CURRICULUM VITAE

PROF. DR. SUFIN YAP

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NATIONALITY

Irish

MEDICAL SCHOOL

1982-1988: Royal College of Surgeons in Ireland

SUMMARY OF ACADEMIC QUALIFICATIONS

- MB, BCh, BAO (NUI), LRCPI, LRCSI. – 1988
- DCH (RCP&SI) – 1995
- MD (RCSI/NUI) – 2000

PROFESSIONAL QUALIFICATION

- FRCPCH – 2004

FULL MEDICAL REGISTRATION

General Medical Council (Full & Specialist Register-Paediatrics) Reg. No. 7034375
Irish Medical Council Reg. No. 12742
Irish Medical Council Paediatrics Specialist Register – since July 2003; Certificate No. 465409
Malaysian Medical Council Reg. No. 27980
National Specialist Register (General Paediatrics & Clinical Genetics), Malaysia since 2009

CURRENT POST

Consultant Paediatrician in Metabolic Medicine

Department of Inherited Metabolic Diseases,
Sheffield Children's Hospital, NHS Foundation Trust,
Sheffield,
United Kingdom.

SUMMARY OF EDUCATION

- ***Undergraduate:***

Completed 6 months of a Pharmacy Degree at South Australia Institute of Technology, Adelaide, Australia, prior to medical school.

Medical Degree: MB, BCh, BAO(NUI), LRCPI, LRCSI;
Royal College of Surgeons in Ireland.

Honours in Medicine and Surgery (Third Med), Ophthalmology, Psychiatry and Pathology.

Electives: University Hospital, Kuala Lumpur, Malaysia (teaching hospital for University Malaysia) - completed 1 month of general medicine.

St. Columcille's Hospital, Dublin, Ireland
– completed 6 weeks of general medicine and 2 weeks of general surgery.

- ***Post-Graduate Diploma:*** DCH (RCS&PI) - 1995
- ***Post-Graduate Degree:*** MD (RCSI/NUI) - 2000
- ***Professional Qualification::*** FRCPCH - 2004
- ***Internship***
  1989 – 1990 (Medicine, Paediatrics, Neonates, Surgery, Orthopaedics, Paediatric/Neonatal Surgery)
  Hospital Kuala Lumpur, Malaysia.
- ***Haematology Training:***
  1990 – 1994 National Blood Services Centre
  Hospital Kuala Lumpur, Malaysia.
- ***Paediatric Training:***
  July 1994 – Dec 1994 The Children's Hospital, Temple Street, Dublin.
  Jan 1995 – June 1995 National Maternity Hospital, Dublin
  July 1995 – Dec 1995 Our Lady's Hospital for Sick Children, Crumlin, Dublin.
  Jan 1996 – Dec 1996 The Children's Hospital, Temple Street, Dublin.
- ***Sub-Speciality Training in Inherited Metabolic Disorders/Paediatrics:***
  Jan 1997 – Dec 2001 National Centre for Inherited Metabolic Disorders
  The Children's Hospital, Temple Street, Dublin.
- ***Doctorate in Medicine*** awarded without corrections November 8, 2000 by The Royal College of Surgeons in Ireland/National University of Ireland (Academic year 1999-2000)

  Early detection and commencement of dietary treatment alters the morbid natural history of patients with severe hyperhomocysteinaemia due to cystathionine β-synthase deficiency.

  Supervisor: Dr. Eileen Naughten Advisor: Prof. Denis Gill

This thesis summarises my work on defining the effectiveness of early detection and commencement of treatment in preventing major complications in patients with the inherited metabolic disorder, Homocystinuria, particularly the prevention of vascular events. It also defines a level of biochemical control necessary for good clinical outcome in 25 years of national newborn screening in Ireland. Gene-gene interactions in vascular risks in both homozygotes and obligate heterozygotes were also documented. Data from a multicentre observational study further strengthens the benefits of homocysteine lowering treatment in reducing vascular risk.

SUMMARY OF POSTS HELD

	POST	ON-CALL ROTA
October 2009 - September 2012	***Professor of Paediatrics (Inherited Metabolic Disorders) and Consultant Metabolic Paediatrician*** Department of Paediatrics, Faculty of Medicine and University Malaya Medical Centre University Malaya, Kuala Lumpur, Malaysia.	1 in 1 (Inherited Metabolic Disorders) 1 in 12 (Gen Paeds)
January 2002 – June 2007	***Consultant Paediatrician with special interest in Inherited Metabolic Disorders*** National Centre for Inherited Metabolic Disorders, The Children's Hospital, Temple Street, Dublin and Our Lady's Hospital for Sick Children, Crumlin, Dublin	1 in 1 to 1 in 2 (Inherited Metabolic Disorders)
July 1999 – Dec 2001	***Clinical and Research Registrar*** National Centre for Inherited Metabolic Disorders, The Children's Hospital, Temple Street, Dublin ▪ Inherited Metabolic Disorders (Adult, Paediatric, Neonates)	1 in 2 (Inherited Metabolic Disorders)
January 1997 – June 1999	***Clinical Registrar*** National Centre for Inherited Metabolic Disorders, The Children's Hospital, Temple Street, Dublin ▪ Inherited Metabolic Disorders (Adult, Paediatric, Neonates)	1 in 2 to 1 in 3 (Inherited Metabolic Disorders)
January 1996 – December 1996	***Paediatric Medical Registrar*** The Children's Hospital, Temple Street, Dublin ▪ Metabolic Medicine (Adult, Paediatric, Neonates)	1 in 4 to 1 in 5 (General Paediatrics)
July 1995 – December 1995	***Paediatric Senior House Officer*** Our Lady's Hospital for Sick Children,Crumlin, Dublin ▪ General Paediatrics, Rheumatology, Neurology and Accident & Emergency	1 in 3 to 1 in 4 (General Paediatrics)
January 1995 – June 1995	***Paediatric Senior House Officer*** National Maternity Hospital, Holles Street, Dublin ▪ Neonatology	1 in 2 to 1 in 3 (Neonates)
September 1994 – December 1994	***Paediatric Senior House Officer*** The Children's Hospital, Temple Street, Dublin ▪ Accident & Emergency	1 in 2 to 1 in 3 (General Paediatrics)
June 1994 – August 1994	***Paediatric Senior House Officer*** The Children's Hospital, Temple Street, Dublin ▪ Metabolic Medicine and Cardiology	1 in 2 to 1 in 3 (General Paediatrics)
February 1990 – June 1994	***Haematology Medical Officer*** Hospital Kuala Lumpur, Malaysia (Adults, Paediatric, Neonates)	1 in 3 to 1 in 4
February 1989 – January 1990	***Internship*** Hospital Kuala Lumpur, Malaysia	1 in 2 to 1 in 3

