

Dr. Avihu Boneh is the head of the Clinical Metabolic team at the Victorian Clinical Genetics Services (VCGS), Murdoch Childrens Research Institute, Royal Children's Hospital Victoria Australia.

Dr. Boneh graduated from the Hadassah Hebrew University Medical School in Jerusalem, Israel. His MD thesis dealt with Glycogen Synthetase in Diabetes Mellitus. He trained in Paediatrics at Hadassah University Hospital, Jerusalem. He then did a fellowship in Biochemical Genetics with Professor Charles Scriver at Montreal Children's Hospital and graduated from McGill University with a PhD in Experimental Medicine (Biochemical Genetics), investigating X-Linked Hypophosphataemia. He then worked as a consultant in Paediatrics with a special interest in inborn errors of metabolism at Hadassah Mt Scopus hospital, Jerusalem. In 1997, Dr. Boneh took the position of head of Metabolic Genetics at VCGS, MCRI and the Royal Children's Hospital. The VCGS is one of the largest genetics service in Australia delivering genetics service to over 10, 000 Australian families every year. Dr. Boneh is also a member of the Human Genetics Society of Australasia (HGSA) newborn-screening sub-committee, the Victorian Clinical Genetics Services (VCGS) newborn screening advisory committee, the Steering Committee of the European Registry and network for Intoxication type Metabolic Diseases (Scientific Collaborating member) and the International Guideline Group for Glutaric Aciduria type I.

In addition to clinical work and teaching, Dr. Boneh has been very active in various areas of research related to metabolic diseases and has published extensively. His current research projects include:

- 1) early diagnosis and natural history of patients with inherited metabolic disorders
- 2) improving the laboratory diagnosis of inborn errors of metabolism
- 3) adequacy of medically prescribed diets
- 4) role of perturbation of signal transduction in the pathogenesis of disorders: signal transduction is the mechanism by which extra cellular signals exert their intracellular effects. Perturbation of this biological process may act as the pathogenetic mechanism underlying these diseases.

Dr. Boneh is well known internationally and is a great teacher and mentor in the field of inherited metabolic diseases. He has provided high quality metabolic training for many Australasian as well as overseas trainees in metabolic medicine, genetics as well as other related specialties like neurology. He is well known to paediatricians in Hong Kong and has been very supportive to the establishment of metabolic service in Hong Kong. In 2010, Dr. Boneh conducted the first workshop on Paediatric Metabolic diseases for paediatricians, metabolic physicians and pathologists in Hong Kong. The workshop was well attended and received good feedback comments from all who attended. He continues to provide support and invaluable advice on the management of a number of the local metabolic patients.