Genetics and Metabolism Unit
Department of Paediatrics, University of Malaya Medical Centre

1. Staff members
   1. Professor Dr Thong Meow Keong (Head)
   2. Dr Premala Muthukumarasamy
   3. Ms Rifhan Azwani bt Mazlan (Associate Genetic Counsellor)

2. Services provided
   1. Birth defects and genetic diseases in newborns, children and adults – diagnosis and management
   2. Inborn errors of metabolism – multi-disciplinary management
   3. Expanded newborn screening programmes
   4. Adult genetics, reproductive genetics, cancer genetics services
   5. Genetic counselling service
   6. Clinical trials and collaborative research

3. Current Research
   1. Prader-Willi syndrome and Silver-Russell syndrome project
   2. Metabolomics study in infants
   3. Rare diseases: novel therapies
   4. Long term survival of children with birth defects
   5. Expanded newborn screening for inherited metabolic diseases

4. Research funding
   2. Co-Investigator. Study of Uniparental Disomy (UPD) and Genomic Imprinting among Patients with Beckwith-Wiedemann Syndrome and Russell-Silver Syndrome in Malaysia. UMRG grant RG427/12HTM
   3. Principal Investigator: Study of micro ribonucleic acid (miRNA) patterns and messegner RNA splicing among myotonic dystrophy (RM1) patients of different ages and clinical features. Fundamental Research Grant Scheme (FRGS/1/10/SP/UM/01/16)
   6. Co-Investigator Pulse oximetry screening for critical congenital heart disease 2012-2015 UMRG grant (RG435/12HTM),
5. University / National / International collaborations

1. The University of California (Irvine), United States of America on the project “A Comparison of Perceived Barriers to Healthcare between Malaysian and Californian Patients with Rare Genetic Disorders”.

2. The Malaysia Rare Disorders Society, a non-governmental organization, on improving the quality of life of patients with rare disorders and genetic diseases in Malaysian children

3. The Cancer Research Malaysia and Breast Unit, Department of Surgery University of Malaya on hereditary breast and ovarian cancer syndrome project

4. Department of Genetics, Hospital Kuala Lumpur on various research projects.

5. Human Variome Project and Universiti Sains Malaysia

6. Selected publications


7. Fellows
   1. Ms Emily Qian (26th June 2015 – 7th August 2015; University of California Irvine, USA)

8. Activities / Courses

   1. Dr Premala, Prof Thong and Ms Rifhan submitted an abstract for oral presentation entitled “Wiskott-Aldrich syndrome: counselling issues” at the International Congress on Human Genetics 2016 in Kyoto Japan 3rd – 7th April 2016. The abstract was selected for Travel Award.
   2. Prof Thong chaired the scientific committee for the ‘International Society of Neonatal Screening – Asia Pacific Regional Meeting’ in Penang from 7th – 9th December 2015.
   3. Prof Thong, Dr Premala, Ms Rifhan attended the 11th Asia-Pacific Conference on Human Genetics” in Hanoi, Vietnam from 16th-18th September 2015 and presented a poster “Berardinelli-Seip congenital lipodystrophy and its diagnostic implications” and oral presentation “Treatment-focused genetic testing (TFGT)—is it too soon for Malaysia?”. Prof Thong gave the Presidential Address entitled “Personalized and precision medicine: are we there yet?”
   4. A fund-raising charity fashion show was organized by Melinda Looi and a total of RM50,000 was raised and donated to the Genetic Patients’ Welfare Fund of University of Malaya Medical Centre on the 25th May 2015.
   5. “Rare Disease Day” was celebrated by the Genetic Medicine Unit with the Malaysia Rare Disorders Society on the 28th February 2015 at the Department of Paediatrics, University of Malaya Medical Centre.

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