INTERNSHIP (1989)

HOSPITAL KUALA LUMPUR, MALAYSIA (1 year)
Teaching hospital for the National University of Malaysia.
2500 bedded National Referral Centre for Malaysia and 150 bedded Institute of Paediatrics.

General Medicine, 5 months (On call rota: 1 in 2 to 1 in 3)

- An extremely busy firm (350 beds) with 50-70 acute admissions on-take every 1 in 3.
- Initial diagnosis and management of all emergency medical cases and day-to-day management of ward patients.
- Diagnostic and therapeutic procedures performed including phlebotomy, arterial blood gas, lumbar puncture, bone marrow and trephine biopsies insertion of central venous pressure lines, pleural biopsy.

Paediatrics/ Neonatology (1 month): (On call rota: 1 in 2 to 1 in 3.)

- Initial diagnosis and management of all emergency paediatric cases e.g., seizures, severe dehydration, meningitis with cerebral abscesses.
- Experience in neonatal exchange transfusions, resuscitation and ventilation.

General Surgery (3 months); Orthopaedics (2 months) (On call rota 1 in 3)

- Initial diagnosis and management of all adult and paediatric emergency surgical, trauma & orthopaedic cases.
- Pre and post-operative care for all surgical, trauma & orthopaedic patients.
- Performing minor surgical procedures e.g.. Appendicectomy, removal of lipomas, incise & draining of abscesses, toilet & suturing of wounds, removal of ingrown toe nails, etc.
- Assisted in surgical and orthopaedic procedures in theatre including joint replacement, spinal surgery, gall bladder surgery, laparotomies, repair of severed tendons, etc.
- Acute management of patients admitted to a busy Casualty Ward.

Paediatric/Neonatal Surgery (1 month): (In-house daily on call for 1 month)

- All paediatric surgical cases were referred to Hospital Kuala Lumpur as it is the only paediatric surgical unit in the country.
- Acute pre, peri and post operative management of **all neonatal surgical patients**, including ventilation, fluid management, antibiotic management, the use of total parenteral nutrition, umbilical venous and arterial line access
- Part of the team for conjoint twin separation.
- Assisted in all neonatal surgical procedures e.g. release of pyloric tumours, repair of diaphragmatic hernia, repair of inguinal hernias, insertion of shunts for hydrocephalus.

HAEMATOLOGY TRAINING (1990-1994)

Hospital Kuala Lumpur, Malaysia. (4.5 years)
Teaching hospital for the National University of Malaysia.

- Worked in a very busy specialised national referral centre for clinical and laboratory haematology, transfusion medicine, virology (hepatitis B, C, D and HIV), coagulopathies and blood banking.
- It is the Central Registry for Coagulation bleeding disorders and is the reference for bleeding problems and Haemophilias.
- The centre is one of the few accredited foreign training centre for the Fellowship of the Royal College of Pathology Australasia (FRCPA) and for medicine (MRCP, UK and MRCPI) and pathology (MRCPath, UK – Haematology).
- Initially started training as medical officer (equivalent to SHO for 6 months and Registrar for 1.5 year) under the direct supervision of Prof. G Duraisamy.
- My final 2.5 years of training (equivalent to a senior specialist registrar) included Consultant level of responsibilities in all aspects of clinical and laboratory haematology, transfusion medicine, blood banking and transfusion transmitted diseases. Full responsibilities especially for Paediatric Leukaemias and all Paediatric (including neonatal) transfusion medicine.
- Provided on call cover (including consultant level cover during Prof Duraisamy's leave) principally for Hospital Kuala Lumpur, which essentially included referral cover for the whole country (on call rota: 1 in 3 to 1 in 4).

Paediatric Leukaemias **(2.5 years):**

- Investigation, diagnosis, care and follow-up of leukaemia patients with full responsibilities for all paediatric haematology, particularly paediatric leukaemia at the Institute of Paediatrics, Hospital Kuala Lumpur for 2.5 years as the equivalent of a senior registrar with consultant level of responsibilities.
- Training includes interpretation of peripheral blood films, bone marrows and trephine biopsies in the initial diagnosis and subsequent follow-up of the patients through chemotherapy and remission. Understanding the effects of chemotherapy and predicting time related potential side effects of the particular drug treatment and monitoring for them. Appropriate use of blood and blood products to prevent bleeding diathesis during chemotherapy. Diagnosis of relapse clinically and confirmation on bone marrow and trephine biopsies. Follow –up and management of relapses.

