cytogenetic Investigation of an Australian Cohort.
McQuade LR, Di K, Fagan K, Colley PW, Christodoulou J, Emanuel B, Lipson AH, Colley AF.
Presented at the Human Genetics Society of Australasia Annual Scientific Meeting, Adelaide, September 1996. (oral)

45. Molecular Studies of Galactosaemia: Frequently Occurring Mutations of Galactose-1-Phosphate Uridyl Transferase (GALT).
Urwin RE, Christodoulou J, Wilcken B, Wiley VC.
Presented at the Human Genetics Society of Australasia Annual Scientific Meeting, Adelaide, September 1996. (oral)

46. Effect of Rhodamine 6G on Cytochrome c Oxidase Activity in Cultured Skin Fibroblasts.
Williams A, Coakley J, Christodoulou J.
Presented at the Faculty of Medicine Research Conference, held in Wollongong, October 1996. (miniposter)

47. Molecular Bases of Galactosaemia in Australia.
Urwin R, Christodoulou J, Murrell M, Wiley V, Wilcken B.
Presented at the Faculty of Medicine Research Conference, held in Wollongong, October 1996. (miniposter)

1997:

48. Complex III Deficiency with Pruritus: a Novel Vitamin Responsive Clinical Feature.
Christodoulou J, Mowat D, Kirby DM, Kamath KR, Arbuckle S, Thorburn DR.
Presented at the 7th International Congress of Inborn Errors of Metabolism, Vienna, Austria, May 1997. (oral)

49. Evaluation of a Second Tier to Newborn Screening for Galactosemia: Utility of N314D Mutation Screening.
Urwin R, Christodoulou J, Wiley V, Murrell M, Wilcken B.
Presented at the 7th International Congress of Inborn Errors of Metabolism,Vienna, Austria, May 1997. (oral)

50. Reduction of Mitochondrial Enzyme Activities in Human Skin Fibroblasts using Rhodamine 6G.
Williams AJ, Murrell M, Coakley J, John Christodoulou J.
Presented at the 7th International Congress of Inborn Errors of Metabolism,Vienna, Austria, May 1997. (poster)

51. The Effect of Rhodamine 6G on the Structure and Function of Mitochondria.
Williams AJ, Murrell M, Coakley J, Christodoulou J.
Presented at the Human Genetics Society of Australasia Annual Scientific Meeting, Perth, July 1997. (poster)

52. Rett Syndrome: Phenotypic Overlap with Angelman Syndrome but not with Methylation Studies.
Ellaway C, Buchholtz T, Smith A, Leonard H, Christodoulou J.
Presented at the Human Genetics Society of Australasia Annual Scientific Meeting, Perth, July 1997. (oral)

53. VCFS/DiGeorge Syndromes: Lessons from Mice and Chicks.
McQuade L, Christodoulou J, Colley PW, Fagan KA, Colley AF.
Presented at the Human Genetics Society of Australasia Annual Scientific Meeting, Perth, July 1997. (oral)

54. Plasma Methylmalonate Estimation: Its Potential Role as a Therapeutic Marker.
Carpenter K, Wilcken B, Wai I, Christodoulou J.
Presented at the Human Genetics Society of Australasia Annual Scientific Meeting, Perth, July 1997. (oral)

55. Carnitine: Double Blinded Crossover Trial in 35 Females with Rett Syndrome.
Ellaway C, Sim KG, Leonard H, Hayden M, Wilcken B, Christodoulou.
Presented at the Human Genetics Society of Australasia Annual Scientific Meeting, Perth, July 1997. (oral)

56. Radiological Clues to the Neuroendocrine Basis of Rett Syndrome.
Thompson R, Glasson E, Fyfe S, Leonard S, Ellaway CJ, Christodoulou J, Bower C, Leonard H.
Presented at the Human Genetics Society of Australasia Annual Scientific Meeting, Perth, July 1997. (poster)

57. The Hands and Feet in Rett Syndrome.

Glasson E, Thompson R, Fyfe S, Leonard S, Bower C, Rousham E, Ellaway CJ, Christodoulou J, Leonard H.
Presented at the Australasian College of Paediatric Annual Meeting, Christchurch, New Zealand, August 1997. (poster)

1998:

58. Genetic Analysis of an Australian Case of Surfactant Protein B Deficiency.
Christodoulou J, Williams GD, Stack J, Symons P, Wert SE, Murrell MJ, Nogee LM.
Presented at the Human Genetics Society of Australasia/Australasian Society of Inborn Errors of Metabolism Conference, Melbourne, July 1998. (poster)

59. Mutation Analysis in Ornithine Transcarbamoylase Deficiency.
Minchenko J, McQuade L, Bennetts B, Christodoulou J.
Presented at the Human Genetics Society of Australasia/Australasian Society of Inborn Errors of Metabolism Conference, Melbourne, July 1998 . (poster)

60. The 22q Deletion Disorders: an Evolving Phenotype.
Sachdev RK, Colley A, Wilson M, McQuade L, Christodoulou J.
Presented at the Human Genetics Society of Australasia/Australasian Society of Inborn Errors of Metabolism confernce, Melbourne, July 1998. (poster)

61. Dietary Management of Long Chain 3-hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency.
Thompson S, Wilcken B, Christodoulou J.
Presented at the Human Genetics Society of Australasia/Australasian Society of Inborn Errors of Metabolism confernce, Melbourne, July 1998. (oral)

62. Oral Health of Children with Phenylketonuria.
Kilpatrick N, Christodoulou J, Wilcken B.
Presented at the Faculty of Medicine Research Conference, held in Leura, October 1998. (miniposter)

63. Two Further Cases of 22q11.2 deletions with no overlap to the DiGeorge Syndrome Critical Region.
McQuade L, Christodoulou J, Sachdev R, Budarf ML, Emanuel B, Colley A.
Presented at the American Society of Human Genetics conference, held in Denver, USA, October 1998 (published in the Amer J Human Genet 1998: 63 (4); A335). (poster)

64. Utility of Rhodamine 6G (R6G) in assessing the mode of inheritance in mitochondrial respiratory Chain Disorders.
Christodoulou J, Williams AJ, Murrell MJ, Brammah S, Minchenko J.
Presented at the American Society of Human Genetics conference, held in Denver, USA, October 1998 (published in the Amer J Human Genet 1998: 63 (4); A264). (poster)

1999:

65. Rett Syndrome: Diagnostic Strategies and Therapeutic Intervention
Ellaway CJ, Williams K, Leonard H, Wilcken B, Christodoulou J.
Presented at the Australasian Society for the Study of Intellectual Disability, 35th Quality Lifestyles Conference, Sydney, September 1999. (oral)

66. Cytochrome c Oxidase Deficiency – Let's Go Surfing!
Thorburn DR, Hutchison WM, Reed KJ, Kirby DM, White SL, Boneh A, Christodoulou J, Dahl H-HM.
Presented at the Human Genetics Society of Australasia/ Australasian Society for Inborn Errors of Metabolism Annual Scientific Conference, Sydney, September 1999. (oral)

67. Effect of Timing of Protein Substitute on Phenylalanine Level.
Holliday K, Christodoulou J.
Presented at the Human Genetics Society of Australasia/ Australasian Society for Inborn Errors of Metabolism Annual Scientific Conference, Sydney, September 1999. (oral)

68. *MEFV* Mutations Detected in a Sydney Population with Familial Mediterranean Fever.
Roscioli R, Bennetts B, Kamath RK, McQuade L, Murrell M, Lonergan S, Christodoulou J.
Presented at the Human Genetics Society of Australasia/ Australasian Society for Inborn Errors of Metabolism Annual Scientific Conference, Sydney, September 1999. (poster)

69. Genetic Studies in Rett syndrome: identification of further familial cases and X-inactivation studies.
Ellaway CJ, Bennetts B, Christodoulou J.
Presented at the Human Genetics Society of Australasia/ Australasian Society for Inborn Errors of Metabolism Annual Scientific Conference, Sydney, September 1999. (oral)

70. Spinocerebellar Ataxia type 7 (SCA7): Genetic Anticipation and Prenatal Diagnosis.
Christodoulou, Healey S, Hyland B, Nicholson G, Grigg J, Wilson M, Grattan-Smith P.
Presented at the Human Genetics Society of Australasia/ Australasian Society for Inborn Errors of Metabolism Annual Scientific Conference, Sydney, September 1999. (oral)

71. *MEFV* Mutations in Familial Mediterranean Fever (FMF): An Australian Experience.
Christodoulou J, Roscioli T, Kamath RK, McQuade L, Murrell M, Bennetts B.
Presented at the Society for the Study of Inborn Errors of Metabolism Annual Scientific Meeting, Genoa, Italy, September 1999 (Published in abstract form in the J Inher Metab Dis 1999 22 (suppl 1); 128). (poster)

2000:

72. Detection of Defects of the Mitochondrial Respiratory Electron Transport Chain Using Flow Cytometry.
Williams A, Setterfield, Thorburn D, Kirby D, Christodoulou J.
AIMS Conference, Sydney, June 2000. (oral)

73. Rett Syndrome in Australia: Seven Years On.
Leonard H, Leonard S, Bower C, Christodoulou J, Davis M, Raffaele L, Fyfe S, Ellaway C.
World Congress on Rett Syndrome, Karuizawa, Nagano, Japan, July 2000. (poster)

74. Recurrent Mutations in Rett Syndrome (RS) Patients.
Raffaele L, Williamson S, Bennetts B, Davis M, Leonard H, Ellaway C, Christodoulou J.
World Congress on Rett Syndrome, Karuizawa, Nagano, Japan, July 2000. (poster)

75. Effect of *MECP2* Mutation Type and Skewing of X-Inactivation on Severity of Outcome in Rett Syndrome (RS).
Raffaele L, Williamson S, Bennetts B, Davis M, Leonard H, Ellaway C, Christodoulou J.
World Congress on Rett Syndrome, Karuizawa, Nagano, Japan, July 2000. (poster)

76. Sleep Dysfunction in Rett Syndrome
Ellaway C, Peat J, Leonard H, Christodoulou J.
World Congress on Rett Syndrome, Karuizawa, Nagano, Japan, July 2000. (poster)

77. Development of the Rett Syndrome: Symptom Index Score.
Ellaway C, Peat J, Christodoulou J.
World Congress on Rett Syndrome, Karuizawa, Nagano, Japan, July 2000. (poster)

78. *MECP2* and Rett Syndrome (RS) – Time for a Mutation Database?
Christodoulou J, Weaving L, Ellaway C, Leonard H, Bennetts B.
World Congress on Rett Syndrome, Karuizawa, Nagano, Japan, July 2000. (poster)

79. Long Term Open Label Trial of L-carnitine in Rett Syndrome.
Ellaway C, Peat J, Williams K, Leonard H, Christodoulou J.
World Congress on Rett Syndrome, Karuizawa, Nagano, Japan, July 2000. (poster)

80. Flow Cytometry in the Study of Mitochondrial Respiratory Chain Disorders.
Setterfield K, Williams A, Donald J, Christodoulou J.
Westmead Hospital Research Symposium, Sydney, Australia, August 2000. (poster)

81. Relationship Between Laboratory Findings, Phenotype and Genotype in OTC Deficiency.
Green K, Minchenko J, Bennetts B, Wilcken B, Christodoulou J.
Human Genetics Society of Australasia Annual Scientific meeting, Wellington, New Zealand, September 2000. Bulletin of HGSA 2000 13(2); 33. (oral)

82. Effect of *MECP2* Mutation type and Skewing of X-inactivation on Prognosis in Rett Syndrome.
Raffaele LS, Williamson S, Bennetts B, Davis M, Leonard H, Ellaway C, Christodoulou J.
Human Genetics Society of Australasia Annual Scientific Meeting, Wellington, New Zealand, September 2000. Bulletin of HGSA 2000 13(2); 43 . (oral)
(LR was winner of award for best student oral presentation)

83. The Clinical Spectrum of Cytochrome c Oxidase Deficiency in Leigh Syndrome Patients with and without Mutations in the SURF1 gene.
Dahl HHM, Hutchison WM, Dasvarma A, Reed K, Kirby DM, Boneh A, Christodoulou J, Freckmann ML, Wilcken B, McGill JJ, Fletcher JL, Van Hove J, Thorburn DR.
American Society of Human Genetics Annual Meeting, Philadelphia, October 2000
Am J Hum Genet 2000 67 (4); 64. (poster)

84. Qunatitative Fibroblast Acylcarnitines Profiling and Correlation of Disease Phenotype in the Diagnosis of Fatty Acid Oxidation Disorders
Sim KG, Christodoulou J, Carpenter K, Hammond J, Wilcken B.

University of Sydney College of Health Sciences Congress, Leura, NSW, Australia, November 2000. (miniposter)

85. MeCP2, X-inactivation and Rett Syndrome (RS)
Raffaele LS, Williamson S, Davis M, Leonard H, Ellaway C, Bennetts B, Christodoulou J.
University of Sydney College of Health Sciences Congress, Leura, NSW, Australia, November 2000. (miniposter)

86. Towards Identifying the Genetic Origin of Respiratory Chain Defects.
Minchenko J, Williams AJ, Thorburn DR, Newbold RF, Reddell RR, Trounce IA, Christodoulou J.
University of Sydney College of Health Sciences Congress, Leura, NSW, Australia, November 2000. (miniposter)

2001:

87. The Australian Lebanese Community has a High Prevalence and Wide Variety of "Mitochondrial" Disorders: Implications for other Consanguineous Groups.
Thorburn DR, Christodoulou J, Mowat D, Halliday J, Kirby DM, Skadal D.
17th World Congress of Neurology, London, England, June 2001. (poster)

88. Raffaele LS, Williamson Sl, Bennetts B, Davis M, Ellaway C, Leonard H, Christodoulou J. *MECP2* Mutation Type, Location and X-inactivation and Phenotype in Rett Syndrome (RTT). Lorne Genome Meeting, Lorne, Victoria, February 2001. (oral)

89. Raffaele LS, Williamson S, Davis M, Ellaway C, Bennetts B, Leonard H, Christodoulou J. *MECP2* Mutation Type, Location and X-inactivation and Outcome in Rett Syndrome (RTT). Dept of Paediatrics Postgraduate Student Conference, Parramatta, NSW, March 2001. (miniposter)

90. Minchenko J, Williams AJ, Thorburn DR, Newbold RF, Reddell RR, Trounce IA, Christodoulou J. Towards Identifying the Genetic Origin of Respiratory Chain Defects. Dept of Paediatrics Postgraduate Student Conference, Parramatta, NSW, March 2001. (miniposter)

91. Ch'ng's presentation at the Dept of Paediatrics Postgraduate Student Conference, Parramatta, NSW, March 2001. (miniposter)

92. Raffaele LS, Williamson S, Davis M, Ellaway C, Bennetts B, Leonard H, Christodoulou J. *MECP2* Mutation Type, Location and X-inactivation and Outcome in Rett Syndrome (RTT). Annual Hospital Meeting, Children's Hospital at Westmead, Sydney March 2001 (Winner of Young Investigator Award). (oral)

93. Chung M, Ellaway C, Christodoulou J. The Rett syndrome multidisciplinary management clinic – survey of parental satisfaction. Australasian Society of Genetic Counsellors, Newcastle, May 2001 (poster).

94. Raffaele LS, Williamson S, Leonard H, Davis M, Ellaway C, Bennetts B, Christodoulou J. *MECP2* Mutation Type, X-inactivation and Outcome in Rett Syndrome. Australian Society for Medical Research Annual Scientific Conference, Sydney, June 2001 (oral).

95. Thorburn D, Christodoulou J, Mowat D, Halliday J, Kirby D, Skladal D. The Australian Lebanese community has a high prevalence and wide variety of "mitochondrial" disorders. Australasian Society for Inborn Errors of Metabolism Annual Scientific Conference, Cairns, July 2001 (oral).

96. Christodoulou J, Minchenko J, Williams A, Kirby D, Thorburn D. Somatic cell strategies and the genetic origin of mitochondrial respiratory chain (RC) disorders. Australasian Society for Inborn Errors of Metabolism Annual Scientific Conference, Cairns, July 2001 (oral).

97. Sim K, Carpenter K, Hammond J, Christodoulou J, Wilcken B. Acylcarnitine profiles in fibroblasts from patients with respiratory chain defects can resemble those with mitochondrial fatty acid β-oxidation disorders. Australasian Society for Inborn Errors of Metabolism Annual Scientific Conference, Cairns, July 2001 (oral).

98. Lal V, Wiley V, Haydon M, Bayliss U, Alexander I, Christodoulou J, Ellaway C, Wilcken B. BH_4 loading in hyperphenylalaninaemia. Australasian Society for Inborn Errors of Metabolism Annual Scientific Conference, Cairns, July 2001 (poster).

99. Lewis S, Haquet E, Christodoulou J, Thorburn D, Dahl H. Characterisation of a chromosomal breakpoint in a patient with complex I deficiency. Human Genetic Society of Australasia Annual Scientific Conference, Cairns, July 2001 (oral).

100. *MECP2* Mutation Type, X-inactivation and Outcome in Rett Syndrome. Human Genetics Society of Australasia Annual Scientific Conference, Cairns, July 2001 (winner of best student oral presentation). (oral)

101. Christodoulou J, Gray P, Murrell M, Bennetts B, McQuade L, Nogee L. Surfactant-protein B deficiency: novel pathogenetic mechanism. Human Genetics Society of Australasia Annual Scientific Conference, Cairns, July 2001 (oral).

102. Christodoulou J. Clinical update on Rett syndrome. Human Genetics Society of Australasia Annual Scientific Conference, Cairns, July 2001 (oral).

103. Christodoulou J, Milledge J, Shaw P, Bennetts B, Williamson S, Roscioli T, Mansour A. Bone marrow transplantation (BMT): cure for familial Mediterranean fever? Human Genetics Society of Australasia Annual Scientific Conference, Cairns, July 2001 (poster).

104. Leonard H, Raffaele L, Eastaugh P, Smith L, Delatycki M, Witt-Engerstrom I, Christodoulou J. Trisomy 21 and Rett syndrome: a double burden. . Human Genetics Society of Australasia Annual Scientific Conference, Cairns, July 2001 (poster).

105. Raffaele LS, Williamson S, Bennetts B, Davis M, Leonard H, Ellaway C, Christodoulou J. *MECP2* Mutation Type, location and X-inactivation on Phenotypic Outcome in Rett Syndrome. Westmead Hospital Annual Scientific Meeting, Sydney, August 2001 (poster).

106. Raffaele LS, Williamson SL, Bennetts B, Davis M, Ellaway CJ, Leonard H, Thong MK, Delatycki M, Thompson EM, Laing N, Christodoulou J. MECP2 Mutation Type, Affected Domain, X-inactivation and Phenotypic Outcome in Rett Syndrome. American Society of Human Genetics Annual Scientific Conference,San Diego, October 2001 (poster).

