



Dr. Minal Menezes

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Education

2010 – 2014 PhD (Medicine), University of Sydney, Australia
2000 – 2002 MSc (Microbiology), Bangalore University, India
1999 – 2002 BSc (Biotechnology, Microbiology), Bangalore University, India

Employment history

2016 October

Research Officer/ Conjoint Senior Lecturer, Sydney Medical School
The Department of Anaesthesia/The Children's Hospital at Westmead Clinical School
The Children's Hospital at Westmead

2014 September – 2016 September

Post-doctoral Scientist
Metabolic Research Unit/Kids Research Institute (KRI)
The Children's Hospital at Westmead

2010 – 2014

PhD student
Sydney Medical School, Discipline of Pediatrics and Child Health
Kids Research Institute (KRI), University of Sydney

2009 – 2014

Metabolic Research Unit/Kids Research Institute (KRI)
The Children's Hospital at Westmead

2005 – 2009

Gene Expression Specialist
Dubaiomics FZ-LLC – Dubai United Arab Emirates

2004

Research Associate

Molecular Connections (A Division of Dr. Reddy's Pharmaceuticals) – Bangalore, India

2002 – 2004

Research Assistant

Institute of Bioinformatics – Bangalore, India

Certifications

- Clinical Genomics Data Analysis Practical Course - The Garvan Institute of Medical Research
- Certification of completion for training on Essential GCP Training for Clinical Investigators - ARCS Australia
- Certificate of Attendance - Informed Consent Training Workshop - Caledonian Clinical Training
- Certificate of attendance – Trials and Translation Research Studies – NHMRC Clinical trials centre

Awards and Scholarships

- **2016** Fresh Science New South Wales Finalist
- **2015** Peter Bancroft Prize – University Medal awarded by Sydney Medical School for outstanding thesis.
- **2015** Recipient of Early Career Researcher Kick Start Grant (\$25,000) from University of Sydney
- **2015** Award for Excellence in Mitochondrial Research - Australian Mitochondrial Disease Foundation
- **2015** New Investigator Award - Human Genetics Society of Australasia (HGSA)
- **2015** Best Oral presentation - Australian Society of Inborn Errors of Metabolism (HGSA conference Perth)
- **2015** Balnaves Foundation People's Choice Award at Early Careers Research Showcase
- **2015** ECR overseas Travel Grant (\$2500) to attend SSIEM conference in Lyon France
- **2014** Awarded Australian Society of Inborn Errors of Metabolism: Best Poster award at Human Genetics Society of Australasia conference in Adelaide
- **2014** Human Genetics Society of Australasia International Travel Grant (\$5000)
- **2014** Awarded Australian Society of Inborn Errors of Metabolism: Travel Scholarship to attend EurMit 2014 Conference in Finland
- **2015** Awarded Best Publication by a higher degree research student- Discipline of Paediatrics and Child health 2013

- **2013** Human Genetics Society of Australasia Student Prize
- **2013** Awarded the Burroughs Wellcome Scholarship by the United Mitochondrial Disease Foundation USA
- **2011** Travel scholarship (\$3000) to attend the 12th International Conference of Human Genetics in Montreal Canada 2011 from Australian Mitochondrial Disease Foundation

Committee Membership

- 2017 - Present Sydney Children's Hospital Network Human Research Ethics Executive Committee member.
- 2017 AMED (Applied Medical Science) 3003 and 3004 Curriculum Development committee member at Westmead.
- 2017 Human Genetics Society of Australasia (HGSA) - NSW executive committee member
- Australian Society for Biochemistry and Molecular Biology (ASBMB) 2011-2012
- American Society of Human Genetics (ASHG) 2010-2011
- Australian Society for Medical Research (ASMR) 2014-2015
- Australian Society of Inborn Errors of Metabolism (ASIEM) 2011- Current