PAEDIATRIC TRAINING (JUNE 1994 – DECEMBER 1996)

THE CHILDREN'S HOSPITAL, TEMPLE STREET, DUBLIN

Senior House Officer (June 1994-December 1994)
With Registrar responsibilities when they are on leave (General paediatrics on call rota of 1 in 3)

- **Inherited Metabolic Disorders**
  - Consultant: Dr. E. Naughten

- **Cardiology**
  - Consultant: Dr. D. Duff

- **Accident & Emergency**
  - Consultant: Dr. P. Keenan

NATIONAL MATERNITY HOSPITAL, HOLLES STREET, DUBLIN

Senior House Officer (January 1995 – June 1995)
With Registrar responsibilities when they are on-leave or the neonatal intensive care unit is extremely busy. (On call rota: 1 in 2 to 1 in 3; First call to all neonatal emergencies)

- **Neonatology**
  - Consultant: Dr. W. A. Gorman, Dr. J. Murphy, Dr. N. O'Brien

OUR LADY'S HOSPITAL FOR SICK CHILDREN, CRUMLIN, DUBLIN

Senior House Officer (July 1995 – December 1995)
With Registrar responsibilities when they are on-leave.
(General paediatric on call rota: 1 in 3 to 1 in 4, includes covering Accident & Emergency)

- **General Paediatrics** (on take once a week)
- **Rheumatology**
- **Neurology**
  - Consultant: Dr. H. Monaghan

THE CHILDREN'S HOSPITAL, TEMPLE STREET, DUBLIN

Paediatric Medical Registrar (January 1996 – December 1996)
(General paediatric on call rota: 1 in 4 to 1 in 5)

- **Inherited Metabolic Medicine/General Paediatrics**
  - Consultant: Dr. E. Naughten, Dr. P. Thornton

SUB-SPECIALITY TRAINING IN INHERITED METABOLIC DISORDERS/PAEDIATRICS (1997 - 2001)

National Centre for Inherited Metabolic Disorders, The Children's Hospital, Temple Street, Dublin. (Inherited Metabolic Medicine on call rota 1 in 2 to 1 in 3)

- **With Consultant responsibilities working under close and direct supervision of the Consultant.**

CLINICAL REGISTRAR (JANUARY 1997-JUNE 1999)

(Inherited Metabolic Medicine on call rota 1:2)

- Consultant: Dr. E. Naughten, Dr. P. Thornton

Responsibilities:

Worked 5 busy outpatient clinics per week for patients with a wide spectrum of metabolic conditions. Involves also routine neonatal and paediatric developmental and growth assessment and monitoring.

Participated in daily Consultant-led ward rounds.

Providing medical care for episodes of acute metabolic decompensation due to intercurrent illnesses.

NEONATES: Acute intensive management of all neonates with metabolic problems. Involves intensive management of fluid, electrolytes, blood gas monitoring for ventilation, antibiotic therapy, use of insulin, use of specialised therapeutic treatment as indicated by metabolic condition, peritoneal dialysis for the removal of toxic metabolites, specialised nutritional treatment as indicated by metabolic condition, etc. (First on-call with consultant responsibilities for all neonatal acute intensive management with close and direct supervision of Consultant.)

Liasing management of neonates in peripheral hospital for transfer to this centre for further management.

Management of minor general paediatric intercurrent illnesses over the phone with parents and general practitioners.

Performing diagnostic procedures e.g. skin biopsy, low dose Dexamethasone tests, fasting studies, etc.

Monitoring results arising from investigations and discussing with Consultants for further appropriate management as indicated.

ADULTS: Acute intensive metabolic management of all known adults with inherited metabolic disorders admitted to other hospitals including;

- Mater Hospital, Dublin (ICU and ward)
- Beaumont Hospital, Dublin (ICU and ward)

Monitoring biochemical control of patients, in particular the 40 patients with classical homocystinuria.

Providing counselling for the different metabolic conditions.

Monitoring Maternal Phenylketonuria patients and liasing with maternity hospital re: High-risk screen on newborns.

Liasing with General Practitioners and staff of peripheral hospitals on the care of metabolic patients.

Providing cover for junior registrar and SHO for inherited metabolic disorders

Participating in Case Conferences, Journal Clubs and publication of scientific papers.

Giving lectures and tutorials to medical students.

Attending weekly Grand Rounds.

Training:

Training in clinical diagnosis, work-up, both acute and outpatients/home management and treatment of patients. Emergency/ crisis management of:

Aminoacidopathies, Organic acidurias, Hyperammonaemia and urea cycle defects, Disorders of carbohydrate metabolism, Fatty acid oxidation defects, Lysosomal storage disorders, Disorders of lipid metabolism, Peroxisomal disorders, Mitochondropathies.

Training in interpretations of amino acid, urinary organic acid, and fasting study profiles.

Training in normal requirements of protein, energy, vitamins and minerals for normal growth and development.

Training in the principles of dietary treatment in inherited metabolic medicine; consequences of changes in nutrient intakes, under-nutrition and specific nutritional deficiencies.

Training in the indications for, planning and interpretation of assays arising from: e.g. skin biopsies for fibroblast cultures and enzyme assays, liver biopsy, muscle biopsy.

Interpretation of psychological and neuropsychological testing.

Training in the appreciation of the patient and families understanding of metabolic disorder and the psychological stress of a metabolic diagnosis.

Training in working as a part of a multidisciplinary team.

CLINICAL AND RESEARCH REGISTRAR (JULY 1999 TO DECEMBER 2001)

- Consultant: Dr. E. Naughten, Dr. E. Treacy

(1 research : 3 clinical time; On-call during research time rota: 1 in 2)

Responsibilities:

Consultant level of responsibilities for three outpatient clinics per week.