107.Christodoulou J, Raffaele LS, Bennetts B, Ouvrier RA. A Syndrome of Deafness, Neurogenic Muscle Weakness and Optic Atrophy: Rosenberg-Chutorian Syndrome (RCS)? A Disease Looking for a Gene. American Society of Human Genetics Annual Scientific Conference,San Diego, October 2001 (poster).

108.Raffaele LS, Williamson SL, Bennetts B, Davis M, Ellaway CJ, Leonard H, Thong M-K, Delatycki M, Thompson EM, Laing N, Christodoulou J. *MECP2* Mutations, X-inactivation and Phenotype in Rett Syndrome. ASBMB ComBio Conference, Canberra, October 2001 (poster).

109.Shaw PJ, Christodoulou J, Milledge J, Williamson S, Bennetts B, Roscioli T, Mansour A. Can bone marrow transplantation cure familial Mediterranean fever? 43rd Annual Society of Hematology Conference, US, December 2001 (Abstract published in Blood 2001: 98; 380b – 381b). (poster)

110.Leonard H, Colvin L, Fyfe S, Leonard S, Schiavello T, Bower C, De Klerk N, Nagarajan L, Johansson C, Christodoulou J, Ellaway C, Raffaele L, Davis M. Epidemiological update of Rett syndrome following recent genetic advances. Princess Margaret Hospital Annual Research Meeting, Perth, October 2001. (oral)

2002:

111.Rochefort MJ, Lloyd J, Christodoulou J, Nogee L. Surfactant Protein B (SP-B) Deficiency in a Preterm Infant. Perinatology Society of Australia and New Zealand Annual Scientific Conference, To be held in Christchurch, New Zealand, March 2002. (poster)

112.Rae C, Joy P, Harasty JA, Kemp A, Kuan S, Christodoulou J, Cowell CT, Coltheart M. The X Chromosome and the Brain: a Magnetic Resonance and Neuropyschological Study of Turner Syndrome. International Society of Magnetic Resonance Medicine, Honolulu, US, May 2002. (poster) Refereed conference paper – Proc Intl Soc Magn Reson Med 2002 XX; 696.

113.Raffaele LS, Christodoulou J, Bennetts B, Ouvrier RA. Microsatellite Mapping and Screening of a Candidate Gene, *TRPC5* (Transient Receptor Potential Channel) in a Second Family with Arts Syndrome. European Society of Human Genetics Conference, Franc, June 2002. (poster)

114.Christodoulou J, Grimm A, Maher T, Bennetts B. RettBASE: the IRSA MECP2 Variation Database. A New Mutation Database Offering Enhanced Capabilities. Rett Syndrome Research Foundation Conference, Baltimore, US, June 2002. (poster)

115.Leonard H, Colvin L, Fyfe S, Christodoulou J, Raffaele L, Davis M, Ellaway C, Leonard S, de Klerk N. Now that the gene has been found: describing the phenotype in Rett syndrome using a national database. Rett Syndrome Research Foundation Conference, Baltimore, US, June 2002. (poster)

116.Williamson S, Bennetts B, Gedeon A, Raffaele L, Turner G, Ellaway C, Leonard H, Christodoulou. Screening Methyl-CpG Binding Protein-2 (*MECP2*) by Denaturing High Performance Liquid Chromatography (DHPLC). Mutation Screening Satellite Conference, Adelaide, SA, July 2002. (oral)

117.Williamson S, Bennetts B, Gedeon A, Raffaele L, Turner G, Ellaway C, Leonard H, Christodoulou J. Screening of the Methyl-CpG Binding-Protein 2 (*MECP2*) Gene in Non-

Syndromic Mental Retardation (MRX). Mutation Screening Satellite Conference, Adelaide, SA, July 2002. (oral)

118.Minchenko J, Williams A, Biggin A, Thorburn D, Trounce I, Christodoulou. A Family with Mitochondrial Respiratory Chain Defects: X-linked or Mitochondrial? Australasian Society for Inborn Errors of Metabolism Conference, Adelaide, SA, July 2002. (oral)

119.Kennedy C, Christodoulou J, Lonergan S, Nogee L, Bennetts B. Mutation Detection for Hereditary Surfactant Protein-B Deficiency. Human Genetics Society of Australasia Conference, Adelaide, SA, July 2002. (poster)

120.Leonard H, Colvin L, Fyfe S, Christodoulou J, Raffaele L, Ellaway C, Davis M, de Klerk N. The Evolution of Rett Syndrome from Phenotype to Genotype. Human Genetics Society of Australasia Conference, Adelaide, SA, July 2002. (oral)

121.Raffaele LS, Christodoulou J, Bennetts B, Ouvrier RA. Refinement of the Candidate Region for Arts Syndrome. Human Genetics Society of Australasia Conference, Adelaide, SA, July 2002. (oral)

122.Minchenko J, Williams A, Biggin A, Christodoulou. Towards Identifying the Genetic Origin of Respiratory Chain Defects. School of Paediatrics & Child Health Postgraduate Student Workshop, Sydney, July 2002. (miniposter)

123.Pelka G, Watson C, Christodoulou J, Tam P. Development of Mouse Models of Rett Syndrome. School of Paediatrics & Child Health Postgraduate Student Workshop, Sydney, July 2002. (miniposter)

124.Gibson J, Bennetts B, Slobedman B, Armati P, Christodoulou J. Gene Expression in Rett Syndrome. Experimental Design in Microarrays: Issues and Solutions Conference (Sydney Microarray Users' Group), Sydney, August 2002. (oral)

125.Sim KG, Carpenter K, Hammond J, Christodoulou J, Wilcken B. Fibroblast Acylcarnitine Profiling in the Diagnostic and Prognostic Evaluation of Fatty Acid β-oxidation Defects. Society for the Study of Inborn Errors of Metabolism Conference, Dublin, Ireland, September 2002. (poster) Published Abstract – J Inher Metab Dis 2002 25 (suppl. 1); 75

126.Sim KG, Carpenter K, Hammond J, Christodoulou J, Wilcken B. Fibroblast Acylcarnitines in Mitochondrial Fatty Acid β-oxidation Defects: Phenotype/Metabolite Correlation. Society for the Study of Inborn Errors of Metabolism Conference, Dublin, Ireland, September 2002. (poster)

127.Booth D, Bennetts B, Christodoulou J, Stewart G. FMF in Australia – an under-diagnosed disease? 3rd International Conference on Familial Mediterranean Fever and Hereditary Inflammatory Disorders, Montpellier, France, September 2002. (oral)

128.Bennetts B, Oxley P, Lonergan S, Roscioli T, Christodoulou J. MEFV Mutations in Familial Mediterranean Fever (FMF): An Australian Experience. 3rd International Conference on Familial Mediterranean Fever and Hereditary Inflammatory Disorders, Montpellier, France, September 2002. (poster)

129.Williamson S, Bennetts B, Gedeon A, Weaving L, Turner G, Ellaway C, Leonard H, Christodoulou J. Screening of the Methyl-CpG Binding-Protein 2 (*MECP2*) Gene in Non-Syndromic Mental Retardation (MRX). University of Sydney College of Health Sciences Conference, Leura, September 2002. (miniposter)

130.Pelka GJ, Hayward M, Gad J, Watson C, Christodoulou J, Tam PPL. Development of Mouse Models for Rett Syndrome. University of Sydney College of Health Sciences Conference, Leura, September 2002. (miniposter)

131.Gibson J, Bennetts B, Slobedman B, Armati P, Christodoulou J. Gene Expression in Rett Syndrome. University of Sydney College of Health Sciences Conference, Leura, September 2002. (miniposter)

132.Minchenko J, Williams A, Biggin A, Thorburn D, Trounce I, Christodoulou. A Family with Mitochondrial Respiratory Chain Defects: X-linked or Mitochondrial? University of Sydney College of Health Sciences Conference, Leura, September 2002. (miniposter)

133.Weaving LS, Lahooti H, Williamson S, Bennetts B, Christodoulou J. MeCP2 Function in Rett Syndrome (RS) Compared to Normal Controls. University of Sydney College of Health Sciences Conference, Leura, September 2002. (miniposter)

134.Christodoulou J, Grimm A, Maher T, Bennetts B. RettBASE: the IRSA MECP2 Variation Database. A New Mutation Database Offering Enhanced Capabilities. Human Genome Variation Society Conference, Baltimore, US, October 2002. (oral)

135.Christodoulou J, Biggin A, Thorburn D, Bennetts B. Mutation Screening of the Mitochondrial Genome Using Denaturing High-Performance Liquid Chromatography. American Society of Human Genetics Congress, Baltimore, US, October 2002. (poster)

136.Pelka G, Watson C, Christodoulou J, Tam PPL. Development of Mouse Models of Rett Syndrome. ComBio Conference, Sydney, October 2002. (poster)

137.Weaving LS, Lahooti H, Bennetts B, Christodoulou J. Functional Properties of Methyl CpG Binding Prtien 2 (MeCP2) in Rett Syndrome. ComBio Conference, Sydney, October 2002. (poster)

2003:

138.Singh-Grewal D, Williamson S, Chaitow J, Roscioli T, Bennetts B, Oxley P, Mutton P, Christodoulou J. Familial Mediterranean Fever and a Complex Allele (E148Q/I692del): Association with Sensorineural Deafness. American College of Rheumatology Pediatrics Conference, Denver, USA, March 2003. (poster)

139.Webster B, Lammi A, Curtin J, Wilcken B, Christodoulou J. Persistent Hyperhomocysteinaemia in a Boy with Cobalamin E Defect. 3rd Congress of Hyperhomocysteinemia, Saarbrücken, Germany, April 2003. (poster)

140.Moore H, de Klerk N, Robertson I. Fyfe S, Christodoulou J, Weaving L, Williamson S, Leonard S, Colvin L, Leonard H. Health Service Utilisation in Rett Syndrome. RACP Conference, Hobart, Tasmania, May 2003. (poster)

141.Pelka G, Watson CM, Loebel DAF, Christodoulou J, Tam PPL. Quantitation of Alternatively Polyadenylated MECP2 mRNA Transcripts. International Congress of Genetics, Melbourne, July 2003. (poster)

142.Watson CM, Pelka G, Radziewic T, Shahbazian MD, Zoghbi H, Christodoulou J. Tam PPL. X-inactivation patterns in *Mecp2* Mutant Mice. International Congress of Genetics, Melbourne, July 2003. (poster)

143.Gibson J, Williamson S, Arbuckle S, Christodoulou J. Regional Variation of X Chromosome Inactivation Patterns of the Brain in Rett Syndrome: Implications for the Disease Phenotype. Rett Syndrome Research Foundation Conference, Baltimore, June 2003. (oral)

144.Pelka GJ, Watson CM, Loebel DAF, Christodoulou J, Tam PPL. Analysis of *Mecp2* Expression by Real Time PCR and *in situ* Hybridisation. Rett Syndrome Research Foundation Conference, Baltimore, June 2003. (poster)

145.Gibson JH, Bennetts B, Slobedman B, Christodoulou J. Downstream Effects of *MECP2* Mutations in the Frontal Cortex of Rett Syndrome Patient Brains. 3rd Australian Microarray Conference, Couran Cove, July 2003 (poster)

146.Gibson JH, Bennetts B, Slobedman B, Christodoulou J. Downstream Effects of *MECP2* Mutations in the Frontal Cortex of Rett Syndrome Patient Brains. Westmead Hospital Association Hospital Week Research Symposium, Sydney, August 2003 (poster)

147.Watson CM, Pelka G, Radziewic T, Shahbazian MD, Zoghbi H, Christodoulou J. Tam PPL. X-inactivation patterns in *Mecp2* Mutant Mice. Westmead Hospital Association Hospital Week Research Symposium, Sydney, August 2003 (poster)

148.Earl JW, Coakley JC, Ouvrier RA, Antony J, Grattan-Smith, P, Christodoulou J. Screening for Neurotransmitter Disorders in Australia. 9th International Congress of Inborn Errors of Metabolism, Brisbane, September 2003. (poster)

149.Earl JW, Nath C, Alexander I, Wilcken B, Christodoulou J, Cheung NW, Lewis B, Boneh A, McGill J, Fletcher J, Wilson C. Biogenic Amine Disorders in Australia and New Zealand. 9th International Congress of Inborn Errors of Metabolism, Brisbane, September 2003. (poster)

150.Allen JR, Arrowsmith FE, Gruca M, Christodoulou J, Gaskin KJ. Nutritional evaluation in inborn errors of protein metabolism. Urea Cycle Satellite Conference, Sydney, September 2003. (oral)

151.Moore H, de Klerk N, Robertson I. Fyfe S, Christodoulou J, Weaving L, Williamson S, Leonard S, Colvin L, Leonard H. Health Service Utilisation in Rett Syndrome. Australasian Epidemiological Society Conference, Perth, September 2003. (poster)

152.Christodoulou J, Minchenko J, Thorburn D, Cooper S. Forced Myogenesis of Fibroblasts Can Unmask Mitochondrial Respiratory Chain Defects. American Society of Human Genetics Congress, Los Angeles, November 2003. (oral)

2004:

153.Malandris M, Bennetts B, Wilson MJ, Widmer RP, Cameron AC, Christodoulou J. A Clinical and Molecular Genetic Study of Oligodontia in Three Australian Families. Ectodermal Dysplasia Conference, University College London, England, May 2004 (poster)

154.Weaving LS, Christodoulou J, Williamson SL, Friend KL, McKenzie OLD, Archer H, Evans J, Clarke A, Pelka GP, Tam PPL, Watson CM, Lahooti H, Ellaway CJ, Bennetts B, Leonard H, Gécz J. Identification of a Second Rett Syndrome Gene: Mutation of *STK9* in Rett Syndrome, Autism and Neonatal Encephalopathy. Rett Syndrome Research Foundation Conference, Baltmore, June 2004. (oral)

155.Stormon MO, Barclay AR, Christodoulou J, Shun A, Sholler G, Arbuckle S, Dorney S. Primary Pulmonary Hypertension in Association with Mitochondrial Liver Disease. Presented at the 2nd World Congress of Paediatric Gastroenterology, Hepatology and Nutrition, Paris, July 2004. (poster)

156.Weaving LS, Christodoulou J, Williamson SL, Friend KL, McKenzie OLD, Archer H, Evans J, Clarke A, Pelka GP, Tam PPL, Watson CM, Lahooti H, Ellaway CJ, Bennetts B, Leonard H, Gécz J. Identification of a Second Rett Syndrome Gene: Mutation of *STK9* in Rett Syndrome, Autism and Neonatal Encephalopathy. 28th Annual Scientific Meeting of the Human Genetics Society of Australasia, Fremantle, August 2004. (oral)

157.Neas K, Bennetts B, Carpenter K, White R, Kirk EP, Wilson M, Kelley R, Baric I, Christodoulou J. *OPA3* mutation screening in patients with unexplained 3-methylglutaconic aciduria. 28th Annual Scientific Meeting of the Human Genetics Society of Australasia, Fremantle, August 2004. (oral)

158.Bezler A, Lahooti H, Christodoulou J. Functional Promoter Mutations of the *MECP2* Gene: Possible Significance in Rett Syndrome. 28th Annual Scientific Meeting of the Human Genetics Society of Australasia, Fremantle, August 2004. (oral)

159.Evans JC, Archer HL, Gecz J, Christodoulou J, Butler R, Whatley SD, Clarke A. Novel STK9 Mutation in a Patient with Rett-like Syndrome. British Society of Human Genetics, where, September 2004 (poster)

160.Pelka GJ, Radziewic T, Hayward M, Christodoulou J, Tam PL. Analysis of *MECP2* Deficiency on Cell Totipotency by *LacZ* Reporter Expression. Combio Conference, Perth, September 2004. (poster)

161.Christodoulou J, Weaving LS, Williamson SL, Friend KL, McKenzie OLD, Archer H, Evans J, Clarke A, Pelka GP, Tam PPL, Watson CM, Lahooti H, Ellaway CJ, Bennetts B, Leonard H, Gécz J. Mutations of *STK9* Cause Early Onset Seizures, Mental Retardation (MR), an Autistic Disorder and a Rett Syndrome (RTT)-like Phenotype. American Society of Human Genetics Congress, Toronto, October 2004. (oral)

162.Gibson JH, Stern J, Slobedman B, Christodoulou J. Downstream Effects of *MECP2* Mutations in the Frontal Cortex of Patients with Rett Syndrome. American Society of Human Genetics Congress, Toronto, October 2004. (oral)

163.Pelka GJ, Watson C, Christodoulou J, Tam PPL. Mecp2 Transcripts Exhibit Dynamic Changes Throughout Life. Fourth Research Conference: From Cell to Society 4, College of Health Sciences, University of Sydney, Leura, November 2004. (miniposter)

164.Christodoulou J, GrimmA, Maher T, Bennetts B. RettBASE: a Web-based *MECP2* Database. Fourth Research Conference: From Cell to Society 4, College of Health Sciences, University of Sydney, Leura, November 2004. (miniposter)

2005:

165.Mitchell JJ, Wilcken B, Alexander I, Ellaway C, O'Grady H, Wiley V, Earl J, Christodoulou C. Tetrahydrobiopterin Responsive Phenylketonuria: The New South Wales Experience. International Conference on Phenylketonuria and Tetrahydrobiopterin, Carnival Imagination, Miami FL, March 2005. (oral)

166.Thorburn D, Davidzon G, Mancuso M, Kirby K, Peters HL, Christodoulou J, Ferraris S, Pancrudo J, Quinzii C, Hirano M, DiMauro S. Mitochondrial DNA Depletion in Hepatocerebral Syndromes. 29th Annual Scientific Meeting of the Human Genetics Society of Australasia, Newcastle, July 2005. (oral/poster?)

167.Marriott J, Mary M-A, Thompson S, Christodoulou J. Case Study: Breast Fed Infant with Classic MSUD. 29th Annual Scientific Meeting of the Human Genetics Society of Australasia, Newcastle, July 2005. (oral/poster?)

168.Chiong M-A, Marinaki T, Duley J, Bennetts B, Ouvrier R, Christodoulou J. Lesch Nyhan Disease Presenting as "Cerebral Palsy" after General Anaesthesia : A Case Report. 29th Annual Scientific Meeting of the Human Genetics Society of Australasia, Newcastle, July 2005. (oral/poster?)

169.Sugiana C, Salemi R, Kirby DM, Bell K, Dahl HHM, Ohtake A, Ryan MT, Christodoulou J, Thorburn DR. Hunting for the First Mitochondrial Complex I Assembly gene Defect. 29th Annual Scientific Meeting of the Human Genetics Society of Australasia, Newcastle, July 2005. (oral/poster?)