Publications

- Michael Nafisinia, **Minal Juliet Menezes**, Wendy Anne Gold et al.. "Tread carefully: A functional variant in the human NADPH oxidase 4 (NOX4) is not disease causing" Molecular Genetics and Metabolism Volume 123, Issue 3, March 2018, Pages 382-387
- **Menezes, M. J.**, L. G. Riley and J. Christodoulou (2014). "Mitochondrial respiratory chain disorders in childhood: insights into diagnosis and management in the new era of genomic medicine." Biochim Biophys Acta **1840**(4): 1368-1379.
- Riley, L. G., M. J. **Menezes, J.** Rudinger-Thirion, R. Duff, P. de Lonlay, A. Rotig, M. C. Tchan, M. Davis, S. T. Cooper and J. Christodoulou (2013). "Phenotypic variability and identification of novel YARS2 mutations in YARS2 mitochondrial myopathy, lactic acidosis and sideroblastic anaemia." Orphanet J Rare Dis **8**: 193.
- Gaignard, P*, **M. Menezes***, M. Schiff, A. Bayot, M. Rak, H. Ogier de Baulny, C. H. Su, M. Gilleron, A. Lombes, H. Abida, A. Tzagoloff, L. Riley, S. T. Cooper, K. Mina, P. Sivadurai, M. R. Davis, R. J. Allcock, N. Kresoje, N. G. Laing, D. R. Thorburn, A. Slama, J. Christodoulou and P. Rustin (2013). "Mutations in CYC1, encoding cytochrome c1 subunit of respiratory chain complex III, cause insulin-responsive hyperglycemia." Am J Hum Genet **93**(2): 384-389.

- Miller, D. K.*, **M. J. Menezes***, C. Simons*, L. G. Riley, S. T. Cooper, S. M. Grimmond, D. R. Thorburn, J. Christodoulou and R. J. Taft (2014). "Rapid Identification of a Novel Complex I MT-ND3 m.10134C>A Mutation in a Leigh Syndrome Patient." PLoS One **9**(8): e104879.
- **Menezes, M.J.**, Guo, Y., Zhang, J., Riley, L.G., Cooper, S.T., Thorburn, D.R., Li, J., Dong, D., Li, Z., Glessner, J. et al. (2015) Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure, and lactic acidemia. Human molecular genetics, 2015, 1-11
- Guo, Y., **Menezes, M.J.***, Menezes, M.P., Liang, J., Li, D., Riley, L.G., Clarke, N.F., Andrews, P.I., Tian, L., Webster, R. et al. (2014) Delayed diagnosis of congenital myasthenia due to associated mitochondrial enzyme defect. Neuromuscular Disorders 12/2014; DOI:10.1016/j.nmd.2014.11.017
- Harsha, H. C., S. Suresh, R. Amanchy, N. Deshpande, K. Shanker, A. J. Yatish, B. Muthusamy, B. M. Vrushabendra, B. P. Rashmi, K. N. Chandrika, N. Padma, S. Sharma, J. L. Badano, M. A. Ramya, H. N. Shivashankar, S. Peri, D. R. Choudhury, M. P. Kavitha, R. Saravana, V. Niranjana, T. K. Gandhi, N. Ghosh, S. Chandran, **M. Menezes**, M. Joy, S. S. Mohan, N. Katsanis, K. S. Deshpande, C. Raghothama, C. K. Prasad and A. Pandey (2005). "A manually curated functional annotation of the human X chromosome." Nat Genet **37**(4): 331-332.
- Peri, S., J. D. Navarro, R. Amanchy, T. Z. Kristiansen, C. K. Jonnalagadda, V. Surendranath, V. Niranjana, B. Muthusamy, T. K. Gandhi, M. Gronborg, N. Ibarrola, N. Deshpande, K. Shanker, H. N. Shivashankar, B. P. Rashmi, M. A. Ramya, Z. Zhao, K. N. Chandrika, N. Padma, H. C. Harsha, A. J. Yatish, M. P. Kavitha, **M. Menezes**, D. R. Choudhury, S. Suresh, N. Ghosh, R. Saravana, S. Chandran, S. Krishna, M. Joy, S. K. Anand, V. Madavan, A. Joseph, G. W. Wong, W. P. Schiemann, S. N. Constantinescu, L. Huang, R. Khosravi-Far, H. Steen, M. Tewari, S. Ghaffari, G. C. Blobel, C. V. Dang, J. G. Garcia, J. Pevsner, O. N. Jensen, P. Roepstorff, K. S. Deshpande, A. M. Chinnaiyan, A. Hamosh, A. Chakravarti and A. Pandey (2003). "Development of human protein reference database as an initial platform for approaching systems biology in humans." Genome Res **13**(10): 2363-2371.
- Peri, S., J. D. Navarro, T. Z. Kristiansen, R. Amanchy, V. Surendranath, B. Muthusamy, T. K. Gandhi, K. N. Chandrika, N. Deshpande, S. Suresh, B. P. Rashmi, K. Shanker, N. Padma, V. Niranjana, H. C. Harsha, N. Talreja, B. M. Vrushabendra, M. A. Ramya, A. J. Yatish, M. Joy, H. N. Shivashankar, M. P. Kavitha, **M. Menezes**, D. R. Choudhury, N. Ghosh, R. Saravana, S. Chandran, S. Mohan, C. K. Jonnalagadda, C. K. Prasad, C. Kumar-Sinha, K. S. Deshpande and A. Pandey (2004). "Human protein reference database as a discovery resource for proteomics." Nucleic Acids Res **32**(Database issue): D497-501.