Includes all of the above responsibilities held as the clinical registrar with increasing independent responsibilities at consultant level. All decisions made independently were subsequently discussed with the Consultant.

NEONATES: Consultant level of responsibilities for all acute management of neonates with metabolic referral. Provided in house on-call management for these neonates in intensive care at other hospitals including:

- ICU, Our Lady's Hospital for Sick Children, Crumlin, Dublin,
- Neonatal ICU, Rotunda Hospital, Dublin,
- Neonatal ICU, Coombe Women's Hospital, Dublin.

Liased management with neonatal registrar and SHO to stabilise neonates in peripheral hospital for transfer to the National Centre for further management of metabolic disorder.

Specific involvement in the diagnosis, management, long term-follow-up and biochemical control of the 40 patients with classical homocystinuria attending this centre under the supervision of Dr. Naughten

ADULTS: Consultant level of responsibility for all acute metabolic management of all known adults with inherited metabolic disorders admitted to other hospital including;

- Mater Hospital, Dublin (ICU and ward)
- Beaumont Hospital, Dublin (ICU and ward)

Provided in-house cover and management of these patients at the above hospitals.

Did night and weekend ward rounds for all inherited metabolic patients when on-call.

Research component as detailed in the "Doctorate of Medicine Thesis section".

Academic/Research Training:

Training in molecular biological techniques for mutational analyses.

Training in setting up of study protocols, data collection and interpretation.

Training in leading and co-ordinating international multicentre study including data collection and interpretation

Training in computerised data processing.

Appreciation and understanding of statistical methods used for analyses of data.

Training and research leading to the degree of Doctor of Medicine.

FURTHER TRAINING IN INHERITED METABOLIC DISORDERS/ PAEDIATRICS

1st Orphan Europe Academy course on Inborn Metabolic Diseases.
Dept of Paediatrics 1, University Children's Hospital, Heidelberg, Germany.
October 10-14, 2001
One of 33 internationally selected participants to attend based on submission of C.V.

EXPERIENCE IN FOREIGN CENTRES OF INHERITED METABOLIC DISORDERS

University College London Hospital, The Middlesex Hospital, Mortimer Street, London, U.K. – March 1999 (Supervisor: Dr. P.J. Lee)

Reviewed medical notes of 41 patients with homocystinuria

- Collection and analysis of clinical data.
- Collection and analysis of biochemical data.

The Biochemical Genetics Department, The Montreal Children's Hospital, Montreal, Canada – May 2001 (Supervisor: Dr. E.P. Treacy)

- Invited to present Irish experiences on Homocystinuria and Phenylketonuria at the Biochemical Genetics Grand rounds.
- Attended General Paediatrics Grand rounds.
- Attended Nephrology-genetics and Biochemical-genetics clinics
- Visited the Neonatal and Paediatric ICU; Tandem Mass Spectrometry Laboratory
- Participated in discussions and Research Round with Dr. D. Rosenblatt at Royal Victoria Hospital – Montreal.
- Laboratory for Tay-Sach's screening and white cell cysteine measurements for cystinosis.
- Discussions on the screening programs for β-thalasseamia in Montreal.

Hospital Necker Enfant Malades, Paris, France - March-April 2002 (Supervisor: Prof. J.M. Saudubray)

- Attended all daily ward rounds, consultants clinics and weekly clinico-laboratory/dietetic meeting on all laboratory results.
- Reviewed management and treatment protocols of metabolic disorders.
- Observed percutaneous pancreatic venous and arterial sampling on patients with hypersulinism under **Prof. Brunelle,** radiologist.
- Visited the mitochondrial diagnostic laboratories - **Dr. P. Rustin**

- I was invited to present my work on the treatment and long term outcome of patients with Homocystinuria at this centre. This provided an opportunity for further discussion on the management of these patients and future research into the area to improve patient care.

- 21st March 2002 - Invited to present my work on Homocystinuria at the *British Inherited Metabolic Disease Group & Association of Clinical Biochemist Meeting at Doncaster, UK.*

- 5-6th April 2002 - Invited to attend and participate at the *Annual Metabolic SOS Meeting, Fountaineblau, France.* Cases with diagnostic and treatment difficulties were discussed at this meeting attended by about 30 European experts in the field.

Harvard Medical School, Boston, USA Massachusetts General Hospital, Boston, USA - May 2002 (Supervisor: Prof. V. Shih, Professor of Paediatric Neurology (Harvard) and Director of Amino Acid Disorder Laboratory/Metabolic Disorders Unit (Massachusetts)

- I also attended other metabolic/genetic clinics held by the following:
  Prof. Harvey Levy
  Sr. Associate in Medicine and Genetics, Children's Hospital, Boston and Associate Professor of

Paediatrics, Harvard Medical School of Medicine, Boston, USA.

Dr. D. Marsden
Director of Metabolism Program, Children's Hospital, Boston, USA.

Dr. M. Ampola
Chief, Division of Metabolism and Director, PKU-Inborn Errors of Metabolism Clinic, Clinical Genetics/Genetic Counselling Clinic, Tufts-New England Medical Center, Tufts University School of Medicine, Boston, USA.

Dr. M. Korson
Associate Chief, Division of Metabolism and Associate Professor of Paediatrics, Tufts-New England Medical Center, Tufts University School of Medicine, Boston, USA.

- Involved in clinical laboratory discussions/meetings, including interpretations of amino acids and organic acids profiles at the **State Laboratory Institute, New England Newborn Screening Program, University of Massachusetts, Boston, USA.**
- Invited to present my work on Homocystinuria and provided some consultations on patients with Homocystinuria who attended these centres.