170.Thompson S, Ghosh-Jerath S, Christodoulou J. Essential Fatty Acid Status in PKU. 29th Annual Scientific Meeting of the Human Genetics Society of Australasia, Newcastle, July 2005. (oral/poster?)

171.Thompson S, Ghosh-Jerath S, Schindler S, Joy P, Rocca A, Rae C, Kemp A, Christodoulou J. The Effect of Large Neutral Amino Acids on Brain Phenylalanine (Phe) Level and Brain Functioning in Early Treated Patients with Phenylketonuria (PKU). 29th Annual Scientific Meeting of the Human Genetics Society of Australasia, Newcastle, July 2005. (oral/poster?)

172.Gibson J, Minchenko J, Stern J, Slobedman B, Christodoulou J. Abnormal Expression of a Subset of Genes in the Frontal Cortex of Rett Syndrome (RTT) Patient Brains. 29th Annual Scientific Meeting of the Human Genetics Society of Australasia, Newcastle, July 2005. (oral – plenary session)

173.Pelka GJ, Hayward M, Radziewic T, Christodoulou J, Tam P.PL. Insights into Rett Syndrome Pathophysiology through Mouse Models. 6th Rett Syndrome Research Foundation Conference, Chicago, June 2005. (oral)

174.Gibson J, Minchenko J, Stern J, Slobedman B, Christodoulou J. Abnormal Expression of a Subset of Genes in the Frontal Cortex of Rett Syndrome (RTT) Patient Brains. 6th Rett Syndrome Research Foundation Conference, Chicago, June 2005. (poster)

175.Giak Sim K, Carpenter K, Kirk E, Christodoulou J, Wilcken B. Riboflavin Deficiency Presenting as a Functional Defect of Multiple Acyl-CoA Dehydrogenase Defect. SSIEM Conference, Paris, September 2005. (poster)

176.Chan PA, Duraisamy S, Murphy JA, McBride C, Bond JP, Raevaara T, Nystrom M, Grimm A, Christodoulou J, Oetting WS, Greenblat MS. Predicting Benign Versus Pathogenic Genetic Variants: Validation of Four Methods of Comparative Sequence Analysis in Human Disease Genes *CDKN2A, MLH1, MECP2* and Tyrosinase. Human Genome Variation Society Annual Scientific Conference, Salt Lake City, October, 2005. (oral)

2006:

177.Pelka GJ, Hayward M, Radziewic T, Christodoulou, J, Tam, PPL. Investigation of the impact of regionalized *Mecp2* deficiency and the manifestation of the RTT phenotype using chimera analysis. 7th Rett Syndrome Research Foundation Conference, Chicago, USA, June 2006 (oral)

178.White R, Bennetts B, Christodoulou J. Extended Mutation Screening in Rett Syndrome and Related Clinical Disorders. International Congress of Human Genetics, Brisbane, August, 2006 (poster)

179.Saxena A, Leonard H, de Lagarde D, Krueger R, Christodoulou C, Ravine R. Analysis of MeCP2 protein expression in peripheral blood cells of RTT patients with common recurrent mutations. International Congress of Human Genetics, Brisbane, August, 2006 (poster)

180.Vasudevan VL, Lahooti H, Christodoulou J. Role of CDKL5/STK9 in Rett Syndrome. Discipline of Paediatrics and Child Health Postgraduate Student Conference, Sydney, August, 2006 (oral)

181.Perry M, Christodoulou J, Bennetts B, Street N. Mutation Screening of the Ryanodine Receptor Gene in Malignant Hyperthermia. Discipline of Paediatrics and Child Health Postgraduate Student Conference, Sydney, August, 2006 (oral)

182.Williamson S, Lahooti H, Christodoulou J. Methyl CpG Binding Protein 2 (MeCP2), Cyclin Dependent Like Kinase 5 (CDKL5) and Rett Syndrome. Discipline of Paediatrics and Child Health Postgraduate Student Conference, Sydney, August, 2006 (oral)

183.White, R, Bennetts, B, Christodoulou, J. *CDKL5* Mutation Screening in Rett Syndrome and Related Clinical Disorders. Discipline of Paediatrics and Child Health Postgraduate Student Conference, Sydney, August, 2006 (oral)

184.Christodoulou J, Sim KG, Rhead WJ, Carpenter K. Clinical & biochemical features of glutaric aciduria type II in the infant of a mother with riboflavin deficiency. International Congress of Inborn Errors of Metabolism, Chiba, Japan, September, 2006 (poster)

185.Fletcher JM, Ketteridge DB, Simpson K, Thompson S, Kirby C, Christodoulou J. Successful pregnancies after low phenylalanine levels improved spermatogenesis in 2 men with PKU. International Congress of Inborn Errors of Metabolism, Chiba, Japan, September, 2006 (oral)

186.Christodoulou J, Ho G, Alexander I, Mitchell J, Ellaway C, Bratkovic D, Wilcken B. Molecular Analysis of the PAH Gene in Phenylketonuria (PKU): Correlation with BH4

Responsiveness. International Congress of Inborn Errors of Metabolism, Chiba, Japan, September, 2006 (oral)

187.Inwood AC, Gangemi R, O'Grady H, Fletcher J, Ketteridge D, Kirk E, Sillence D, Christodoulou J, Coman D, McGill JJ. Review of clinical course for 9 Australian children who have MPS 1 treated with bone marrow transplantation. Congress of Inborn Errors of Metabolism, Chiba, Japan, September, 2006 (poster)

188.Thompson S, Schindeler S, Ghosh-Jerath S, Rocca A, Joy P, Kemp A, Rae C, Green K, Chin C, Worthington R, Kim J, Wilcken B, Christodoulou. Effects of large neutral amino acid supplements in PKU: an MRS and neurospyschological study. Congress of Inborn Errors of Metabolism, Chiba, Japan, September, 2006 (oral)

189.Ghosh-Jerath S, Thompson S, Schindeler S, Christodoulou J. Essential fatty acid status in phenylketonuria (PKU). Indo-British Joint Conference of Pediatric Gastroenterology, Hepatology and Nutrition, Dehli, India, October, 2006 (poster)

190.Williamson S, Lahooti HS, Vasudevan V, Bezler A, Cloosterman D, Curphy L, Ho G, Reuter K, Mohamedali A, White R, Christodoulou J. Methyl CpG Binding Protein 2 (MECP2) Mutation Screening of a Rett Syndrome and Non-syndromic Mental Retardation Cohort. Medical Sciences Research conference, Leura, Sydney, November, 2006. (miniposter)

191.Grimm AJ, Chan PA, Duraisamy S, Bond JP, Greenblatt M, Newell JA, Christodoulou J. Predicting pathogenicity of *MECP2* mutations using the RettBASE mutation database. Medical Sciences Research conference, Leura, Sydney, November, 2006. (miniposter)

192.Vasudevan VL, Lahooti H, Christodoulou J. Role of *CDKL5* in Rett syndrome. Medical Sciences Research conference, Leura, Sydney, November, 2006. (miniposter)

2007:

193.Christodoulou J. Large Neutral Amino Acids – current opinion and use in PKU. SHS International Dietary Management of Inborn Errors of Metabolism Meeting, London, March 2007. (invited plenary lecture)

194.Thompson S, Schindler S, Ghosh-Jerath S. Phenylalanine and essential fatty acid intake and status in Australian teenagers and adults with phenylketonuria. SHS International Dietary Management of Inborn Errors of Metabolism Meeting, London, March 2007. (poster)

195.Kondo M, Pelka GJ, Christodoulou J, Tam PPL, Hannan AJ. Environmental enrichment ameliorates the behavioural phenotype in a *Mecp2*-null mouse model of Rett syndrome. International Brain Research Organisation conference, Melbourne, July 2007. (poster)

196.Williams SR, Edwards M, Hardwick SA, Slater K, Bennetts B, Christodoulou J, Smith RL. A familial *MECP2* gene rearrangement: variable phenotype and the role of X-inactivation in Rett syndrome. Royal Australasian College of Physicians conference, Melbourne, 2007. (poster)

197.Lahooti H, Williamson SL, Cloosterman D, Christodoulou J. Functional analysis of *MECP2* promoter sequence variations in Rett Syndrome (RTT). Rett Syndrome Research Foundation conference, Chicago, USA, June 2007. (poster)

198. Lahooti H, Williamson SL, Cloosterman D, Christodoulou J. Functional analysis of *MECP2* promoter sequence variations in Rett Syndrome (RTT). Human Genetics Society of Australasia conference, Auckland, New Zealand, August 2007. (oral presentation)

199. Williamson SL, Nelaturi V, Grimm A, Christodoulou J. A novel isoform of Cyclin Dependent Kinase Like 5 (CDKL5). Human Genetics Society of Australasia conference, Auckland, New Zealand, August 2007. (poster)

200. Ho G, B. Wilcken B, Alexander I, Ellaway C, Thompson S, Dennison B, Bratkovic D, Christodoulou J. Molecular analysis of the phenylalanine hydroxylase (*PAH*) gene in NSW: mutation profile and tetrahydrobiopterin (BH_4) responsiveness. Human Genetics Society of Australasia conference, Auckland, New Zealand, August 2007. (poster)

201. Carpenter K, Christodoulou J, Fietz M, Silink M. Congenital disorder of glycosylation type 1a (phosphomannomutase deficiency) presenting as isolated hyperinsulinaemic hypoglycaemia. Australasian Society for Inborn Errors of Metabolism conference, Auckland, New Zealand, August 2007. (poster)

202. Junek R, Dennison B, Thompson S, Watson P, Gick J, Wilcken B, Ellaway C, Alexander I, Christodoulou J. Dealing with PKU: a lifestyle not an illness. Australasian Society for Inborn Errors of Metabolism conference, Auckland, New Zealand, August 2007. (poster)

203. Bijarnia S, Wiley V, Carpenter K, Christodoulou J, Ellaway CJ, Wilcken B. Glutaric Aciduria type 1 in New South Wales in the last decade - Follow up and impact of newborn screening. Australasian Society for Inborn Errors of Metabolism conference, Auckland, New Zealand, August 2007. (oral presentation)

204. de Brouwer APM, Christodoulou J, Williams K, Nabuurs S, Hamel B, Duley J, Weaving L, White R, Donald J, van Bokhoven H. Loss of function mutations in the phosphoribosyl pyrophosphate synthase I *(PRPSI)* gene cause Arts syndrome. Human Genetics Society of Australasia conference, Auckland, New Zealand, August 2007. (oral presentation)

205. Dennison B, Thompson S, Ellaway C, Alexander I, Christodoulou J. Nutritional Management Concerns in 3 Phenylketonuria (PKU) Patients on BH4. Society for the Study of Inborn Errors of Metabolism conference, Hamburg, Germany, September 2007. (poster)

2008:

206. Ellen de Leon, Sandra Cooper, Jim Minchenco, David Thorburn and John Christodoulou. Unmasking Respiratory Chain Defects by Forced Myogenesis. United Mitochondrial Diseases Foundation Congress, Indianapolis, June 2008 (oral presentation).

207. Lahooti H, Williamson SL, Nelaturi V, Christodoulou J. TECHNOLOGICAL ADVANCENMENT IN STUDIES OF RETT SYNDROME (RTT): A LABORATORY PERSPECTIVE. Australian Institute of Medical Scientists conference, Albury, May 2008 (oral presentation).

208. Sugiana C, Pagliarini DJ, McKenzie M, Kirby DM, Salemi R, Hutchison WM, Vascotto KA, Smith SM, Lazarou M, Calvo S, Christodoulou J, Abu-Amero KK, Dahl HHM, Mootha VK, Ryan MT and Thorburn DR. Mutations in *C20orf7* cause Complex I deficiency and

Lethal Neonatal Mitochondrial Disease. Euromit conference, Stockholm, June 2008 (oral presentation).

209. Nelaturi VL, Williamson SL, Lahooti H, & Christodoulou J. Role of CDKL5 in Rett Syndrome. Human Genetics Society of Australasia Conference, Adelaide, July 2008 (oral presentation – winner of the best student presentation).

210. Horton Z., Thompson S., Newsom J., Cowell J., Kirkland R., Jay E., Lopes E., Bennetts B., Christodoulou J., Ellaway C. RETROSPECTIVE ANALYSIS OF PATIENTS ATTENDING AUSTRALIA'S FIRST RETT SYNDROME MULTIDISCIPLINARY MANAGEMENT CLINIC. Human Genetics Society of Australasia Conference, Adelaide, July 2008 (oral presentation).

211. Mohamedali, A., Pelka, G.J., Christodoulou, J., Tam, P.P.L. *MECP2* Deficiency Leads To Metabolic and Synaptic Changes in the Rett Brain. Discipline of Paediatrics and Child Health Annual Postgraduate Student Conference, Sydney, August 2008 (oral presentation).

212. Slater, K, Williamson, S, Bennetts, B, Lahooti, H, Christodoulou, J. Sodium phenylbutyrate, a therapeutic benefit in Rett syndrome? Discipline of Paediatrics and Child Health Annual Postgraduate Student Conference, Sydney, August 2008 (oral presentation).

213. Perry M., Christodoulou J., Bennetts B., Street N. Mutation Screening of the Ryanodine Receptor Gene in Malignant Hyperthermia. Discipline of Paediatrics and Child Health Annual Postgraduate Student Conference, Sydney, August 2008 (oral presentation).

214. Ho, G., Reichardt, J., Christodoulou, J. Phenylketonuria: Genetic, Functional and Structural Analyses of Phenylalanine Hydroxylase. Discipline of Paediatrics and Child Health Annual Postgraduate Student Conference, Sydney, August 2008 (oral presentation).

215. Armani R, Cloosterman D, Christodoulou J. The Use of High Resolution Melt in Detecting Sequence Variations in the Transcription Factor 4 (*TCF4*) Gene. Discipline of Paediatrics and Child Health Annual Postgraduate Student Conference, Sydney, August 2008 (oral presentation).

216. Vasudevan V, Lahooti H, Christodoulou J. Role of *CDKL5/STK9* in Rett syndrome. Discipline of Paediatrics and Child Health Annual Postgraduate Student Conference, Sydney, August 2008 (oral presentation).

217. De Greef E, Christodoulou J, Dorney S, Shun A, Verran D, O'Loughlin EV, Stormon MO. MITOCHONDRIAL RESPIRATORY CHAIN HEPATOPATHIES: ROLE OF LIVER TRANSPLANTATION. A CASE SERIES OF 5 PATIENTS. Australian Transplantation Congress, Sydney, August 2008 (poster presentation).

---

218. Relf B, Larkin E, de Torres C, Baur L, Christodoulou J, Waters K. Genomewide scan for obesity and obesity related traits in a single large Filipino family supports multifactorial disturbances in MES and OSA. European Congress of Obesity, 2008 (poster)

219. Cloosterman D, Williamson S.L., Pelka G.J., Tam P.P.J., and Christodoulou J. Detection of Mecp2 Gene Targets Using ChIP-on-Chip. 6th World Rett Syndrome Congress, Paris France, 10th – 13th October 2008 (oral presentation).

220. Ben Zeev B, Ho G, Bebbington A, Leonard H, de Klerk N, Eva Gak E, Vecksler M, John Christodoulou J. The Common BDNF Polymorphism May be a Modifier of Disease Severity in Rett Syndrome. 6th World Rett Syndrome Congress, Paris France, 10th – 13th October 2008 (oral presentation).

221. Horton Z., Thompson S., Newsom J., Cowell J., Kirkland R., Jay E., Lopes E., Bennetts B., Christodoulou J., Ellaway C. RETROSPECTIVE ANALYSIS OF PATIENTS ATTENDING AUSTRALIA'S FIRST RETT SYNDROME MULTIDISCIPLINARY MANAGEMENT CLINIC. 6th World Rett Syndrome Congress, Paris France, 10th – 13th October 2008 (oral presentation).

222. Mohamedali, A., Pelka, G.J., Christodoulou, J., Tam, P.P.L. *MECP2* Deficiency Leads To Metabolic and Synaptic Changes in the Rett Brain. COMBIO Conference, Canberra, October 2008 (oral presentation).

223. Ellen de Leon, Sandra Cooper, Jim Minchenco, David Thorburn and John Christodoulou. Unmasking Respiratory Chain Defects by Forced Myogenesis. AussieMit Conference, Melbourne, November 2008 (oral presentation).

224. Williamson S.L., Pelka G.J., Tam P.P.L., and Christodoulou J. Mitochondrial Function in the *Mecp2*^{tm1Pplt} Mouse. AussieMit Conference, Melbourne, November 2008 (poster presentation).

2009:

225. Christodoulou J. Genetic Testing for Rare Disorders: What Referring Labs Want. RCPA Pathology Update, Sydney, March 2009 (invited presentation).

226. Roy B, Bhattacharya K, Christodoulou J, Wiley V, Wilcken B. Outcome of Maple Syrup Urine Disease (MSUD) diagnosed by newborn screening. To be presented at the Paediatric Society of Australia and New Zealand congress, in April 2009 (oral presentation).

227. Riley L, Hickey P, Cooper S, Thorburn D, Compton A, Ryan M, de Leon E, Bahlo M, **Christodoulou J**. Autozygosity mapping reveals *YARS2* as a cause of MLASA: myopathy, lactic acidosis and sideroblastic anaemia. Human Genetics Society of Australasia Conference, Fremantle, May 2009 (platform presentation – conference highlights).

228. Mohamedali, A., Pelka, G.J., **Christodoulou, J**., Tam, P.P.L. *MECP2* Deficiency Leads To Metabolic and Synaptic Changes in the Rett Brain. 14th Lorne Proteomics Symposium, Lorne, March 2009 (oral presentation).

229. Ho G, Chiong M-A, Sim KG, Carpenter K, Rhead W, Olsen RKJ, Peters G, **Christodoulou J**. Riboflavin deficiency caused by deletion of the riboflavin transporter, *GPR172B*, detected

by quantitative real-time PCR (qRT-PCR). Human Genetics Society of Australasia Conference, Fremantle, May 2009 (platform presentation).

230. Perry M, **Christodoulou J**, Bennetts B, Street N. Mutation Screening of the Ryanodine Receptor Gene in Malignant Hyperthermia. Submitted to the European Malignant Hyperthermia Group conference, Melbourne, May 2009 (platform presentation).

231. Williamson SL, Nelaturi V, **Christodoulou J.** Novel *CDKL5* Isoform: Implications for Rett Syndrome and Related Disorders. International Rett Syndrome Foundation Conference, Washington, US, June 2009 (poster presentation).

232. Ho G, Inui K-I, Yonezawa A, Masuda S, Sim KG, Carpenter K, Mitchell JJ, Rhead W, Olsen RKJ, Peters G, **Christodoulou J.** Maternal riboflavin deficiency, mimicking neonatal-onset glutaric aciduria type II, is caused by deletion of the riboflavin transporter, *GPR172B*. ICIEM conference, San Diego, September 2009 (platform presentation).