Talks

- **Invited Speaker -Minal Menezes** -Blue Babies: How Do They Survive? IARS (International Anesthesia Research Society) 2018 Annual Meeting –Chicago USA
- **Minal J. Menezes**, Yiran Guo⁷, Jianguo Zhang, Lisa G. Riley, Sandra T. Cooper, David R. Thorburn, Jiankang , Daoyuan Dong, Zhijun Li, Joseph Glessner, Ryan L. Davis, Carolyn M. Sue, Stephen I. Alexander, Susan Arbuckle, Paul Kirwan¹, Brendan J. Keating, Xun Xu, Hakon Hakonarson, and John Christodoulou
Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic academia
SSIEM Scientific meeting 2015 (selected for oral), Lyon, France
- **Minal J. Menezes**, Yiran Guo⁷, Jianguo Zhang, Lisa G. Riley, Sandra T. Cooper, David R. Thorburn, Jiankang , Daoyuan Dong, Zhijun Li, Joseph Glessner, Ryan L. Davis, Carolyn M. Sue, Stephen I. Alexander, Susan Arbuckle, Paul Kirwan¹, Brendan J. Keating, Xun Xu, Hakon Hakonarson, and John Christodoulou
Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic academia. HGSA 36th Annual Scientific Meeting 2015, Perth, Australia
- **Minal. Menezes**, M. Menezes, L. Riley, S. Cooper, J. Christodoulou.
The Need for Careful diagnosis of Congenital Myasthenic Syndromes with Secondary Complex I Disorders.
HGSA 36th Annual Scientific Meeting 2013, Queenstown, New Zealand
- **M. Menezes**, L. Riley, S. Cooper, V. Lukic, M. Bahlo, D. Thorburn, J. Christodoulou
The Whole Exome Sequencing Approach to Identify Novel Genes in Mitochondrial Respiratory Chain Disorders
HGSA 36th Annual Scientific Meeting 2012, Canberra, Australia

Abstracts for Poster presentations

- Andrew D Weatherall, Matt D Rogerson, **Minal J Menezes**, Professor Paul G McMenamin, Michael G Cooper, Justin W Adams
Developing novel paediatric airway models: 3D printing, casting and hybrid modelling. IARS 2017 Washington DC USA
- **Minal Menezes**, Yiran Guo, Lisa G Riley, Sandra T. Cooper, David R Thorburn, Xun Xu, John Christodoulou
Mutation of a mitochondrial ribosomal protein gene causes progressive renal failure and congenital sensorineural deafness
EUROMIT 2014 Scientific Meeting, Tampere, Finland
- **Minal J. Menezes**, Manoj P. Menezes, Lisa. Riley, Sandra. Cooper, John. Christodoulou.

- The Need for Careful Diagnosis of Congenital Myasthenic Syndromes with Secondary Complex I Disorders
United Mitochondrial Disease Foundation (UMDF) 2013, LA, USA
- **Minal Menezes**, Lisa Riley, Sandra Cooper, David Thorburn, John Christodoulou.
Transdifferentiation of Patient Fibroblasts for the Study of Tissue Specific Mitochondrial Respiratory Chain Disorders
12th International Conference of Human Genetics Montreal Canada (2011)
 - **Minal Menezes**, Lisa Riley, Sandra Cooper, David Thorburn, John Christodoulou.
Can MyoD-mediated myogenesis help unmask muscle specific Mitochondrial respiratory chain disorders?
2012 Human Genome Meeting (HGM) Sydney, Australia
 - **Menezes M**, Riley LG, Cooper ST, Tzagoloff A, Mina K, Davis M, Thorburn D, Christodoulou, J.
Whole exome sequencing reveals a novel gene associated with complex III deficiency
2012 Aussie Mit Conference, Melbourne, Australia

REFERENCES

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 Dept of Paediatrics, University of Melbourne
 Murdoch Childrens Research Institute, The Royal Children's Hospital ,
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