John Hopkins University & Division of Metabolism, Kennedy Krieger Institute, Baltimore, Maryland, USA - May 2002 (Supervisor: Prof. R.I. Kelley, Associate Professor of Paediatrics, John Hopkins University and Director, Kennedy Krieger Institute

- Attended multiple clinics and conferences and reviewed the standard protocols for diagnosis and treatment of inborn errors of metabolism.
- Contributed to discussion on the management of pregnancy in patients with Homocystinuria.
- Also attended Dr. H. Morton's weekly clinic and laboratory at the **Clinic for Special Children, Lancaster, Maryland, USA**.

Nijmegen Centre for Mitochondrial Disorder and Metabolic Diseases, University Hospital Nijmegen, The Netherlands - June-July 2002 (Supervisor: Prof. J. Smeitink)

- Attended all clinics, ward rounds and clinico-laboratory diagnostic meetings.
- Observed laboratory preparations of muscle biopsies and assays for mitochondrial disorders.
- Participated in the weekly clinico-laboratory discussions on results of amino acids, organic acids and muscle biopsies.
- Reviewed investigative protocols for mitochondrial disorders.
- Attended metabolic clinical round table meeting with paediatricians at a peripheral hospital in Armalo, where the metabolic service was covered by this centre.
- Also attended **Dr. G. Boers'** weekly adult metabolic clinics and Marfan polyclinics. Observed echocardiography in the assessment of these patients. Provided input and discussion for the future of research into the area of homocysteine metabolism.

- Attended the *7th International Symposium on Mucopolysaccharide and Related Diseases and the 3rd Scientific Lysosomal Storage Disorders Congress*, Paris, France.

Institute of Child Health and Great Ormond Street Children's Hospital, London, UK - July-August 2002 (Supervisor: Prof. James Leonard, Professor of Paediatric Metabolic Medicine and Prof. Peter Clayton, Professor of Hepatology and Metabolism)

- Attended all ward rounds, clinics, consults and unit meetings.
- Reviewed management and treatment protocols for metabolic disorders.
- Reviewed treatment and management protocols for hyperinsulinism (endocrine dept.).
- Also attended the adult metabolic service/clinic by **Dr. P. Lee** at *the Hospital of Neurology and Neurosurgery, Queen Square, London, UK.*

- During this time, I also provided a two week on-call cover for The Children's University Hospital, Temple Street, Dublin.

METABOLIC CONSULTANT POST (JAN 2002 – JUNE 2007)

National Centre for Inherited Metabolic Disorders, Children's University Hospital, Temple Street, Dublin, Ireland, and
Our Lady's Hospital for Sick Children, Crumlin, Dublin, Ireland.
Teaching hospitals for the Medical Schools of the Royal College of Surgeons in Ireland, University College Dublin and Trinity College Dublin.

National Centre for Inherited Metabolic Disorders (NCIMD), Children's University Hospital

- This is the only tertiary referral centre for the investigation and treatment of inherited metabolic diseases (IMD) linked to the newborn screening program in Ireland.
- Catchment area: 1.4 million population from county Dublin, and
- 4.1 million total Irish population
- The Centre comprises of: a dedicated outpatients department and
- 7-beded high dependency metabolic inpatient ward.
- Over 1500 patients nationwide, with a wide variety of inherited metabolic disorders ascertained by newborn screening and clinical presentations, are followed at the NCIMD for life.
- The delivery of care for all IMD patients in Ireland is delivered entirely from the NCIMD
- After 40 years of screening, the Centre actively follows over 600 patients with PKU, over 100 patients with Galactosaemia, 40 with Homocystinuria and 14 patients with MSUD.
- About 45% of the patients followed at the Centre are over the age of 16 years.
- In addition to screening, a major proportion of the caseload are suspected IMD cases referred from Paediatricians and Neurologists from the three Paediatric Hospitals and three Maternity Hospitals in Dublin and from all regional hospitals throughout the country for consultation and follow up of confirmed cases.
- The Irish metabolic practice reflects a relatively homogenous isolated population with subsequent higher prevalence of specific recessive IMDs than in other population, contributing to the NCIMD having one of the largest cohorts of IMD patients in a single centre worldwide (eg. high incidence of Phenylketonuria, Galactosaemia, Homocystinuria, Glutaric Aciduria type 1 in Travellers and Mitochondropathies.)
- The unique position of the NCIMD and the relatively homogenous Irish population have allowed opportunities for Clinician Investigators at the Centre to conduct health related research and investigated the epidemiology and successful treatment outcomes of many IMDs
- The Centre also provides outreach dietetic and nursing support and advice to Dublin and regional hospitals by telephone.

The NCIMD team consists of: 4 funded Metabolic Consultant posts (only two posts are filled)
2 Specialist Registrars, 2 Registrars and 1 Senior House Officer
3 Administrative staff, 1 Clinic Coordinator
2 Metabolic Nurse Specialist (CNM3)
1 Enzyme replacement nurse coordinator and 1 Staff Nurse
5 Dietitians
2 Clinical Psychologists; 1 Medical Social Worker

The Metabolic Ward is staffed by: 1 Ward Sister (CNM3)
1 Ward Sister (CNM1)
Post graduate Staff Nurses
1 Play Specialist

Clinical Responsibilites: (Inherited Metabolic Medicine on- call rota: 1 in 1 to 1 in 2)

Full personal clinical responsibility for a caseload of over 300 IMD patients and cross cover for all 1300 IMD patients followed at the NCIMD.