233. Wilcken B, Haas M, Joy P, Wiley V, Bowling F, Carpenter K, **Christodoulou J**, Cowley D, Ellaway C, Fletcher J, Kirk EP, Lewis B, McGill J, Peters H, Pitt J, Ranieri E, Yaplito-Lee J, Boneh A. Newborn Screening by Tandem Mass Spectrometry: a Cohort Study Comparing Outcome in Screened and Clinically Diagnosed Patients at Six Years of Age. ICIEM conference, San Diego, September 2009 (poster presentation).

234. Georgiou Th, Ho G, Dionysiou M, Nicolaou A, Vogazianos M, Chappa G, Stylianidou G, **Christodoulou J**, Drousiotou A. The mutation spectrum of phenylalanine hydroxylase deficiency in Cyprus. ICIEM conference, San Diego, September 2009 (poster presentation).

235. Thompson S, Dennison B, Wiley V, Carpenter K, Wilcken B, Bhattacharya K, Alexander I, Ellaway C, **Christodoulou J**. Dietetic issues in the management of Medium Chain Acyl Coenzyme A Dehydrogenase deficiency diagnosed by Newborn Screening. ICIEM conference, San Diego, September 2009 (poster presentation).

236. Watson P, Dennison B, Junek Z, Garnett S, Thompson S, **Christodoulou J**. Children with non-Phenylketonuria-Hyperphenylalaninaemia: Management and Problems. ICIEM conference, San Diego, September 2009 (poster presentation).

237. Olsen RKJ, Fletcher J, Rhead WJ, Garn B, Kjeldsen M, Das AM, Pelken L, Olpin S, Bhattacharya K, Sim KG, **Christodoulou J**, Gregersen N, Andresen BS. Multiple Acyl-CoA dehydrogenation deficiency, detected by abnormal acylcarnitine profiles through MS/MS based newborn screening programmes, are not always associated with *ETF/ETFDH* mutations. ICIEM conference, San Diego, September 2009 (poster presentation).

238. Bhattacharya K, Wile V, Roy B, Ellaway C, Alexander, I, Dennison B, Thompson S, **Christodoulou J**, Carpenter K, Wilcken B. Newborn Screening for Maple Syrup Urine Disease (MSUD) by Tandem Mass Spectrometry: Outcome. ICIEM conference, San Diego, September 2009 (poster presentation).

239. Dalkeith T, Bhattacharya K, Holmes-Walker J, **Christodoulou J**, Wilcken B. Issues for the integrated care of children and adults with inborn errors of metabolism. ICIEM conference, San Diego, September 2009 (poster presentation).

240. **Christodoulou J**. Genetic and Epigenetic Factors Contributing to the Rett Syndrome Phenotype. European Rett Syndrome Working Group conference, Stresa, Italy, September 2009 (invited platform presentation).

241. **Christodoulou J**. New Developments in Rett Syndrome Research: a View from Down Under. Invited seminar presentation, University of Padua, Padua, Italy, September 2009.

242. Riley L, Hickey P, Cooper S, McKenzie M, Ryan M, Compton A, Thorburn D, Rudinger-Thirion J, de Leon E, Bahlo M, **Christodoulou J**. A *YARS2* mutation is a novel cause of mitochondrial myopathy characterized by lactic acidosis and sideroblastic anemia. ASHG conference, Hawaii, October 2009 (poster presentation).

243. Hynes K, Tarpey P, Dibbens LM, Bayly MA, Berkovic SF, Vandeleur J, Turner SJ, Brown NJ, D Desai T, Haan E, Turner G, Christodoulou J, Leonard H, Gill D, Afawi Z, Lerman-Sagie T, Lev, D, Stratton M, Scheffer IE, Gecz J. Epilepsy and mental retardation limited to females with *PCDH19* mutations can present *de novo* or in single generation families. ASHG conference, Hawaii, October 2009 (poster presentation).

244. Ho G, Inui K-I, Yonezawa A, Masuda S, Sim K.G., Carpenter K, Mitchell JJ, Rhead W, Olsen RKJ, Peters G, Christodoulou J. Maternal Riboflavin Deficiency, Mimicking Neonatal-Onset Glutaric Aciduria Type 2, Is Caused by Mutations in the Riboflavin Transporter GPR172B. Westmead Hospital Annual Conference, Westmead, August 2009 (poster presentation).

245. Bayles R, Kaipananickal H, Baker E, Ciccotosto J, Maxwell S, Cappai R, Pelka G, Tam P, **Christodoulou J**, Lambert G, El-Osta A. Transcriptional inhibition of the noradrenaline transporter gene Slc6a2. Australian Epigenetics conference. Melbourne, December, 2009 (poster presentation).

246. **Christodoulou J**. New Frontiers of Rett Syndrome Research. ASMR conference, Hobart, November 2009 (invited oral plenary presentation).

247. **Christodoulou J**. New Frontiers of Rett Syndrome Research: the Sydney Perspective Invited presentation, Brain Research Institute, University of Queensland, Brisbane November 2009 (invited lecture).

248. M. Kondo, L. J. Gray, G. J. Pelka, **J. Christodoulou**, P. P.L. Tam, A. J. Hannan. Effect of *Mecp2* mutation on BDNF expression and rescue of autistic spectrum disorder endophenotypes by environmental modulation in Rett syndrome mice. Australian Epigenetics conference. Melbourne, December, 2009 (poster presentation).

2010:

249. **Christodoulou J**. Evolution of the RCPA-HGSA Molecular Genetics Quality Assurance Program: Partnership in Action. RCPA Pathology Update, Melbourne, February 2010 (invited oral presentation).

250. Dalkeith T, Thompson S, Dennison B, **Christodoulou J**, Ellaway C, Wilcken B, Bhattacharya K. Difficulties in the dietetic management of patients with early childhood onset – multiple acyl Co-A dehydrogenase deficiency (MADD).

251. Williamson SL, Gold WA, Pelka GJ, Tam PPL, Gibson J, **Christodoulou J**. Mitochondrial abnormalities in the Mecp2 tm1Tam mouse model of Rett syndrome. United Mitocondrial Diseases Foundation Conference, Scottsdale, US, June 2009 (oral presentation).

252. Riley L, Cooper, S, Hickey P, Rudinger-Thiron J, McKenzie M, Compton A, Thorburn D, Ryan M, Giegé R, Bahlo M, **Christodoulou J**. A *YARS2* mutation is a novel cause of Mitochondrial Myopathy, Lactic Acidosis and Sideroblastic Anaemia (MLASA) syndrome. United Mitocondrial Diseases Foundation Conference, Scottsdale, US, June 2010 (oral presentation).

253. Williamson SL, Gold WA, Pelka GJ, Tam PPL, Gibson J, **Christodoulou J**. Mitochondrial abnormalities in the *Mecp2*^{tm1Tam} mouse model of Rett syndrome. International Rett Syndrome Foundation Conference, Washington, US, June 2009 (poster presentation).

254. Bhattacharya K, Ho G, Dalkeith T, Dennison B, Thompson S, **Christodoulou J**. HMG CoA Lyase paper. Submitted to Society for the Study of Inborn Errors of Metabolism conference, Istanbul, September 2010.

255. Ho G, Reichardt J, **Christodoulou J**. Effects of missense mutations in *PAH* on protein quaternary structure and enzyme function: implications for disease severity in phenylketonuria. Submitted to the OzBio conference, Melbourne, September 2010.

256. Gold WA, Williamson SL, Lahooti H, Pelka GJ, Tam PPL, **Christodoulou J**. Will pharmacological restoration of MeCP2 levels be of therapeutic value in Rett syndrome? Submitted to the Second European Rett Syndrome Conference, Edinburgh, Scotland, October 2010.

257. Huppke P, Brendel C, Korenke GC, Marquart I, Elpeleg O, Moller LB, **Christodoulou J**, Kaler S, Gartner J. Mutations in *CCS*, the copper chaperone for superoxide dismutase (SOD1) cause a novel disorder of human copper metabolism. Submitted to American Society of Human Genetics conference, Washington, US, October 2010.

258. Bettella E, Ho G, Murgia A, **Christodoulou J**. Non-coding elements that regulate MeCP2 expression. Human Genetics Society of Australasia conference, Melbourne, November 2010. (poster presentation)

259. Armani R, Yang N, Cloosterman, D. **Christodoulou, J.** No Novel Pathogenic *TCF4* and *MEF2C* Sequence Variations identified in RTT Patients. Human Genetics Society of Australasia conference, Melbourne, November 2010. (poster presentation)

260. Riley L, Cooper, S, Hickey P, Rudinger-Thiron J, McKenzie M, Compton A, Thorburn D, Ryan M, Giegé R, Bahlo M, **Christodoulou J**. A *YARS2* mutation is a novel cause of Mitochondrial Myopathy, Lactic Acidosis and Sideroblastic Anaemia (MLASA) syndrome. AussieMit conference, Sydney, December 2010 (poster presentation).

2011:

261. Duley JA, **Christodoulou J**, de Brouwer APM. The PRPP synthetase neuropathology spectrum: What can we learn from it? 14th International Symposium on Purine and Pyrimidine Metabolism in Man, Tokyo, Japan, February 2011(oral presentation given by Dr Duley).
262. Gold W, Ravine D, Saxena A, **Christodoulou J**. Therapeutic amelioration of functional abnormalities in RTT using HDAC inhibitors. Reproductive, Maternal and Child Health Research Theme day, Sydney University, June 2011. (Poster)
263. Alodaib A, Gold W, Wilson M, Collins F, Bennetts B, Sillence D, Bahlo M, **Christodoulou J**. Use of high throughput genomic screening technologies for gene discovery in Mendelian disorders. Reproductive, Maternal and Child Health Research Theme day, Sydney University, June 2011. (poster)
264. Kaur S, Gold WA, Pelka GJ, Tam PPL, **Christodoulou J**. Mitochondrial abnormalities in Rett syndrome with particular focus on the dysregulation of the respiratory chain in the *Mecp2*^{tm1Tam1} Rett mouse model. Reproductive, Maternal and Child Health Research Theme day, Sydney University, June 2011. (poster)
265. Menezes M, Riley L, Cooper S, Thorburn D, **Christodoulou J**. Transdifferentiation of patient fibroblasts for the study of tissue specific mitochondrial respiratory chain disorders. Reproductive, Maternal and Child Health Research Theme day, Sydney University, June 2011. (poster)
266. Riley LG, Cooper S, Christodoulou J. Investigating phenotypic variation and tissue specificity in a mitochondrial respiratory chain disorder. Euromit 8, European Meeting on Mitochondrial Pathology, Zaragoza, Spain, June, 2011 (poster)
267. Christodoulou J, Gold W, Ravine D. MeCP2, microtubule dynamics and HDAC6 inhibitors: is there a link? 12th Annual Rett Syndrome Symposium, Leesburg, Virginia, USA, June 2011 (invited oral presentation).
268. Watson P, Dennison B, Junek Z, Garnett S, Wiley V, Thompson S, Christodoulou J. Children with non phenylketonuria-hyperphenylalaninemia: management and problems. Human Genetics Society of Australasia/ASIEM Conference, Gold Coast, July 2011. (poster presentation)
269. Ellaway C, Collins F, Hackett A, McKenzie, F, Ho, G, Darmanian A, Peters G, Fagan K, Christodoulou J. 14q12 microdeletion: a Rett syndrome-like phenotype. Human Genetics Society of Australasia Conference, Gold Coast, July 2011. (oral presentation)
270. Kennedy C, Du Sart D, Bennetts B, Christodoulou, J. RCPA/HGSA Molecular Genetics Quality Assurance Program - moving forward. Human Genetics Society of Australasia Conference, Gold Coast, July 2011. (poster presentation)
271. Ormshaw E, Jenkins G, Brett M, Mihelec M, St Heaps L, Christodoulou J, Bennetts B. Establishment of a diagnostic service for Alagille syndrome. Human Genetics Society of Australasia Conference, Gold Coast, July 2011.
272. Tucker EJ, Hershman SG, Koehrer C, Belcher-Timme CA, Goldberger OA, **Christodoulou J**, Silberstein J, McKenzie M, Ryan MT, Compton AG, Calvo SE, Rajbhandary UL, Thorburn DR, MoothaVK. Mutations in *MTFMT* underlie a human disorder of formylation causing impaired mitochondrial translation. Human Genetics Society of Australasia Conference, Gold Coast, July 2011.
273. Thompson S, Dalkeith T, Bhattacharya K, Ellaway C, Christodoulou J. Simplifying the ketogenic diet – experience in pyruvate dehydrogenase

complex deficiency. Human Genetics Society of Australasia Conference, Gold Coast, July 2011.

274. Thompson S, Yip Q, Dennison D, Watson P, Alexander I, Bhattacharya K, Ellaway C, **Christodoulou J**. Nutritional assessment of patients with metabolic disorders. Human Genetics Society of Australasia Conference, Gold Coast, July 2011.

275. Brown RM, Kirby DM, Christodoulou J, Thorburn DR, Brown GK. Pyruvate dehydrogenase E3 binding protein deficiency associated with prolonged survival, dystonia and behavioural disturbances. SSIEM Conference, Geneva, Switzerland, August 2011.

276. Thompson S, Dalkeith T, Bhattacharya K, Ellaway C, Christodoulou J. Simplifying the ketogenic diet – experience in pyruvate dehydrogenase complex deficiency. SSIEM Conference, Geneva, Switzerland, August 2011.

277. Thompson S, Yip Q, Dennison D, Watson P, Alexander I, Bhattacharya K, Ellaway C, **Christodoulou J**. Nutritional assessment of patients with metabolic disorders. SSIEM Conference, Geneva, Switzerland, August 2011.

278. Menezes M, Riley L, Cooper S, Thorburn D, **Christodoulou J**. Transdifferentiation of patient fibroblasts for the study of tissue specific mitochondrial respiratory chain disorders. International Congress of Human Genetics Congress, Montreal, October 2011.

279. Compton AG, Calvo SE, Hershman SG, Lim SC, Garone C, Tucker EJ, Laskowski A, Lieber DS, Liu S, Christodoulou J, Fletcher JM, DiMauro S, Thorburn DR, Mootha V. Targeted next-generation DNA sequencing of the mitochondrial proteome. Menezes M, Riley L, Cooper S, Thorburn D, **Christodoulou J**. Transdifferentiation of patient fibroblasts for the study of tissue specific mitochondrial respiratory chain disorders. International Congress of Human Genetics Congress, Montreal, October 2011.

280. Kondo M, Gray LJ, Pelka GJ, **Christodoulou J**, Tam PPL, Hannan AJ. A study of environmental modulation and gene-dosage in a mouse model of Rett syndrome. Presented at the 12th Annual Rett Syndrome Symposium, Leesburg, VA, USA, 26th – 28th June (Poster).

281. Bebbington A, Leonard H, Jacoby P, **Christodoulou J**, Ben-Zeev B, Bahi-Buisson N, Pineda M, Anderson A, Ravine D, Ho G, Smeets E, Percy A. The phenotype of deletions in MECP2: what do we know? Presented at the 12th Annual Rett Syndrome Symposium, Leesburg, VA, USA, 26th – 28th June (Poster).

282. Ho G, Reichardt J, Christodoulou J. Nonsense read-through: a potential treatment for PKU. Presented at Discipline of Paediatrics and Child Health Postgraduate Research Conference, Sydney, August 2011 (oral presentation)

283. Alodaib A, Gold W, Wilson M, Collins F, Bennetts B, Sillence D, Bahlo M, Christodoulou J. The use of High Throughput Genomic Screening Technologies for Gene Discovery in Mendelian Disorders. Presented at Discipline of Paediatrics and Child Health Postgraduate Research Conference, Sydney, August 2011 (oral presentation)

284. Kaur S, Gold WA, Williamson SL, Gibson JH, Pelka GJ, Tam. PPL, Christodoulou J. Mitochondrial abnormalities in Rett syndrome with particular focus on the dysregulation of the respiratory chain in the *Mecp2*^{tm1Tam} Rett mouse model. Presented at Discipline of Paediatrics and Child Health Postgraduate Research Conference, Sydney, August 2011 (oral presentation)

285. AL Hafid N, XingZhang T, Carpenter K, Alexander I, Christodoulou J. Evaluation of Novel Treatment Strategies for Phenylketonuria (PKU). Presented at Discipline of Paediatrics and Child Health Postgraduate Research Conference, Sydney, August 2011 (oral presentation)

286. Menezes M, Riley L, Cooper S, Thorburn D, Christodoulou J. Transdifferentiation of Patient Fibroblasts for the Study of Tissue Specific Mitochondrial Respiratory Chain (RC) Disorders. Presented at Discipline of Paediatrics and Child Health Postgraduate Research Conference, Sydney, August 2011 (oral presentation)
287. Middleton A, Munns C, Selvadurai H, Christodoulou J. Whole body vibration training in chronic disease: muscle, bone, function. Presented at Discipline of Paediatrics and Child Health Postgraduate Research Conference, Sydney, August 2011 (oral presentation)
288. Fisk K, Williamson S, Bennetts B, Christodoulou J. Modulators of MECP2 expression are of therapeutic importance in Rett syndrome (RTT). Presented at Discipline of Paediatrics and Child Health Postgraduate Research Conference, Sydney, August 2011 (oral presentation)

2012:

289. Ellaway C Ho, G, Collins F, Hackett A, McKenzie, F, Ho, G, Peters G, Christodoulou J. 14q12 microdeletion and a Rett syndrome-like phenotype. Human Genome Conference, Sydney, March 2012. (poster presentation)
290. Riley JG, Cooper S, John Christodoulou J. Investigating phenotypic variation and tissue specificity in a mitochondrial respiratory chain disorder. Human Genome Conference, Sydney, March 2012. (poster presentation)
291. Alodaib A, Gold W, Wilson M, Collins F, Bennetts B, Sillence D, Bahlo M, Christodoulou J. Use of Next Generation Sequencing for Gene Discovery in Mendelian disorders. Human Genome Conference, Sydney, March 2012. (poster presentation)
292. Kaur S, Gold WA, Williamson SL, Gibson JH, Pelka GJ, Tam PPL, Christodoulou, J. Cardiolipin synthase – A novel basis for the mitochondrial respiratory chain defects in the *Mecp2*^{tm1Tam} Rett mouse model? Human Genome Conference, Sydney, March 2012. (poster presentation)
293. Menezes M, Riley L, Cooper S, Thorburn D, Christodoulou J. Can MyoD-mediated myogenesis help unmask muscle specific mitochondrial respiratory chain disorders? Human Genome Conference, Sydney, March 2012. (poster presentation)
294. Gold WA, Saxena S, Ravine D, and Christodoulou J. HDAC6 inhibitors ameliorate functional abnormalities in RTT. Human Genome Conference, Sydney, March 2012. (poster presentation)
295. Ho G, Reichardt J, Christodoulou J. Nonsense read-through: a potential treatment for phenylketonuria. Human Genome Conference, Sydney, March 2012. (poster presentation)
296. Huppke PM, Brendel C, Kahlscheuer V, Korenke G, Marquardt I, Freisinger P, Christodoulou J, Hillebrand M, Pitelet G, Wilson C, Gruber-Sedlmayr U, Ullmann R, Haas S, Elpeleg O, Nürnberg G, Nürnberg P, Dad S, Møller L, Kaler S, Gärtner J. Mutations in *SLC33A1* cause an autosomal recessive lethal disorder with congenital cataracts, bilateral hearing loss and reduced copper and ceruloplasmin levels in serum. German Pediatric Society conference, XXX 2012 (submitted)
297. Christodoulou J, Gold WA, Williamson SL, Kaur S, Gibson JH, Pelka GJ, Hargreaves IP, Land JM, Tam PPL. Mitochondrial Defects In Rett Syndrome. UMDF Conference, Bethedsa, USA, June 2012 (poster presentation)
298. Christodoulou J, Gold WA, Williamson SL, Kaur S, Gibson JH, Pelka GJ, Hargreaves IP, Land JM, Tam PPL. Mitochondrial Defects In Rett Syndrome. World Rett Congress, New Orleans, USA, June 2012 (poster presentation)

299. Christodoulou J, Williamson SL, Ellaway CJ, Pelka GJ, Tam PPL. Deletion of a Novel Phosphatase results in a Rett Syndrome-like Phenotype. World Rett Congress, New Orleans, USA, June 2012 (poster presentation)
300. Leonard H, Downs J, Bebbington A, Ellaway E, Anderson A, Bathgate K, Wong K, Torode T, Jacoby P, Christodoulou J. Why we need population-based and longitudinal study designs: the example of the Australian Rett Syndrome Study. World Rett Congress, New Orleans, USA, June 2012 (oral presentation)
301. Christodoulou J, Gold WA, Williamson SL, Kaur S, Gibson JH, Pelka GJ, Hargreaves IP, Land JM, Tam PPL. Mitochondrial Defects In Rett Syndrome. World Rett Congress, New Orleans, USA, June 2012 (poster presentation)
302. Fehr S, Wilson M, Christodoulou J, Downs J, Murgia A, Sartori S, Vecchi M, Ho G, Polli R, Psoni S, Bao X, de Klerk N, Leonard H. The CDKL5 disorder: a new cause of early-onset encephalopathy. World Rett Congress, New Orleans, USA, June 2012 (oral presentation)
303. Ho G, Reichardt J, Christodoulou, J. Complementation between specific phenylalanine hydroxylase (*PAH*) missense mutations and correlation to clinical phenotype in phenylketonuria (PKU). Human Genetics Society of Australasia Conference, Canberra, July 2012 (oral presentation)
304. Gold WA, A. Saxena A, Ravine D, Christodoulou J. HDAC6 inhibitors: a novel therapy for Rett syndrome. Human Genetics Society of Australasia Conference, Canberra, July 2012 (poster)
305. Menezes M, Riley L, Cooper S, Lukic V, Bahlo M, Thorburn DR, J. Christodoulou J. The whole exome sequencing approach to identify novel genes in mitochondrial respiratory chain disorders. Human Genetics Society of Australasia Conference, Canberra, July 2012 (oral presentation)
306. Williamson SL, Ellaway C, Pelka GJ, Tam PPL, Christodoulou J. Deletion of a novel phosphatase results in a Rett syndrome-like phenotype. Human Genetics Society of Australasia Conference, Canberra, July 2012 (oral presentation)
307. Al Hafid N, Carpenter K, Tong X-Z, Cunningham S, Alexander I, J. Christodoulou J. Towards the development of a genetically modified probiotic as a novel therapy for phenylketonuria (oral presentation)
308. Alodaib A, Gold W, Lek M, Collins F, Wilson M, Bennetts B, Sillence D, Scerri T, Bahlo M, Christodoulou J. Whole exome sequencing identifies a novel missense mutation in two affected children with a suspected rare Mendelian disorder. Human Genetics Society of Australasia Conference, Canberra, July 2012 (oral presentation)
309. Williams S, Leo A, Jenkins G, Bennetts B, Christodoulou J, Fietz M. Niemann-Pick disease type C caused by maternal uniparental isodisomy of chromosome 18. Human Genetics Society of Australasia Conference, Canberra, July 2012 (oral presentation)
310. Gold WA, Williamson SL, Kaur S, Gibson JH, Pelka GJ, Hargreaves IP, Land JM, Tam PPL, Christodoulou J. Mitochondrial defects in Rett syndrome. Human Genetics Society of Australasia Conference, Canberra, July 2012 (oral presentation)
311. Sim K, Devanapalli B, Mowat D, Fietz M, Christodoulou J, Carpenter K. Prenatal diagnosis analysing amniotic fluid amino acids failed to detect classical maple syrup urine disease. Human Genetics Society of Australasia Conference, Canberra, July 2012 (poster presentation)
312. Kennedy CT, Bennetts BH, Christodoulou J. 2011 molecular genetics quality assurance program review. Human Genetics Society of Australasia Conference, Canberra, July 2012 (oral presentation)
313. Huppke PM, Brendel C, Kahlscheuer V, Korenke G, Marquardt I, Freisinger P, Christodoulou J, Hillebrand M, Pitelet G, Wilson C, Gruber-Sedlmayr U, Ullmann R, Haas S, Elpeleg O, Nürnberg G, Nürnberg P, Dad S, Møller L, Kaler S, Gärtner J. Mutations in *SLC33A1* cause an autosomal recessive lethal disorder with congenital cataracts, bilateral

hearing loss and reduced copper and ceruloplasmin levels in serum. SSIEM conference, Birmingham, September 2012 (submitted)

314. Ho G, Bennetts B, Christodoulou J. RettBASE: Locus-specific mutation database for Rett syndrome and related disorders Australasian Mutation Detection & Molecular Genetics Society of Australasia conference, Port Douglas, September 2012 (submitted)

315. Menezes M, Riley LG, Cooper ST, Tzagoloff A, Mina K, Davis M, Allcock JN, Kresoje N, Laing NG, Mountford HS, Thorburn D, **Christodoulou, J**. Whole exome sequencing reveals a novel gene associated with complex III deficiency. AusMit conference, Melbourne, December, 2012 (submitted)

316. **Christodoulou J.** Clinical and Laboratory Aspects of Mitochondrial Disorders in Childood. AusieMit conference, Melbourne, December 2013 (invited oral presentation).

2013:

317. Williamson SL, Ellaway C, Pelka GJ, Tam PPL, **Christodoulou J**. Deletion of protein tyrosine phosphatase, non-receptor type 4 (PTPN4) results in a Rett Syndrome–Like Phenotype. Human Genome (HUGO) Meeting, Singapore, April 2013 (submitted).

318. **Christodoulou J.** Gene Discovery for Orphan Diseases: This is why you do it. Royal College of Pathologists conference, Melbourne, March 2013 (invited oral presentation).

319. **Christodoulou J.** Advances in the Genetic Investigation of SIDS. Royal College of Pathologists conference, Melbourne, March 2013 (invited oral presentation).

320. Ho G, Reichardt J, **Christodoulou, J**. Complementation between specific phenylalanine hydroxylase (*PAH*) missense mutations and correlation to clinical phenotype in phenylketonuria (PKU). Human Genome (HUGO) Meeting, Singapore, April 2013 (submitted).

321. Menezes MJ, Menezes MP, Riley L, Cooper S, **Christodoulou J**. The need for careful diagnosis of congenital myasthenic syndromes with secondary complex I disorders. United Mitochondrial Disorders Foundation conference, Newport Beach, Ca, USA, June, 2013.

322. Dennison B, Thompson S, **Christodoulou J**. How much protein can he tolerate? A study of phenylalanine tolerance in a boy with phenylketonuria. Human Genetics Society of Australasia conference, Queenstown, New Zealand, August, 2013.

323. Ho G, Ormshaw E, Bennetts B, Christodoulou J. Custom targeted resequencing methods in a clinical diagnostic setting: genetic testing for early infantile epileptic encephalopathies.

324. Alodaib A, Menezes M, Gold W, Sugano K, Yonezawa A, Lek M, Webster R, Carpenter K, Bennetts B, Ouvrier R, Christodoulou J. Whole exome sequencing identifies mutation in the riboflavin transporter gene *SLC52A2* in three affected children with Brown-Vialetto-van Laere syndrome. Human Genetics Society of Australasia conference, Queenstown, New Zealand, August, 2013.

325. Nafisinia M, Riley L, Gold, W Menezes M, Thorburn D, **Christodoulou J**. Revealing underlying mechanisms of mitochondrial respiratory chain disorders by gene discovery. Human Genetics Society of Australasia conference, Queenstown, New Zealand, August, 2013.

326. Menezes MJ, Menezes MP, Riley L, Cooper S, **Christodoulou J**. The need for careful diagnosis of congenital myasthenic syndromes with secondary complex I disorders. Human Genetics Society of Australasia conference, Queenstown, New Zealand, August, 2013.

327. R.J. Taft, C. Simons, A. Vanderve[2], R.J. Leventer, M.S. van der Knaap, N.I. Wol[5], R. Schiffmann, S. Damiani, P. Pearl, M. Bloom, S.M. Grimmond, D. Miller, D.R. Thorburn, **J. Christodoulou**, M. Gabbet, J. McGaughran. Application of high-throughput sequencing to

pediatric patients with unresolved diagnoses. Submitted to the American Society of Human Genetics Conference, Boston October, 2013

Invited Presentations:

Since 1993 over 200 lectures have been given to many groups, including undergraduate and graduate medical students, paediatric trainees, paediatricians, adult physicians, general practitioners, nursing staff, allied health staff, laboratory research groups, lawyers, and the lay public.

The breakdown from 1993 – 2002 is as follows:

Local Institutional Lectures	- at the Children's Hospital at Westmead	108
	- at other Hospitals	30
University of Sydney Lectures		18
Lectures at other Universities		6
Invited Presentations at National Conferences		11
Invited Presentations at International Conferences		20
Invited Presentations to Professional Organisations		16
Lectures to Lay Organisations		10

Year	Number of Lectures Given
1993	24
1994	23
1995	35
1996	26
1997	30
1998	13
1999	12
2000	20
2001	17
2002	19
Total	**219**

Invited Speaker Presentations:

2001:

The Human Genome Project: Opportunities, Challenges and Consequences for Population Screening. Plenary speaker at the 4th Asia Pacific Regional Meeting of the International Society for Neonatal Screening, Manila, The Philippines, October 2001.

A Clinical Approach to Inborn Errors of Metabolism Presenting in the Newborn. Invited speaker at the 4th Asia Pacific Regional Meeting of the International Society for Neonatal Screening, Manila, The Philippines, October 2001.

2002:

IRSA Rett Databases: International Phenotype Database and RettBASE: IRSA MECP2 Variation Database. Rett Syndrome Satellite Meeting, Beijing, September 2002.

Rett Syndrome and MECP2: Phenotype – Genotype Correlations. Rett Syndrome Satellite Meeting, Beijing, September 2002.

2003:

Clinical, Biochemical and Genetic Aspects of the Mitochondrial Respiratory Chain Defects. Presented at Pathology Update 2003 (Royal College of Pathologists of Australia), Darling Harbour, Sydney, March 2003.

Rett Syndrome and MECP2: Challenges in Searching for Phenotype-Genotype Links. Presented at the Rett Syndrome of UK Family Weekend, Northampton, UK, October 2003.

2004:

Mitochondrial Encephalomyopathies – Something for Everyone. Presented at Pathology Update 2004 (Royal College of Pathologists of Australia), Darling Harbour, Sydney, March 2004.

The Human Genome Project: Promises and Problems in Pediatric Practice. Plenary presentation at the 41st Annual Convention of the Philippines Pediatric Society, Manila, April 2004.

New Technologies for the Detection of Submicroscopic Chromosomal Rearrangements. Plenary presentation at the 41st Annual Convention of the Philippines Pediatric Society, Manila, April 2004.

Metabolic Catastrophies Masquerading as Sepsis. Plenary presentation at the 41st Annual Convention of the Philippines Pediatric Society, Manila, April 2004.

2005:

CDKL5: a second Rett syndrome gene? Implications for other neurodevelopmental disorders. University of Padua Scientific Program, Padua, Italy, May 2005

Rett syndrome, more than one gene, more than one story. Dept of Molecular Genetics, University of Padua, Padua, Italy, May 2005

2006:

Inherited causes of cardiomyopathy in childhood and beyond. Presented at the Cardiac Society of Australia and New Zealand Working Group conference on Cardiovascular Genetics, Sydney Convention Centre, Darling Harbour, Sydney, March 2006.

Practicalities of genetic investigation (who is doing what, where and why?). Presented at the Royal College of Pathologists of Australasia Pathology Update meeting, Sydney Convention Centre, Darling Harbour, Sydney, March 2006.

Clinical and laboratory aspects of mitochondrial diseases in childhood. Presented at the Royal College of Pathologists of Australasia Pathology Update meeting, Sydney Convention Centre, Darling Harbour, Sydney, March 2006.

Mitochondrial encephalomyopathies: something for everyone. Presented at the Cyprus Institute for Neurology and Genetics, Nicosia, Cyprus, July 2006.

Rett Syndrome: more than one gene, more than one story. Presented at the Genetics Society of Cyprus, Nicosia, Cyprus, July 2006.

The role of large neutral amino acids in PKU. Presented at the Mastering Metabolic Therapy conference, Las Vegas, October 2006.

Long chain polyunsaturated fatty acids: are they needed in PKU and other metabolic disorders? Presented at the Mastering Metabolic Therapy conference, Las Vegas, October 2006.

2007:

Large Neutral Amino Acids – current opinion and use in PKU.
Presented at the Nutricia conference, Dietary Management of Inherited Metabolic Disease, Royal College of Physicians, London, 16th March 2007

A Huge Leap Backwards, Then Small Steps Forward: Profile of a Rett Multidisciplinary Management Clinic, presented at the Rett Syndrome.
Presented at the Rett syndrome: Diagnosis, Genetics, Epidemiology and Clinical Management Workshop, Peking University First Hospital, Beijing, China, 7th – 13th September 2007

Rett Syndrome Research – What's New and Exciting.
Presented at the Rett Syndrome: Diagnosis, Genetics, Epidemiology and Clinical Management Workshop, Peking University First Hospital, Beijing, China, 7th – 13th September 2007

Clinical and Genetic Diagnosis of Rett Syndrome.
Presented at the Rett Syndrome: Diagnosis, Genetics, Epidemiology and Clinical Management Workshop, Peking University First Hospital, Beijing, China, 7th – 13th September 2007

2008:

Lessons from the Mouse: Rett Syndrome is Potentially Treatable.
Presented at the New Zealand Rett Congress, Wellington, New Zealand, 25th April 2008

Rett Syndrome Research: The Sydney Experience.
Presented at the New Zealand Rett Congress, Wellington, New Zealand, 25th April 2008

A Huge Step Backwards, then Small Steps Forward: Profile of a Rett Syndrome Multidisciplinary Management Clinic.
Presented at the New Zealand Rett Congress, Wellington, New Zealand, 25th April 2008

Expanded Newborn Screening: Is Asia Ready to Screen for 50 Disorders?
Presented at the HUGO-Pacific Congress, Cebu, Philippines, 6th April 2008

Stratifying RTT Patients for Clinical Trials: A Mutation Based Approach.
Presented at the IRSF Minisymposium on Clinical Trials in Rett Syndrome, Chicago, 22nd – 23rd June 2008

CDKL5 Deficiency: Overview of Clinical Molecular and Biological Aspects.
Presented at the 6th World Rett Syndrome Congress, Paris France, 10th – 13th October 2008

RettBASE: The IRSF MECP2 Variation Database.
Presented at the 6th World Rett Syndrome Congress, Paris France, 10th – 13th October 2008

Re-investigation of a 25 yr old with mental retardation and seizures.
Presented at the Epilepsy Society of Australia conference, Darling Harbour, 7th November 2008.

2009:

Genetic Testing for Rare Disorders: What Referring Labs Want.
Presented at the XXV WASPaLM Congress, Sydney, 13th – 15th March 2009.

Genetic and Epigenetic Factors Contributing to the Rett Syndrome Phenotype.
Presented at the European Rett Syndrome Working Group conference, Stresa, Italy, 18th – 20th September 2009 (invited platform presentation).

New Developments in Rett Syndrome Research: a View from Down Under.
Invited seminar presentation, University of Padua, Padua, Italy, 15th September 2009.

New Frontiers of Rett Syndrome Research: The Sydney Perspective.
Presented at the Queensland Brain Institute, University of Queensland, 5th November 2009.

New Frontiers of Rett Syndrome Research.
Presented at the ASMR conference, Hobart, 15th November 2009 (invited oral plenary presentation).

2010:

Evolution of the RCPA-HGSA Molecular Genetics Quality Assurance Program: Partnership in Action.
Presented at the Pathology Update, Melbourne, March 2010.

Rett Syndrome: New Clinical and Molecular Insights
Clinical and Laboratory Aspects of Mitochondrial Diseases in Childhood
Genetic Metabolic Disorders Causing Acute Encephalopathy
Inherited Metabolic Causes of Cardiomyopathy in Childhood & Beyond
Presented at the Malaysian Paediatric Congress, Kuala Lumpur, Malaysia, March 2010.

New Frontiers of Rett Syndrome Research – the Sydney Perspective
Presented at the Institute of Child Health, Great Ormond St Hospital, London, 5th October 2010.

PKU Research: The Latest from Sydney
Presented at the MDDA conference, Melbourne, 14th November 2010.

2011:

The Acidotic Neonate: what do we do?
Presented at the 6th World Congress of Paediatric Critical Care, 16th March 2011 (plenary).

MeCP2, microtubule dynamics and HDAC6 Inhibitors: is there a link?
Presented at the 12th Annual Rett Syndrome Symposium, Leesburg, VA, USA, 26th – 28th June (invited speaker).

New insights into the biology of Rett syndrome: implications for therapeutic interventions.
Presented at the PhD seminar series, University of Padova, Padova, Italy, 8th September (invited speaker).

The "Next Generation" Approach to Gene Discovery of the Mitochondrial Disorders
Presented at the PhD seminar series, University of Padova, , Padova, Italy, 7th October (invited speaker).

Genetic heterogeneity of Rett syndrome: multiple genes, multiple phenotypes.
Presented to the Veneto Region Child Neurologists symposium, Padova, Italy, 14th October (plenary speaker).