Weekly out patients clinics: 1 general IMD clinic
1 Homocystinuria clinic
2 PKU (1 Adult & 1 Paeds) Nurse specialist/dietitian delivered clinics
1 Galactosaemia clinic

- To ensure patient safety in the delivery of a responsive and progressive IMD service
- Lead a collaborative multidisciplinary team in the provision of such a service
- Outpatients new metabolic referral nationwide
- Consultative metabolic phone advise to regional hospitals nationwide on initial investigation and management of suspected cases of IMD.
- Lead multidisciplinary pre-clinic meetings
- Inpatients new metabolic referral and consultations onsite and nationwide (averaging about 3-5 new cases/week)
- All metabolic patients admitted to the high dependency metabolic ward, neonatal ward and PICU.
- Continuing responsibilities for the investigation and for treatment of patients without supervision in professional matters.
- Participate in the 24/7 hospital metabolic consultant on-call rota, which also provides nationwide cover.
- Consultative metabolic service (averaging about 7-8 new referrals/week) to Our Lady's Hospital for Sick Children, Dublin – the largest tertiary Paediatric Hospital in Dublin.
- Honorary metabolic consultant to the National Maternity Hospital, Holles Street, Dublin.
- Act as clinical advisor to the national newborn screening and metabolic laboratory.
- Planning and scheduling metabolic investigative and diagnostic tests for patients
- Interpretation of metabolic function tests results
- Planning, scheduling and updating patient-specific emergency treatment regimens and well day regimes.
- Daily management of biochemical levels (in collaboration with the Dietitians) for:
  Maternal PKU (about 7-8 pregnancies at any one time)
  Homocystinuria,
  Maple Syrup Urine Disease
- Review of amino acids profiles and other biochemical markers in routine managements of patients with organic acidurias, aminoacidopathies, fatty acid oxidation defects, urea cycle defects and others.
- Participate in weekly clinico-laboratory organic acid profile meetings
- Participate in weekly unit multidisciplinary psycho-social meeting
- Counselling newly diagnosed patients and their families.
- Liasing acute care of patients with regional hospitals for stabilisation prior to transfer to the NCIMD

Research and Development:

- Actively participating in the process of clinical audits in collaboration with medical and paramedical staff members.
- Actively encouraging and supporting professional development of team staff members.
- In-service teaching and training of medical staff and other staff.
- Participating in active collaboration with colleagues locally and internationally for investigative and diagnostic procedures and tests.
- Active frequent exchange, updating and sharing of management and treatment experiences between colleagues at both local and international levels.
- Leading clinical research in the area of IMD – refer to section on "active research in progress".
- Actively involved in the education of colleagues and the public on metabolic disorders
- Participate in the planning for future development of the IMD service with patient care being central.
- Active involvement in Continued Medical Education (CMD) and Continued Professional Development (CPD)

Development of a dedicated clinic for patients with Homocystinuria:

Assisted by a nurse specialist to coordinate care.
Bimonthly Paediatric clinic and monthly adult clinic
Coordinates annual ophthalmology review and Dexa scans
Development of a quarterly joint cardiac and neurology clinic

Development of diagnostic and management guidelines for the treatment of Homocystinuria

Development of education materials/booklets on Homocystinuria for the patient (child and adult), their parents or carers and physicians.

Development of a comprehensive intranet system for elective inpatients.
This system allows:

- The ward sister to inform me, a week in advance, of all elective admissions of patients.
- The preparation and planning of patient-specific baseline and metabolic function tests and other specialists consultations for the admission in advance by the Consultant.
- Each patient-specific elective admission plan is then posted in a specific folder in the intranet whereby ward staff members can assessed readily on the Ward computer system in preparation for the admission.

This system has worked effectively and efficiently in improving the organisation of elective admissions in a very busy clinical practice.

Managerial and Administrative Responsibilities/ Experience:

The appointment allows for a two to three years rotational Clinical Directorship of the NCIMD with other Metabolic Consultants of the Centre.
Participate in the selection process for Non-Consultant Hospital Doctors
Participate in the selection process for such other staff as the employing authorities agree are appropriate
Participate in the administration outside the management of the NCIMD of the Hospital.

PROFESSOR OF PAEDIATRICS (INHERITED METABOLIC DISORDERS) and CONSULTANT METABOLIC PAEDIATRICIAN POST (MAR 2010 – AUG 2012)

Department of Paediatrics and University Malaya Medical Centre,
Faculty of Medicine,
University Malaya.

Clinical Responsibilites: (General Paediatric on- call rota: 1 in 12)

General Paediatrics and Neonates:

- Consultant in-charge of paediatric/neonatal ward P2
    - 26 bedded ward
    - 3 Interns; 4 Medical Officers; 2 Lecturers

Inherited Metabolic Disorders:

- To develop a safe and progressive IMD service
- To ensure patient safety in the delivery of a responsive and progressive IMD service
- Re-assessment of the diagnoses of known IMD patients
- Inpatients new metabolic referral and consultations onsite
- Planning and scheduling metabolic investigative and diagnostic tests for patients
- Interpretation of metabolic function tests results
- Planning and scheduling of appropriate treatment regimens for newly diagnosed patients
- Planning, scheduling and updating patient-specific emergency treatment regimens and well day regimes.
- Review of amino acids profiles and other biochemical markers in routine managements of patients with organic acidurias, amino-acidopathies, fatty acid oxidation defects, urea cycle defects and others.
- Scheduling and liasing with foreign centres for confirmatory enzymology and molecular testing.
- Screening of siblings of newly diagnosed index case and interpretation of screening results.
- Lead multidisciplinary pre-clinic meetings
- Weekly Outpatients service
    - Outpatients new metabolic referral nationwide
    - Outpatients follow-up of all IMD patients
- Participate in weekly clinico-laboratory organic acid/ amino acid profile meetings
- Advice on Newborn Screeining to Laboratory
- Providing expert IMD and homocystinuria advice locally and internationally

Training and Teaching:

- General Paediatrics teaching for both under and post-graduates
- Supervision of 1 Master of Paediatrics and 1 Master of Science candidates
- Teaching and training of paediatric dietician in Paediatric Metabolic Dietetics
- Provision of IMD examination questions
- Developing an IMD teaching module for both under and post-graduates.
- Training of IMD biochemist – HKL
- Development of a IMD training course for IMD Paediatricians in the Asia-Pacific region
    - **Inaugural Asia Pacific Metabolic Course Kuala Lumpur - October 2011**

- Teaching and training of laboratory colleagues in organic acid and amino acid profile interpretation.

Research and Development:

- Actively participating in the process of clinical audits in collaboration with medical and paramedical staff members.
- Actively encouraging and supporting professional development of team staff members.
- In-service teaching and training of medical staff and other staff.
- Participating in active collaboration with colleagues locally and internationally for investigative and diagnostic procedures and tests.
- Active frequent exchange, updating and sharing of management and treatment experiences between colleagues at both local and international levels.
- Leading clinical research in the area of IMD – refer to section on "active research in progress".
- Actively involved in the education of colleagues and the public on metabolic disorders
- Participate in the planning for future development of the IMD service with patient care being central.
- Active involvement in Continued Medical Education (CMD) and Continued Professional Development (CPD)
- Main member of Biotechnology and Bioproduct Research Cluster – **Genomic research group**

TEACHING EXPERIENCE

UNDERGRADUATE TEACHING:

(HAEMATOLOGY, TRANSFUSION MEDICINE AND BLOOD BANKING, 1990-1994)

- Medical students
- Medical laboratory technician, Institute of Medical Research, Malaysia.

(GENERAL PAEDIATRIC - SINCE 1994 AND INHERITED METABOLIC MEDICINE – SINCE 1996)

Royal College of Surgeons in Ireland

- General Paediatric bedside clinical teaching
- General Paediatric and Inherited Metabolic Medicine outpatient clinical teaching
- Inherited Metabolic Medicine formal lectures since 1999

University College Dublin

- General Paediatric bedside clinical teaching
- General Paediatric and Inherited Metabolic Medicine outpatient clinical teaching
- Inherited Metabolic Medicine formal lectures since 2000

Trinity College, Dublin

- Inherited Metabolic Medicine formal lectures since 2000

University Malaya, Kuala Lumpur

- General Paediatrics and Inherited Metabolic Medicine since 2010

POST-GRADUATE TEACHING:

(HAEMATOLOGY, TRANSFUSION MEDICINE AND BLOOD BANKING, 1990-1994)

- Nurses
- M. Med. (pathology) Students, University Kebangsaan Malaysia
- Training of doctors from peripheral hospitals

(INHERITED METABOLIC MEDICINE – SINCE 1998)

Metabolic workshops for nurses, The Children's Hospital, Temple Street, Dublin – since 1998.

- Formal lectures

Special and intensive care of the newborn course for nurses – National Maternity Hospital, Holles Street, Dublin 2001.

- Formal lecture

MSc in Paediatrics, Trinity College Dublin – since 2000.

- Formal lectures
- Clinical bedside teaching

Metabolic Module for Nurses, Children's University Hospital & Dublin City University – since 1998

- Formal Lectures
- Nursing guidelines for metabolic disorders

Metabolic Module for post graduate Doctors, University Malaya, Kuala Lumpur – since 2010

MEMBERSHIP OF SOCIETIES AND OTHER LEARNED BODIES

- Society for the Study of Inborn Errors of Metabolism (SSIEM) – since 2000
- European Neurological Society (ENS) – since 1999
- European Federation of Neurological Societies (EFNS) – since 2000
- Irish Society of Human Genetics (ISHG) – since 2000
- Faculty of Paediatrics, Royal College of Physicians, Ireland – February 2002
- Royal College of Paediatrics and Child Health, England – December 2004
- Medical Genetic Society Malaysia – April 2010
- British Inherited Metabolic Disease Group (BIMDG)

SCIENTIFIC COMMITTEE

- Member of the Consensus Statement Group for the "Guidelines on the use of Blood and Blood Products
- Planning Committee for the 40th Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Dublin 2002.
- Member of the European Homocystinuria Expert Board – established June 2008.
- Co-Editor of the Inaugural Asia Pacific Metabolic Course, Kuala Lumpur, Malaysia-October 2011
- Participating member of European Network and Registry for Homocystinurias and Methylation Defects – since 2014
- Expert Member of Orphan Europe Advisory Board on Carglumic Acid - 2015

EXAMINER BY INVITATION

- Final Professional Examination for Medicine and Surgery of Childhood, Royal College of Surgeons in Ireland, since 2000.
- Diploma in Child Health, Royal College of Surgeons in Ireland, since 2000.
- Final Professional Examinations for Paediatrics and Child Health, Trinity College, Dublin, since 2000.
- MBBS Phase IIIA clinical examinations, University Malaya - October 2010.
- MBBS Phase IIIB clinical examinations, University Malaya – October 2010.
- Theory Examination for the Conjoint Board (University Malaya/ University Kebangsaan Malaysia/ University Science Malaysia) Master of Paediatrics Part II Examination – November 2010, April 2012

DOCTOR OF MEDICINE (MD) THESIS

Yap S (2000).
Early Detection and Commencement of Dietary Treatment Alters the Morbid Natural History of Patients with Severe Hyperhomocysteinaemia due to Cystathionine β-Synthase Deficiency.
MD Thesis, Royal College of Surgeons in Ireland / National University of Ireland.

- Submitted in October, 2000 to the Royal College of Surgeons in Ireland for the degree of Doctor of Medicine.
- Awarded without corrections in November, 2000 and conferred in May 2001.