New insights into the biology of Rett syndrome: implications for therapeutic interventions.
Presented at the Genetics seminar series, University of Siena, Siena, Italy, 17th October (invited speaker).

The "Next Generation" Approach to Gene Discovery of the Mitochondrial Disorders
Presented at the Genetics seminar series, University of Siena, Siena, Italy, 19th October (invited speaker).

Clinical and Laboratory Aspects of Mitochondrial Diseases in Childhood.
Presentation to Metabolic Masters program, Padova, Italy, 20th October (invited speaker).

2012:

Epileptic encephalopathy in the first year of life: metabolic and other monogenic causes.
Presented at the Hospital Seminar series, DIPARTIMENTO A.I. DI PEDIATRIA "SALUS PUERI", Azienda Ospedaliera – Universita' Di Padova, Padova, Italy, 1st March (invited speaker).

The genetics and biology of Rett syndrome: are there potential avenues for therapy?
Presented at RTT Parent meeting, 26th May, Brisbane, Australia.

Clinical Evaluation and Emergency Management of Inborn Errors of Metabolism Presenting with Neonatal Encephalopathy.
Presented at the 12th International Child Neurology Congress, 27th May, Brisbane, Australia (invited speaker).

Genetic heterogeneity of Rett syndrome: multiple genes, multiple phenotypes.

Presented at the 12th International Child Neurology Congress, 27th May, Brisbane, Australia (invited speaker).

Latest Advances in Mitochondrial Therapies.
Presented at the 12th International Child Neurology Congress, 28th May, Brisbane, Australia (invited speaker).

Therapies for Rett Syndrome: Past and Current Clinical Trials, and Future Prospects.
Presented at the 7th World Rett Syndrome Congress, 22nd June, New Orleans, USA (invited speaker).

CDKL5: What we know so far/Genetics 101.
Presented at the inaugural International Foundation for CDKL5 Research conference, 24th June, New Orleans, USA (invited speaker).

Novel Clinical and molecular aspects relating to CDKL5 -Implications for genetic testing.
Presented at the International CDKL5 Research Symposium, 27th June, New Orleans, USA (invited speaker).

Metaxas-Kakkis Lecture 2012: Gene discovery for orphan genetic disorders: translation from the bench to the bedside. Presented at the Hippocration General Hospital, Thessaloniki, Greece, 18th September. (Plenary speaker)

Genetic Metabolic Disorders in Adults: not so rare but treatable. Presented at the Hippocration General Hospital, Thessaloniki, Greece, 19th September. (invited speaker)

Insights into the genetics and biology of Rett syndrome: implications for therapeutic interventions. Presented at the Hippocration General Hospital, Thessaloniki, Greece, 19th September. (invited speaker)

The Next Generation in Genetic Diagnosis. Presented at the University of Padua Dept of Paediatrics Seminar, Padua, Italy, 27th September 2012. (invited speaker)

2013:
Practical Utility of Next Generation Sequencing for Orphan Diseases. Presented at the RCPA Pathology Update, Melbourne, 22nd February (invited speaker).

Advances in the Genetic Investigation of Sudden Infant Death Syndrome. Presented at the RCPA Pathology Update, Melbourne, 24th February (invited speaker).

Epileptic encephalopathy in the first year of life: metabolic and other monogenic causes. Presented at the IEMCON conference, Delhi, India, 5th April (plenary speaker).

Clinical and Laboratory Aspects of Mitochondrial Disorders in Childhood. Presented at the IEMCON conference, Delhi, India, 6th April (plenary speaker).

Diagnosis and Management of Maple Syrup Urine Disease. Presented at the IEMCON conference, Delhi, India, 6th April (plenary speaker).

Treatment of Disorders of Purine and Pyrmidine Metabolism. To be presented at the ICIEM conference, Barcelona, September, 2013.

Other Presentations:

1. *Inborn errors of metabolism presenting in the newborn period.* Presented as part of the Neonatology Training Program at the Hospital for Sick Children, Toronto, 1990.

2. *A neurologist's guide to genetic metabolic diseases.* Presented as part of the training programme for Neurology residents in Toronto held at the Toronto Western Hospital, 1990.

3. *A case of D-lactic acidosis.* Division of Clinical Nutrition weekly seminar, Hospital for Sick Children, Toronto, 1990.

4. *Complex III deficiency.* Division of Clinical Genetics weekly seminar, Hospital for Sick Children, Toronto, 1991.

5. *A case of Isovaleric acidaemia.* Division of Clinical Genetics weekly seminar, Hospital for Sick Children, Toronto, 1991.

6. *Phenylketonuria- therapeutic successes but what about the survivors?* Continuing Education Seminar, Faculty of Medicine, University of Toronto, 1991.

7. *Four cases of Barth syndrome.* Division of Clinical Genetics weekly seminar, Hospital for Sick Children, Toronto, 1991.

8. *Genetic Metabolic Diseases- from the patient to the laboratory and back again.* Presented at the Cytogeneticists Scientific Meeting held at the Michener Institute for Applied Health Sciences, Toronto, 1991.

9. *Overview of Metabolic Disorders.* Presented to the FRCP candidates, Hospital for Sick Children, Toronto, 1991.

10. *Inherited Metabolic Diseases: When to suspect them and what to do about them.* Presented at Grand Rounds, Hospital for Sick Children, Toronto, 1991.

11. *Metabolic Emergencies: Investigation and management.* Presented to Resident Medical Officers, Hospital for Sick Children, Toronto, 1991.

12. *Pyridoxine Dependent Epilepsy - to GAD or not to GAD?* Presented at Genetic Rounds, Hospital for Sick Children, Toronto, 1991.

13. *MCAD Deficiency- What's old and what's new.* Presented at the GI Research Seminar, Hospital for Sick Children, Toronto, 1992.

14. *Molecular analysis of argininosuccinic acid lyase deficiency.* Presented by J. Christodoulou and H. Craig at the Dept of Genetics weekly research meeting, Hospital for Sick Children, Toronto, 1992.

15. *A case of neonatal sideroblastic anaemia and lactic. Is it Pearson syndrome?* Presented by Dr B. Webster and J. Christodoulou at Grand Rounds held at King George V Hospital, Sydney, 1992.

16. *Inherited Metabolic Disease in Childhood.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Camperdown, September 1992, March 1993, May 1993, August 1993, October 1993, March 1994, May 1994, August 1994, October 1994, February 1995.

17. *PCR and direct sequencing* . Presented at the Renal Research Meeting, The Children's Hospital, Camperdown, September, 1992.

18. *Inherited Metabolic Disease in the Newborn.* Presented to Clinical Genetics Fellows, The Children's Hospital, Camperdown, September, 1992.

19. *Approach to Inherited Metabolic Disease.* Presented to Intensive Care Unit Fellows, The Children's Hospital, Camperdown, September, 1992.

20. *When lightning strikes twice: cytochrome c oxidase deficiency.* Grand Rounds, The Children's Hospital, Camperdown, February, 1993.

21. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Camperdown, March 1993, May 1993, October 1993, March 1994, August (x 2)1994, October 1994, March 1995.

22. *Biochemical Genetics.* Presented as part of the Centre for Continuing Education Lecture Series, "Understanding the New Genetics", University of Sydney, March 1993.

23. *Treatment of Genetic Diseases.* Presented as part of the Centre for Continuing Education Lecture Series, "Understanding the New Genetics", University of Sydney, March 1993.

24. *Mitochondrial DNA and Genetics Diseases.* Presented at the Molecular Group Seminar Series, Royal Prince Alfred Hospital, Camperdown, April, 1993.

25. *Methods of Mutation Analysis.* Presented to Genetics Fellows as part of the Genetics training programme, The Children's Hospital, Camperdown, May 1993.

26. *Insights into Some Inborn Errors of Metabolism-Argininosuccininic Aciduria and Pyridoxine-dependent Epilpesy.* Presented at the Genetics Seminar, Adelaide Children's Hospital, May 1993.

27. *Genetic Metabolic Causes of Intellectual Handicap.* Presented at the Conference on Developmental Disability Medicine, Sheraton Sydney Airport Hotel, June 1993.

28. *Prenatal Diagnosis of Genetic Metabolic Disorders.* Presented at the Perinatal Dept Meeting, King George V Hospital, Camperdown, June 1993.

29. *Inborn Errors of Metabolism.* Presented to Master of Nutrition and Dietetics students from the University of Sydney, August, 1993.

30. *Single Strand Conformation Polymorphism Analysis.* Presented at the Molecular Group Seminar Series, Royal Prince Alfred Hospital, Camperdown, August, 1993.

31. *Genetic Metabolic Disorders: When to Think of Them and What to Do About Them.* Presented at the Diploma of Paediatrics Course, Children's Hospital, Camperdown, October 1993.

32. *Mitochondrial Inheritance and Diseases.* Presented at The Clinical Genetics Update for the Medical Practitioner Meeting, The Children's Hospital, Camperdown, October 1993.

33. *Mitochondrial Inheritance and Diseases.* Presented for the ICU Clinical Seminars series, The Children's Hospital, Camperdown, November 1993.

34. *Practical Approach to Inborn Errors of Metabolism/Newborn Screening.* Presented to FRACP candidates, The Children's Hospital, Camperdown, December 1993.

35. *Genetic Metabolic Disorders in the Newborn, and Practical Aspects of Newborn Screening.* Presented to Midwifery students, Gosford Hospital, Gosford, December 1993, May 1994, September 1994, December 1994, March 1995.

36. *Mitochondrial Inheritance and Diseases.* Presented to FRACP candidates, The Children's Hospital, Camperdown, December 1993.

37. *Defects of Neurotransmitter Metabolism: From the Bedside to the Bench (and Back Again).* Presented at the Paediatric Research Seminar, The Children's Hospital, Camperdown, December 1993.

38. *Scope of Human Genetics.* A 6 hour series of lectures given as part of the Graduate Diploma of Clinical Chemistry course run by the University of Western Sydney, March, 1994.

39. *Prenatal Diagnosis of Inborn Errors of Metabolism.* Presented at the Genetics Seminar Program of the Royal North Shore Hospital, Sydney, May, 1994.

40. *Genetics of Leigh Disease.* Presented to the inaugural Leigh's Disease Support Group Meeting, AGSA, Sydney, June 1994.

41. *Leigh Disease- What's in a Name?* Presented at Grand Rounds, Royal Alexandra Hospital for Children, Sydney, June, 1994.

42. *Metabolic Update - 1994: Genetic Metabolic Disorders: When To Think Of Them, And What To Do About Them.* Presented at the Paediatric Postgraduate Weekend for General Practitioners, Royal Alexandra Hospital for Children, Sydney, September, 1994.

43. *History Taking in Paediatrics.* Presented to 5th year medical students, Royal Alexandra Hospital for Children, Sydney, September, 1994.

44. *Inborn Errors of Metanolism.* Presented to Sydney University Master of Nutrition and Dietetics Students, Royal Alexandra Hospital for Children, Sydney, September, 1994.

45. *Update on Genetic Metabolic Diseases.* Presented at the Royal Australasian College of General Practitioners Annual Meeting, Canberra, September, 1994.

46. *Pyridoxine-dependent Epilepsy - Fact or Fiction?* Presented at the Annual Postgraduate Meeting, Royal Alexandra Hospital for Children, Sydney, October, 1994.

47. *Practical Approach to the Investigation of Genetic Metabolic Disorders.* Presented at the Paediatric Update Meeting, Royal Alexandra Hospital for Children, Sydney, October, 1994.

48. *Clinical and Molecular Aspects of Familial Hypertrophic Cardiomyopathy.* Presented at the Muscle Group Seminars, Dept of Anatomy and Histology, University of Sydney, October 1994.

49. *Clinical, Biochemical, and Molecular Aspects of Inborn Errors of Neurotransmitter Metabolism.* Presented at the Molecular Group Seminar Series, Royal Prince Alfred Hospital, Camperdown, October 1994.

50. *Practical Approach to the Investigation of Genetic Metabolic Disorders.* Presented to Diploma of Child Health candidates, Royal Alexandra Hospital for Children, Sydney, October, 1994.

51. *Inherited Neurodegenerative Disorders: Is There Light At The End Of The Tunnel?* Presented at the Association of Doctors in Developmental Disability Conference Day, Parkroyal Hotel, Parramatta, November, 1994.

52. *Sudden Collapse - Day 5.* Presented at the Dept of Fetal &Perinatal Medicine Monthly Clinical Meeting, King George Vth Hospital for Mothers and Children, Camperdown, November, 1994.

53. *Clinical, Biochemical, and Molecular Aspects of Inborn Errors of Neurotransmitter Metabolism.* Presented at the Research Seminars, NSW Biochemical Genetics Service, North Ryde, December 1994.

54. *FRACP Candidate Tutorial on Genetic Metabolic Disorders.* 3 hr tutorial given at the Royal Alexandra Hospital for Children, Camperdown, January 1995.

55. *Genetics of Leigh Disease.* Presented as an Inservice teaching session to nursing staff at the Royal Alexandra Hospital for Children, Camperdown, January 1995.

56. *One in 50,00, two a week.* Paediatric Grand Rounds presentation at Westmead Hospital, Westmead, February 1995.

57. *Paediatrics Course for Foreign Medical Graduates.* Conducted by the University of Sydney Medical Foundation, University of Sydney, Februrary 1995.

58. *Practical Approach to the Investigation of Genetic Metabolic Disorders.* Presented to N.S.W. Genetics Fellows and Consultants, Royal Alexandra Hospital for Children, Camperdown, February 1995.

59. *FRACP Candidate Question & Answer Session on Genetic Metabolic Disorders.* Tutorial given at the Royal Alexandra Hospital for Children, Camperdown, February 1995.

60. *Scope of Human Genetics.* A 6 hour series of lectures given as part of the Graduate Diploma of Clinical Chemistry course run by the University of Western Sydney, March, 1995.

61. *PKU-What's it all about?* Presented to parents of adolescents with phenylketonuria, Royal Alexandra Hospital for Children, Camperdown, March 1995.

62. *Genes and Chromosomes - What's it all about?* Presented at the Rural Doctors Refresher Conference, Narrandera, March 1995.

63. *A Neurologist's Guide to Inborn Errors of Neurotransmitter Metabolism.* Presented at the NSW Epilepsy Society meeting, University of Sydney, April 1995.

64. *Inherited Metabolic Disease in Childhood.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Camperdown, May 1995.

65. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Camperdown, June 1995.

66. *Inborn Errors of Metabolism.* Presented to Sydney University Master of Nutrition and Dietetics Students, Royal Alexandra Hospital for Children, Sydney, June, 1995.

67. *Update on Metabolic Disorders.* Presented at Dept of Paediatrics Seminar & Teaching sessions, John Hunter Hospital, Newcastle, June 1995.

68. *Creating and Using Your Own Reference Library.* Presented at Paediatric Research Seminar series, Royal Alexandra Hospital for Children, Sydney, June, 1995.

69. *Inherited Metabolic Disorders.* Presented at the Diploma of Child Health Teaching Program, Royal Alexandra Hospital for Children, Sydney, July, 1995.

70. *Genetic Metabolic Disorders in the Newborn, and Practical Aspects of Newborn Screening.* Presented to Midwifery students, Gosford Hospital, Gosford, July 1995.

71. *Inherited Metabolic Disease in Childhood.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Camperdown, July 1995.

72. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Camperdown, August 1995.

73. *Treatment of Genetic Disorders.* Presented at the Paediatric Seminars, Nepean Hospital, Penrith, August 1995.

74. *Mitochondrial Diseases and Mitochondrial Inheritance.* Presented to N.S.W. Genetics Fellows and Consultants, Royal Alexandra Hospital for Children, Camperdown, August 1995.

75 *A Clinical Approach to Inborn Errors of Metabolism.* Presented to FRACP trainees in Paediatrics on behalf of ALJESAL Educational Services, Hunters Hill, September 1995.

76 *The Genetics of Mitochondrial Inheritance: Implications for Aging and Disease.* Keynote address at the 33rd Annual Scientific Meeting of the Australasian Association of Clinical Biochemists, Sydney, October 1995.

77. *Inherited Metabolic Disease in Childhood.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Camperdown, October 1995.

78. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Camperdown, October 1995.

79. *Inborn Errors of Metabolism in the Newborn.* Presented at Genetic Seminar, Royal North Shore Hospital, St Leonards, November 1995.

80. *Mitochondria and You: What you need to know.* Presented at the Royal Australasia College of Physicians Annual Scientific Meeting, Sydney, November 1995.

81. *An Approach to the Investigation of Children with Suspected Inborn Errors of Metabolism.* Presented at the Residential Course in Paediatrics, Auckland, New Zealand, November 1995.

82. *Clinicopathological Conference - Gaucher Disease.* Presented at the Residential Course in Paediatrics, Auckland, New Zealand, November 1995.

83. *Mitochondrial DNA.* Presented at the Reproductive Genetics Symposium, Sydney, November 1995.

84. *Cystic Fibrosis: a Genetic Paradigm.* Presented at the Rural Doctors Resource Network Refresher Conference, Bowral NSW, November 1995.

85. *A Clinical Approach to Inborn Errors of Metabolism.* Presented to FRACP trainees in Paediatrics Royal Alexandra Hospital for Children, Westmead, December 1995.

86. *Genetic Metabolic Disorders in the Newborn, and Practical Aspects of Newborn Screening.* Presented to Midwifery students, Gosford Hospital, Gosford, December 1995.

87. *Paediatrics Course for Foreign Medical Graduates.* Conducted by the University of Sydney Medical Foundation, University of Sydney, February 1996.

88. *PKU and You.* Lecture as part of a workshop held for adolescents with PKU, RAHC, February 1996.

89. *Newborn Screening.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, February 1996.

90. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, March 1996.

91. *Genetic Metabolic Disorders in the Newborn, and Practical Aspects of Newborn Screening.* Presented to Midwifery students, Gosford Hospital, Gosford, April 1996.

92. *Newborn Screening.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, June1996.

93. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, June 1996.

94. A Nongeneticist's Approach to Genetic Metabolic Diseases. Presented to the Psychological Medicine Seminar series, August 1996.

95. *Newborn Screening.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead August 1996.

96. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, August 1996.

97. Research into Velo-Cardio-Facial syndrome. Presented at the Annual General Meeting of the VCFS Foundation of Australia, Westmead, August 1996.

98. Paediatric Inborn Errors of Metabolism: Implications for Adulthood. Presented at the Endocrinology Grand Rounds, Westmead Hospital, Westmead, August 1996.

99. Inborn Errors of Metabolism and Congenital Malformations. Presented at the New Children's Hospital Celebratory Meeting, Parramatta, August 1996.

100. *Genetic Metabolic Disorders in the Newborn, and Practical Aspects of Newborn Screening.* Presented to Midwifery students, Gosford Hospital, Gosford, September 1996.

101. *A Clinical Approach to Inborn Errors of Metabolism.* Presented to FRACP trainees in Paediatrics Royal Alexandra Hospital for Children, Camperdown, September 1996.

102. *PKU and You.* Lecture as part of a workshop held for parents of children with PKU, RAHC, September 1996.

103. *Genetic Metabolic Disorders.* Lecture to Diploma of Child Health Candidates, Royal Alexandra Hospital for Children, September 1996.

104. *Gaucher Disease.* Lecture presented at the Medicolegal Society meeting, Sydney, September 1996.

105. *Practical Approach to Genetic Metabolic Disorders.* Presented at the ALJESAL First Part Preparation Course, Hunters Hill, September 1996.

106. *Newborn Screening.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead October 1996.

107. *Inborn Errors of Metabolism.* Presented to Sydney University Master of Nutrition and Dietetics Students, Royal Alexandra Hospital for Children, Sydney, November, 1996.

108. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, November 1996.

109. *The Clinical, Biochemical and Molecular Consequences of Mitochondrial Disorders.* Presented to the Neuropsychiatry Interest Group, New Children's Hospital, Westmead, November, 1996.

110. *Genetic Metabolic Disorders.* Presented at the FRACP Trainee Program, University of Auckland, Auckland New Zealand, November 1996.

111. *Clinicopathological Conference - Hypoglycaemia.* Presented at the FRACP Trainee Program, University of Auckland, Auckland New Zealand, November 1996.

112. Genetic Testing and Research into Velo-cardio-facial syndrome. Presented at the Second VCFS parents seminar, Turramurra, NSW, November 1996.

113. *Newborn Screening.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead February 1997.

114. *Inborn Errors of Metabolism.* Presented to GMP medical students, Sydney, March 1997.

115. *Is it a Syndrome or is it Metabolic?* Presented at the New Zealand Paediatric Update Meeting, Auckland, New Zealand, March 1997.

116. *Case Discussions* Presented at the New Zealand Paediatric Update Meeting, Auckland, New Zealand, March 1997.

117. *Acute Encephalopathy - could it be an Inborn Error of Metabolism?* Presented at the New Zealand Paediatric Update Meeting, Auckland, New Zealand, March 1997.

118. *Inherited Metabolic Causes of Developmental Regression* Presented at the New Zealand Paediatric Update Meeting, Auckland, New Zealand, March 1997.

119. *Workshop on Hypoglycaemia* Presented at the New Zealand Paediatric Update Meeting, Auckland, New Zealand, March 1997.

120. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, March 1997.

121. *PKU: What's It All About?* Presented to Parent Seminar on PKU, The Children's Hospital, Westmead, March 1997.

122. *Inborn Errors and the PICU.* Presented to Paediatric Intensive Care Registrars, The Children's Hospital, Westmead, April 1997.

123. *Newborn Screening.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead April 1997.

124. *Genetic Metabolic Disorders in the Newborn, and Practical Aspects of Newborn Screening.* Presented to Midwifery students, Gosford Hospital, Gosford, April 1997, September 1997.

125. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, May 1997.

126. *Mitochondrial Disorders.* Presented at the "Genetics for the Neurologist" meeting, New Children's Hospital, June 1997.

127. *Treatment for Genetic Disorders.* Presented at the Genetics Awareness Week, Powerhouse Museum, June 1997.

128. *Newborn Screening.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead July 1997.

129. *Inherited Metabolic Disorders*. Presented as part of the Diploma of Child Health course, New Children's Hospital, August 1997.

130. *PKU - Will History Repeat Itself?* Presented at the Endocrine Grand Rounds, Westmead Hospital, August 1997.

131. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, August and September 1997.

132. *Latest in VCFS Research.* Presented at the VCFS Foundation of Australia Annual Meeting, New Children's Hospital, August 1997.

133. *Metabolic Crises.* Presented at the Practical Management of the Critically Ill Child conference, New Children's Hospital, September 1997.

134. *Treatment of Genetic Diseases.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead September 1997

135. *Inborn Errors of Metabolism.* Presented to Sydney University Master of Nutrition and Dietetics Students, Royal Alexandra Hospital for Children, Sydney, October 1997.

136. *Genetic Services for Western Sydney - where are we now and where are we going?* Presented at the Annual Hospital Meeting, New Children's Hospital, October 1997.

137. *Get Ready for the Graduate Medical Student.* Presented at the Annual Hospital Meeting, New Children's Hospital, October 1997.

138. *Classification & Approach to Inborn Errors of Metabolism.* Presented to FRACP candidates, New Children's Hospital, November 1997.

139. *Classification & Approach to Inborn Errors of Metabolism.* Presented to FRACP candidates, Auckland, New Zealand, November 1997.

140. *Inborn Errors of Metabolism.* Presented at Clinical Paediatric Meeting, Nepean Hospital, Sydney, November 1997.

141. *Inherited Metabolic Disorders that Cause Developmental Regression.* Presented at the Paediatric Update, New Children's Hospital, Westmead, November 1997.

142. *Inborn Errors of Metabolism.* Presented to GMP medical students, Sydney, March 1998.

143. *Inborn Errors of Metabolism that Cause Intellectual Disability.* Presented at the Untreated Phenylketonuria symposium, New Children's Hospital, Westmead, July 1998.

144. *Treatment of Genetic Diseases.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, July and September 1998.

145. *Medical Quiz.* Presented to medical students as part of their Paediatrics rotation, The Children's Hospital, Westmead, July 1998.

146. *Mitochondrial Disorders.* Presented to the Genetics Fellows seminar series, New Children's Hospital, July 1998.

147. *Inborn Errors of Metabolism.* Presented to Sydney University Master of Nutrition and Dietetics Students, Royal Alexandra Hospital for Children, Sydney, September 1998

148. *Mitochondrial Disorders.* Presented to the Dept of Molecular and Clinical Genetics seminar series, Royal Prince Alfred Hospital, Sydney, September 1998.

149. *Practical Approach to Genetic Metabolic Disorders.* Presented at the ALJESAL First Part Preparation Course, Hunters Hill, October 1998.

150. *Genetic Metabolic Disorders in the Newborn, and Practical Aspects of Newborn Screening.* Presented to Midwifery students, Gosford Hospital, Gosford, October 1998.

151. *Mitochondrial Disorders.* Presented at a Macquarie University Dept of Genetics seminar, Macquarie University, November 1998.

152. *Mitochondrial Disorders: from the Bedside to the Bench and Back Again.* Presented to the Human Genetics Society of Australasia Scientific meeting, Newcastle, November 1998.

153. *Classification & Approach to Inborn Errors of Metabolism.* Presented to FRACP candidates, Auckland, New Zealand, November 1998.

154. *The Internet for the Paediatrician.* Interactive workshop presented at the Paediatric Update, New Children's Hospital, Westmead, November 1998.

155. *Inborn Errors of Metabolism.* Presented to GMP medical students, Sydney, March 1999.

156. *The New Genetics: What's on the Horizon?* Presented at the Paediatric Update, New Children's Hospital, Westmead, November 1998.

157. *Somatic Cell Mitochondrial Mutations and Respiratory Chain Disorders.* Presented at the Sereno Symposium, "The Bottleneck: Gamete and Embryo Mitochondria in Humans", Sydney, May 1999.

158. *Treatment of Genetic Disorders.* Presented at the Masters of Human Genetics Lecture Series, Macquarie University, June 1999.

159. *Advances in the Management of Phenylketonuria (PKU) in Australia.* Presented at the International Association of Special Education 6th Biennial Conference, Sydney, July 1999.

160. *Update on PKU.* Presented at the PKU Parents' Support Group Annual General Meeting, Sydney, July 1999.

161. *The New Genetics and Mitochondrial Respiratory Chain Disorders.* Presented at the Australasian Association of Clinical Biochemists Current Concepts Videoconference, Sydney, July 1999.

162. *Inherited Metabolic Disorders.* Presented at the Diploma of Child Health Course, New Children's Hospital, August 1999.

163. *Familial Mediterranean Fever.* Presented at Grand Rounds, New Children's Hospital, Sydney, October 1999.

164. *Metabolic Disease.* Presented at the ALJESAL 1999 FRACP (Paed) First Part Preparatory Course, Sydney, October 1999.

165. *Inborn Errors of Metabolism.* Presented at the Masters of Nutrition and Dietetics Course, New Children's Hospital, Sydney, October 1999 .

166. *Unravelling the Genetics of Mitochondrial Disorders.* Presented at the New Children's Hospital, Annual Hospital Meeting, Sydney, October 1999.

167. *The New Genetics and Mitochondrial Respiratory Chain Disorders.* Presented at the Paediatric Update, Starship Hospital, Auckland, New Zealand, November 1999.

168. *Classificiation & Approach to Inborn Errors of Metabolism.* Presented to FRACP candidates, Auckland, New Zealand, November 1999.

169. *Unravelling the Genetics of Mitochondrial Disorders.* Presented to GMP Medical Students, New Children's Hospital, Westmead, January 2000.

170. *Principles of Human Genetics.* Presented to Psychiatry Fellows, NSW Institute of Psychiatry, Parramatta, February 2000.

171. *Inborn Errors of Metabolism.* Presented to GMP medical students, Sydney, February 2000.

172. *Tutorial on Inborn Errors of Metabolism*. Presented to FRACP Paediatric Trainees, New Children's Hospital, Westmead, April 2000.

173. *Genetic Metabolic Disorders in the Newborn, and Practical Aspects of Newborn Screening.* Presented to Midwifery students, Gosford Hospital, Gosford, May 2000.

174. *Inherited Metabolic Disorders.* Presented at the Diploma of Child Health Course, New Children's Hospital, May 2000.

175. *The Human Genome Project – a View from the Inside.* Presented at the Human Genome Project and Disability: Promises and Threats Conference. Sydney, May 2000.

176. *Mitochondrial Disorders.* Presented at the Physiotherapy Dept, New Children's Hospital, Westmead, June, August, October 2000.

177. *Inborn Errors of Metabolism Tutorial – FRACP Candidates.* New Children's Hospital, Westmead, June 2000.

178. *Rett Syndrome, MECP2 and Transcriptional Silencing: Too Much of a Good Thing?* Presented at the Dept of Biochemistry, University of Sydney, Sydney, August 2000.

179. *Inborn Errors of Metabolism.* Presented at the Masters of Nutrition and Dietetics Course, New Children's Hospital, Sydney, August 2000.

180. *Metabolic Causes of Cardiomyopathy.* Presented to PICU Registrars, New Children's Hospital, Westmead, August 2000.

181. *Mitochondrial Respiratory Chain Disorders.* Presented at the Western Sydney Genetics Program Seminar Series, New Children's Hospital, Westmead, August 2000.

182. *The Human Genome Project – a View from the Inside.* Presented at the Conference on Health Issues in Developmental Disability. St George Hospital, Sydney, November 2000.

183. *Rett Syndrome: Too Much of a Good Thing?* Presented at Paediatric Update, Starship Hospital, Auckland, New Zealand, November 2000.

184. *Classification & Approach to Inborn Errors of Metabolism.* Presented to FRACP candidates, Auckland, New Zealand, November 2000.

185. *Metabolic Disease.* Presented at the ALJESAL 2000 FRACP (Paed) First Part Preparatory Course, Sydney, October 2000.

186. *Rett Syndrome: from the Clinic to the Laboratory and Back Again.* Presented to the Medical Faculty at the University of Siena, Siena, Italy, December 2000.

187. *Inborn Errors of Metabolism.* Presented to USydMP and GDP students, University of Sydney, March 2001.

188. *Principles of Human Inheritance.* Presented to USydMP and GDP students, University of Sydney, March 2001

189. *X-linked Adrenoleukodystrophy.* Presented at Medical Grand Rounds, The Children's Hospital at Westmead, Sydney, March 2001.

190. *Somatic Cell Strategies for the Genetic Dissection of Mitochondrial Respiratory Chain Defects.* Presented at the Children's Medical Research Institute, Sydney, April 2001.

191. *Symposium: Human Genome Project.* Chair of a session at the Royal Australasian College of Physicians Annual Scientific Meeting, Sydney, May 2001.

192. *Inherited Metabolic Disorders.* Presented at the Diploma of Child Health Course, New Children's Hospital, June 2001.

193. *Treatment of Genetic Disorders.* Presented at the Masters of Human Genetics Lecture Series, Macquarie University, June 2001.

194. *Postgraduate Student Recruitment.* Presented at the Academic Forum, University of Sydney, Sydney, August 2001.

195. *Mitochondrial Respiratory Chain Disorders.* Presented at the Neurometabolic Update. Children's Hospital at Westmead, Westmead, August 2001.

196. *Somatic Cell Strategies for the Genetic Dissection of Mitochondrial Respiratory Chain Defects.* Presented at the Children's Cancer Institute Australia Seminar Series, Sydney Children's Hospital, Randwick, September 2001.

197. *Metabolic Disease.* Presented at the ALJESAL 2001 FRACP (Paed) First Part Preparatory Course, Sydney, October 2001.

198. *The Human Genome Project: Opportunities, Challenges and Consequences for Population Screening.* Plenary speaker at the 4th Asia Pacific Regional Meeting of the International Society for Neonatal Screening, Manila, The Philippines, October 2001.

199. *A Clinical Approach to Inborn Errors of Metabolism Presenting in the Newborn.* Invited speaker at the 4th Asia Pacific Regional Meeting of the International Society for Neonatal Screening, Manila, The Philippines, October 2001.

200. *A Clinical Approach to Inborn Errors of Metabolism.* Paediatric Update, Children's Hospital at Westmead, November 2001.

201. *Somatic Cell Strategies for the Genetic Dissection of Mitochondrial Respiratory Chain Defects.* Presented at the Western Sydney Genetics Program Seminar Series, Children's Hospital at Westmead, November 2001

202. *Possible Role of Metabolic Disorders in Stillbirth – Current Status and Future Research.* Invited speaker to the conference sponsored by SIDSNSW - "Investigation and Prevention of Stillbirths: Setting the Policy and Research Agenda, University of Sydney, Sydney, November 2001.

203. *Investigation and Management of Inborn Errors of Metabolism*. Paediatric Education Sessions, Blacktown/Mt Druitt Health, December 2001.

204. *Biochemical Genetics Case of the Week.* Children's Hospital at Westmead Lunchtime Seminars, February & September 2002.

205. *The Human Genome Project.* The Children's Hospital at Westmead Annual Hospital Meeting, Westmead, March 2002.

206. *Inborn Errors of Metabolism.* Presented to USydMP First year Medical Students, March 2002.

207. *Principles of Human Inheritance.* Presented to USydMP First year Medical Students, March 2002.

208. *Inborn Errors Leading to Neurodegeneration.* Bear Cottage Seminar Series, Manly, March 2002.

209. *Clinical, Biochemical and Genetic Aspects of Mitochondrial Disorders.* Genetic Fellows Seminar Series, Children's Hospital at Westmead, April 2002.

210. *The Laboratory Investigation of Genetic Metabolic Disorders Presenting with Liver Disease.* 28th Annual Scientific Meeting of the Australasian Division of the International Academy of Pathology, Sydney, May 2002.

211. *Treatment of Genetic Disorders.* Presented at the Masters of Human Genetics Lecture Series, Macquarie University, June 2002.

212. *Inherited Metabolic Disorders.* Diploma of Child Health course, Children's Hospital at Westmead, Sydney, July 2002.

213. *Meet the Expert – Genetic Disorders.* Regional Paediatrician teleconference, August 2002.

214. *Getting the Ticket: Practical Aspects in the PhD Examination Process.* Professional Development Skills Seminar Series, Children's Hospital at Westmead, September 2002.

215. *IRSA Rett Databases: International Phenotype Database and RettBASE: IRSA MECP2 Variation Database.* Rett Syndrome Satellite Meeting, Beijing, September 2002.

216. *Rett Syndrome and MECP2: Phenotype – Genotype Correlations.* Rett Syndrome Satellite Meeting, Beijing, September 2002.

217. *A Clinical Approach to Genetic Metabolic Disorders.* Presented at the ALJESAL 1999 FRACP (Paed) First Part Preparatory Course, Sydney, October 2002.

218. *Inborn Errors of Metabolism.* Presented at the Masters of Nutrition and Dietetics Course, New Children's Hospital, Sydney, October 2002.

219. *New Research into PKU.* National PKU Conference, Sydney, October 2002.

220. *Research Candidature Working Party Reports.* Faculty of Medicine Workshop presentation, Sydney, November 2002.

221. *Inborn Errors of Metabolism.* FRACP Trainee Seminar Series, Children's Hospital at Westmead, November 2002.

222. *Inborn Errors of Metabolism.* Presented to USydMP first year Medical students and GDP first year Dental Students, March 2003.

223. *Principles of Human Inheritance.* Presented to USydMP first year Medical students and GDP first year Dental Students, March 2003.

224. *Clinical, Biochemical and Genetic Aspects of the Mitochondrial Respiratory Chain Defects.* Presented at Pathology Update 2003 (Royal College of Pathologists of Australia), Darling Harbour, Sydney, March 2003.

225. *Biochemical Genetics Case of the Week – Maple Syrup Urine Disease.* Presented to FRACP trainees, Children's Hospital at Westmead, March 2003

226. *Rett Syndrome and MECP2: Of Mouse and Women.* Presented at Grand Rounds, Children's Hospital at Westmead, Sydney, May 2003.

227. *MECP2 and Beyond: Phenotype-Genotype Correlations in Rett Syndrome.* Presented at Grand Rounds, Sydney Children's Hospital, Sydney, May 2003.

228. *Not Enough Energy? Tell me about it!!* Presented at Grand Rounds, Children's Hospital at Westmead, Sydney, May 2003.

229. *Treatment of Genetic Disorders.* Presented at the Masters of Human Genetics Lecture Series, Macquarie University, June 2003.

230. *Inherited Metabolic Disorders.* Diploma of Child Health course, Children's Hospital at Westmead, Sydney, July 2003.

231. *Genetics and the General Practitioner: Advances that will Affect GPs in Paediatrics in the Next 5 Years.* Presented at the Paediatric Postgraduate Weekend for General Practitioners, Children's Hospital at Westmead, Sydney, August 2003.

232. *Getting the Ticket: Practical Aspects in the PhD Examination Process.* Professional Development Skills Seminar Series, Children's Hospital at Westmead, Sydney, August 2003.

233. *A Jaundiced View of Mitochondrial Respiratory Chain Disorders.* Presented at the Festschrift for Ramanand Kamath, Children's Hospital at Westmead, Sydney, August 2003.

234. *Rett Syndrome and MECP2: Of Mouse and Women.* Presented at Westmead Millennium Institute Seminar Series, Sydney, August 2003.

235. *Incurable Inborn Errors of Metabolism Diagnosis & Management.* Presented at Bear Cottage Education Day, Children's Hospital at Westmead, Sydney, September 2003.

236. *Inborn Errors of Metabolism.* Presented at the Masters of Nutrition and Dietetics Course, New Children's Hospital, Sydney, October 2003.

237. *Rett Syndrome and MECP2: Challenges in Searching for Phenotype-Genotype Links.* Presented at the Rett Syndrome of UK Family Weekend, Northampton, UK, October 2003.

238. *A Double Whammy: Brain Injury and a Rare Genetic Disorder.* Presented at Grand Rounds Ethics Forum, Children's Hospital at Westmead, Sydney, October 2003.

239. *A Clinical Approach to Genetic Metabolic Disorders.* ALJESAL FRACP (Paed) Written Examination Preparation Course, Sydney, October 2003.

240. *Inborn Errors of Metabolism.* FRACP Trainee Seminar Series, Children's Hospital at Westmead, December 2003.

241. *Inborn Errors of Metabolism.* Presented to USydMP first year Medical students and GDP first year Dental Students, March 2004.

242. *Principles of Human Inheritance.* Presented to USydMP first year Medical students and GDP first year Dental Students, March 2004.

243. *Mitochondrial Encephalomyopathies – Something for Everyone.* Presented at Pathology Update 2004 (Royal College of Pathologists of Australia), Darling Harbour, Sydney, March 2004.

244. *The Human Genome Project: Promises and Problems in Pediatric Practice.* Plenary presentation at the 41st Annual Convention of the Philippines Pediatric Society, Manila, April 2004.

245. *New Technologies for the Detection of Submicroscopic Chromosomal Rearrangements.* Plenary presentation at the 41st Annual Convention of the Philippines Pediatric Society, Manila, April 2004.

246. *Metabolic Catastrophies Masquerading as Sepsis.* Plenary presentation at the 41st Annual Convention of the Philippines Pediatric Society, Manila, April 2004.

247. *Treatment of Genetic Disorders.* Presented at the Masters of Human Genetics Lecture Series, Macquarie University, June 2004.

248. *Inherited Metabolic Disorders.* Diploma of Child Health course, Children's Hospital at Westmead, Sydney, July 2004.

249. *Getting the Ticket: Practical Aspects in the PhD Examination Process.* Professional Development Skills Seminar Series, Children's Hospital at Westmead, Sydney, August 2004.

250. *Postgraduate Issues*. Plenary presentation with Professor Masud Behnia, Fourth Research Conference: From Cell to Society 4, College of Health Sciences, University of Sydney, Leura, November 2004. (plenary oral)

251. *A Clinical Approach to Genetic Metabolic Disorders.* ALJESAL FRACP (Paed) Written Examination Preparation Course, Sydney, September 2004.

252. *PDAs in Clinical Practice* – session chaired as part of Paediatric Update, CHW, 18th November 2004

253. *Top 5 Websites* – Paediatric Update, CHW, 19th November 2004

254. *Supervisor Workshop*, Sydney University, 29th November 2004

255. *Supervisor Workshop*, Westmead Hospital, 3rd December 2004

256. *Inborn Errors of Metabolism.* FRACP Trainee Seminar Series, Children's Hospital at Westmead, December 2004.

257. *PKU Research: Recent Advances* – NSW PKU Association AGM, 13th February 2005

258. *Inborn Errors of Metabolism* – USydMP year 1 students, Sydney University, 4th March 2005

259. *Genetic Metabolic Disorders and the Palliative Care Physician* – Annual Paediatric Palliative Care Symposium, CHW, 4th March 2005

260. *Rett Syndrome, More than One Gene More than One Story* – Kolling Institute, RNSH, 7th March 2005

261. *Principles of Human Inheritance* – USydMP year 1 students, Sydney University, 8th March 2005

262. *Supervisor Workshop,* ANZAC Institute – Concord Hospital, 8th March 2005

263. *University Policies – What you should know and how to train your supervisor* – CHW, 9th March 2005

264. *Chaired Epigenetics Session,* RCPA Annual Conference – Darling Harbour, Sydney, 12th March 2005

265. *Progress with CHW Rett Syndrome Research* – Rotary Club of Narellan meeting, Camden, 15th March 2005

266. *Introduction to Inborn Errors of Metabolism* – Lysosomal Storage Disorders Short Course, CHW, 7th April 2005

267. *Rett Syndrome, More than One Gene More than One Story* – Oncology Research Unit, CHW, 12th April 2005

268. *How Sweet It Is – NOT!* – Grand Rounds presentation, CHW, 14th April 2005

269. *Rett Syndrome, More than One Gene More than One Story* – "A Healthy Start to Life" Research Program in honour of Dr Fiona Stanley, Medical Foundation Building, Faculty of Medicine, Sydney University, 15th April 2005

270. *Inborn Errors in the Newborn.* NICU, Nepean Hospital, 28th April 2005

271. *Rett Syndrome, More than One Gene More than One Story*. Dept of Molecular Genetics, University of Padua, 4th May 2005

272. *CDKL5*: a Second Rett syndrome Gene? Implications for other Neurodevelopmental Disorders. University of Padua Scientific Program, 5th May 2005

273. *Rett Syndrome: More than one gene, more than one story*. HGSA (NSW Branch) Annual Scientific Meeting, CHW, 22nd June 2005

274. *Abnormal expression of a subset of genes in the frontal cortex of Rett syndrome patient brains.* WSGP Research Workshop, CHW, 27th May 2005

275. *Forced myogenesis of fibroblasts can unmask mitochondrial respiratory chain (RC) defects.* WSGP Research Workshop, CHW, 27th May 2005

276. *Treatment of Genetic Disorders.* Presented at Macquarie University, 1st June 2005

277. *Inborn Errors of Metabolism.* Presented to Dental students, CHW, 13th July 2005.

278. *Rett Syndrome: A Neurodevelopmental Disorder with a Difference.* Presented at the Neuroscience – from bench to bedside showcase, Sydney University, 15th July 2005

279. *A Clinical Approach to Inborn Errors of Metabolism.* Diploma of Child Health Lecture, CHW, 18th July 2005.

280. *Getting the Ticket: Practical Aspects of the PhD thesis examination process.* Presented to Westmead Hub Postgraduate Research Students, CHW, 5th August 2005.

281. *Inborn errors of metabolism – part 2: Neonatal Epileptic Encephalopathy and Peroxisomal Disorders.* NICU, Nepean Hospital, 18th August 2005.

282. *Rett syndrome: more than one gene, more than one story.* Department of Pharmacology, Sydney University, 24th August 2005.

283. *Galactosaemia.* As part of parent/patient galactosaemia education day, CHW 31st August 2005.

284. *PICU issues with inborn errors of metabolism.* PICU, CHW, 15th September 2005.

285. *Clinical approach to inborn errors of metabolism.* ALJESAL FRACP program, St Joseph's College, Hunters Hill, 27th September 2005.

286. *New research in PKU.* Metabolic Dietary Diseases Association conference, Canberra, 9th October 2005.

287. *Inborn Errors of Metabolism in Adults* – presented to FRACP trainees, Westmead Hospital, 15th November 2005.

288. *Update on research into Rett syndrome.* Rett Syndrome Parents Education Meeting, CHW, 20th November 2005.

289. *Diagnosis and Genetics of Rett syndrome APSU* – Rett Syndrome Health Professionals Workshop, CHW, 21st November 2005.

290. *Inborn Errors of Metabolism* – presented to FRACP trainees, CHW, 21st November 2005.

291. *Medical Journal Club* – presented as part of the Paediatric Update, CHW, 25th November 2005.

292. *University Policies – What you should know and how to train your supervisor* – CHW, 8th March 2006

293. *Inborn Errors of Metabolism* – USydMP year 1 students, Sydney University, 3rd March 2006

294. *Principles of Human Inheritance* – USydMP year 1 students, Sydney University, 3rd March 2006

295. *A Clinical Approach to Inborn Errors of Metabolism.* Diploma of Child Health Lecture, CHW, 21st August 2006

296. *Getting the Ticket: Practical Aspects of the PhD thesis examination process.* Presented to Westmead Hub Postgraduate Research Students, CHW, 12th May 2006

297. *Clinical approach to inborn errors of metabolism.* ALJESAL FRACP program, St Joseph's College, Hunters Hill, 4th October 2006

298. *Regional Paediatrician's Group Teleconference.* Teleconference, 24th October 2006

299. *Clinical and Laboratory Aspects of Mitochondrial Diseases in Childhood*. Presentation to Genetics Fellows, CHW, Sydney, 14th November 2006

300. *Life Threatening Metabolic Acidosis: to B2 or not to B2?* Grand rounds presentation, CHW, Sydney, 23rd November 2006

301. *Inborn Errors of Metabolism* – presented to FRACP trainees, CHW, 27th November 2006

302. *University Policies – What you should know and how to train your supervisor* – CHW, 7th March 2007

303. *Inborn Errors of Metabolism* – USydMP year 1 students, Sydney University, xxth March 2007

304. *Principles of Human Inheritance* – USydMP year 1 students, Sydney University, xxth March 2007

305. *Large Neutral Amino Acids – current opinion and use in PKU*, Nutricia conference, Dietary Management of Inherited Metabolic Disease, Royal College of Physicians, London, 16th March 2007

306. *Biochemical Genetics Journal Club*, CHW, 29th March 2007

307. *Lysosomal Storage Disorders*, presented to genetic and metabolic fellows, CHW, April 2007

308. *Inborn Errors of Metabolism with Metabolic Acidosis*, presented at Nepean Hospital NICU, 10th May 2007

309. *Getting the Ticket: Practical Aspects of the PhD thesis examination process.* Presented to Westmead Hub Postgraduate Research Students, CHW, 6th July 2007

310. *A Clinical Approach to Inborn Errors of Metabolism.* Diploma of Child Health Lecture, CHW, 27th August 2007

311. *A Huge Leap Backwards, Then Small Steps Forward: Profile of a Rett Multidisciplinary Management Clinic, presented at the Rett Syndrome*: Diagnosis, Genetics, Epidemiology and Clinical Management Workshop, Peking University First Hospital, Beijing, China, 7th – 13th September 2007

312. *Rett Syndrome Research – What's New and Exciting*, presented at the Rett Syndrome: Diagnosis, Genetics, Epidemiology and Clinical Management Workshop, Peking University First Hospital, Beijing, China, 7th – 13th September 2007

313. *Clinical and Genetic Diagnosis of Rett Syndrome,* presented at the Rett Syndrome: Diagnosis, Genetics, Epidemiology and Clinical Management Workshop, Peking University First Hospital, Beijing, China, 7th – 13th September 2007

314. *Western Sydney Genetics Program: Services and Functions,* presented at CHW Forum, 20th September 2007

315. *Clinical and Laboratory Aspects of Mitochondrial Diseases in Childhood*. Presentation to Genetics Fellows, CHW, Sydney, 9th October 2007

316. *Clinical Approach to Inborn Errors of Metabolism.* ALJESAL FRACP program, St Joseph's College, Hunters Hill, 11th October 2007

317. *Appropriate Metabolic Investigation of Children with Developmental Delay,* presented at the Developmental Disorders in Paediatric Neurology: A Practical Approach, CHW, 14th November 2007

318. *Inborn Errors of Metabolism* – presented to FRACP trainees, CHW, November 2007

319. *University Policies – What you should know* – CHW, 5th March 2008

320. *One mother or two? Donor mitochondria to correct a mitochondrial DNA defect* – Ethics Forum, CHW, 11th March 2008

321. *Strategies for the prevention and treatment of genetic disease* – presented to USydMP 1st year students, University of Sydney, 24th April 2008

322. *Strategies for the prevention and treatment of genetic disease* – presented to Biological Sciences students, Macquarie University, 23rd May 2008

323. *Something doesn't smell right…* – presented to FRACP trainees, CHW, 28th May 2008

324. *Research into Rett syndrome* – presented to Science Extension students, Barker College, Hornsby, 29th May 2008

325. *Clinical Approach to Inborn Errors of Metabolism.* ALJESAL FRACP program, St Joseph's College, Hunters Hill, 2nd October 2008

326. *A Clinical Approach to Inborn Errors of Metabolism* – presented to FRACP trainees, CHW, 3rd November 2008

327. *The Most Common Metabolic Questions I Get Asked Are…* – presented at the Paediatric Update Conference, CHW, 21st November 2008

328. *Genetic Metabolic Disorders Research Group* – presented at the CMRI-CHW Research Heads symposium, CMRI, 5th December 2008

329. *A Clincial Approach to Genetic Metabolic Disorders* – presented at the Australian Postgraduate Course in Neuropathology, University of Sydney, 16th January 2009.

330. *Lecture on inborn errors of metabolism* – presented to genetics fellows at the CHW, 10th February.

331. *Lecture on autozygosity testing* – presented at the WSGP seminar series, 17th February

332. *There is Art(s) in Science: Therapuetic Implications of a Gene Discovery in an Australian Family* – presented at Grand Rounds, CHW, 19th February.

333. *PKU Research at the Children's Hospital at Westmead* – Presented at the Blacktown Rotary Club meeting, Blacktown, 24th March 2009.

334. *Is it metabolic*? – presented to Advanced Trainees, CHW, 15th April 2009.

335. *What makes a good supervisor?* – presented at a Faculty of Health Sciences supervisor forum, 23rd April 2009.

336. *Strategies for the prevention and treatment of genetic disease* – presented to USydMP 1st year students, University of Sydney, 24th April 2009.

337. *Rett syndrome research: the Australian Experience* – presented at the Rett syndrome parent workshop, Children's Hospital at Westmead, 29th May 2009.

338. *Strategies for the prevention and treatment of genetic disease* – presented to B Biol Sci students, Macquarie University, 5th June 2009.

339. *Getting the Ticket: Practical Aspects of the PhD thesis examination process.* Presented to Westmead Hub Postgraduate Research Students, CHW, 7th August 2009.

340. *Gene Discovery and Treatment of Mitochondrial Disease in Childhood.* Presented to the RMO Journal Club, CHW, 7th August 2009.

341. *A Clinical Approach to Inborn Errors of Metabolism.* Diploma of Child Health Lecture, CHW, 24th August 2009.

342. *Clinical Approach to Inborn Errors of Metabolism.* ALJESAL FRACP program, St Joseph's College, Hunters Hill, 8th October 2009.

343. *RCPA-HGSA Molecular Genetics Quality Assurance Program: Partnership in Evolution.* Presented at the RCPA Annual Conference, Melbourne, 25th February 2010.

344. *Treatment of Mitochondrial Disorders in Children.* Presented at the AMDF Conference, Royal Children's Hospital, Brisbane, 10th April 2010.

345. *Strategies for the Prevention and Treatment of Genetic Disorders.* Presented to USydMP students, Sydney University, 16th April 2010.

346. *Rett syndrome: new clinical and molecular insights.* Presented at Malaysian Genetics Conference, Kuala Lumpur, Malaysia, 22nd April 2010.

347. *Genetic metabolic disorders causing acute encephalopathy.* Presented at Malaysian Genetics Conference, Kuala Lumpur, Malaysia 22nd April 2010.

348. *Clinical and molecular aspects of mitochondrial diseases in childhood.* Presented at Malaysian Genetics Conference, Kuala Lumpur, Malaysia 23rd April 2010.

349. *Inherited metabolic causes of cardiomyopathy in childhood and beyond.* Presented at Malaysian Genetics Conference, Kuala Lumpur, Malaysia 23rd April 2010.

350. *Rett syndrome: recent discoveries and future research directions.* Grand rounds presentation, CHW, 13th May 2010.

351. *PKU: from bedside to bench and back again.* Presented to Year 12 Barker College students, Hornsby, 19th May 2010.

352. *Strategies for the prevention and treatment of genetic disease* – presented to B Biol Sci students, Macquarie University, 4th June 2010.

353. *Innovations in the Treatment of OTC Deficiency: Clinical Blood, Sweat and Tears, and Lessons from the Mouse* – presented at grand rounds, CHW, 29th July 2010.

354. *A Clinical Approach to Inborn Errors of Metabolism.* Diploma of Child Health Lecture, CHW, 9th August 2010.

355. *Giving lecture - Getting the Ticket: Practical Aspects in the PhD Thesis examination process* – given to Westmead Hub postgraduate students, CHW, 24th September 2010.

356. *Clinical Approach to Inborn Errors of Metabolism.* ALJESAL FRACP program, St Joseph's College, Hunters Hill, 29th September 2010.

357. *IEMs causing encephalopathy in the newborn.* Presented to genetics fellows, CHW, 25th February 2010.

358. Plenary lecture at the World Rare Diseases Day event, Opera House, Sydney, 28th February 2011.

359. *Progressive neurological disorders: metabolic perspective.* Presented to Genetics Fellows, 25th March 2011.

360. *Strategies for the Prevention and Treatment of Genetic Disorders.* Presented to USydMP students, Sydney University, 8th April 2011.

361. *Fatty acid oxidation defects.* JMO teaching session, CHW, 11th May 2011.

362. *A Clinical Approach to Inborn Errors of Metabolism.* Diploma of Child Health Lecture, CHW, 16th May 2011.

363. *Strategies for the prevention and treatment of genetic disease.* Presented to B Biol Sci students, Macquarie University, 27th May 2011.

364. *Clinical and Laboratory Aspects of Mitochondrial Diseases in Childhood.* Presented to Genetics Fellows, 14th June 2011.
365. *Inborn errors in paediatric patients* – 2011 Australian Neuropathology Course, 15th July 2011
366. *The genetics and biology of Rett syndrome: are there potential avenues for therapy?* – RTT Parent meeting, 26th May, Brisbane, Australia.
367. *Work/life balance for researchers.* – Early Career Researcher seminar series, 31st July, Children's Hospital at Westmead.
368. *Inborn errors of metabolism causing neonatal encephalopathy.* – RMO teaching session, 7th August, Children's Hospital at Westmead.
369. *Getting the Ticket: Strategies and Tips for Having Your Thesis Passed with the Minimum of Fuss!!* – Discipline of Paediatrics and Child Health Postgraduate Research Student Professional Development Series, University of Sydney, 2nd November.
370. *Getting the Ticket: Strategies and Tips for Having Your Thesis Passed with the Minimum of Fuss!!* – Discipline of Public Health Seminar Series, University of Sydney, 13th November.
371. *Latest advances in mitochondrial therapies.* – mitochondrial parent group meeting, sponsored by the AMDF, Brisbane, 24th November.
372. *Genetics of Rett syndrome.* – Australasian genetics fellows webinar series, 30th November.
373. *Strategies for the prevention and treatment of genetic disease* – presented to USydMP 1st year students, University of Sydney, 24th April 2011.
374.