My work looked, uniquely, at patients with Classical Homocystinuria in 25 years of national newborn screening in Ireland. Long-term clinical outcomes were determined and a level of biochemical control for good clinical outcome was defined. Intellectual abilities were systematically assessed and compared to the untreated patients and their unaffected sibling controls to determine effectiveness of treatment. The effectiveness of dietary treatment in preventing premature osteoporosis was also studied in detail including calcium and phosphate intakes and excretion. Gene-gene interactions in vascular risks in both homozygotes and obligate heterozygotes were also documented. Data from an international multicentre study further strengthens the benefits of homocysteine lowering treatment in reducing vascular risks.

In summary, this thesis summarises my work in determining long-term clinical endpoints and defining the effectiveness of early detection and commencement of treatment in preventing major complications in patients with homocystinuria as well as defining a level of biochemical control necessary for good clinical outcomes.

Research principally conducted at:
National Centre for Inherited Metabolic Disorders,
The Children's Hospital, Temple Street, Dublin, Ireland.

Collaboration with: Professor M. O'Brien, Osteoporosis Unit, Trinity College Dublin, for work on bone mineral densities.

International Multicentre Collaboration with:

- Professor D Brenton, Postgraduate Centre, Middlesex Hospital, London;
- Dr. GHJ Boers, University Hospital Nijmegen, The Netherlands;
- Dr. P. Lee, The National Hospital for Neurology and Neurosurgery, London,
- Dr. J Walter, Willink Biochemical Genetics Unit, Royal Manchester Children's Hospital, Manchester;
- Professor B Wilcken, The Children's Hospital, Sydney, and
- Professor D Wilcken, University of New South Wales, Sydney, Australia,
  for long-term vascular outcomes and biochemical control of patients with classical homocystinuria.

Clinical Direction and Supervision By:
Dr. Eileen Naughten, DCH, FRCPI, FRCPCH.
Clinical Director, National Centre for Inherited Metabolic Disorders, and Consultant Paediatrician with special interest in Inherited Metabolic Disorders, The Children's Hospital, Temple Street, Dublin, Ireland.

Royal College of Surgeons in Ireland Advisor:
Professor Denis Gill, BSc, DCH, FRCPI, FRCPCH.
Professor of Paediatrics, Royal College of Surgeons in Ireland, and
Consultant Paediatrician, The Children's Hospital, Temple Street, Dublin, Ireland.

University College Dublin Nominating Professor:
Professor Hugh Brady
Professor of Medicine, University College Dublin.

Research Funding:
- Mutational analysis work funded by Irish Society for Inherited Metabolic Disorders (ISIMD).

POST-DOCTORAL ACTIVE RESEARCH IN PROGRESS

- Carotid artery ultrasonography in classical homocystinuria.
- MRI and neurological findings in classical homocystinuria.
- International multicentre collaborative study on the origin of Cystathionine β-Synthase c.833T>C (I278T) haplotype.
- Bone mineral densities in patients with Maple Syrup Urine Disease.
- Long-term outcome in treated patients with Maple Syrup Urine Disease.
- Mutational and complementation analysis of 3-methyl-crotonyl CoA carboxylase gene in Irish patients.
- Mutational analysis and enzymatic studies on pyridoxine nonresponsive Homocystinuria.

ACTIVE RESEARCH IN PROGRESS – MALAYSIA

- Mutational analysis and demographic study of Homocystinuria in Malaysia
- aCGH in Ornithine Transcarbamylase Deficiency
- Dominant negative inheritance of 3 Methyl-Crotonyl- CoA Carboxylase deficiency
- Clinical and molecular spectrum of Cobalamin C Defects in Malaysia
- Molecular demographic of Fanconi-Bickel Syndrome in Malaysian patients.
- Diagnostic pitfalls in Carnitine Transporter Defects

AWARDS AND HONOURS

JUNIOR IRISH PAEDIATRIC ASSOCIATION – ANNUAL NEONATAL MEETING 1995

Best Case Presentation:
Oral presentation:

Yap S*, Gorman WA, Murphy J, O'Brien N.*
Juene Syndrome

FINALIST FOR THE ROYAL ACADEMY OF MEDICINE IRELAND/IRISH JOURNAL OF MEDICAL SCIENCE (RAMI/IJMS) AWARD FOR EXCELLENCE IN CLINICAL RESEARCH 2000:

("Risk Factor Medicine" category with Prof. E O'Brien, Beaumont Hospital and Prof. C O'Herlihy, National Maternity Hospital, Dublin)

Yap S*, O'Donnell KA, O'Neill C, Mayne PD, Thornton P, Naughten E.*
Factor V Leiden (Arg506Gln), a confounding genetic risk factor but not mandatory for the occurrence of venous thromboembolism in homozygotes and obligate heterozygotes for cystathionine β-synthase deficiency.
Thromb Haemost 1999; 81: 502-5.

FINALIST IN THE ROYAL ACADEMY OF MEDICINE IRELAND PAEDIATRIC REGISTRAR MEDAL, 2000:

Oral presentation:

Royal College of Physicians, Ireland
Yap S*, Boers GHJ, Wilcken B, Wilcken DEL, Naughten ER.*
Vascular risk in homocystinuria - A three centre study on the effects of long-term treatment.

THE ROYAL ACADEMY OF MEDICINE IRELAND PAEDIATRIC REGISTRAR MEDAL, 2001:

Oral presentation:

Royal College of Physicians, Ireland
Yap S*, Rushe H, Howard PM, Naughten ER.*
Early dietary treatment prevents mental retardation in patients with homocystinuria.

THE ROYAL ACADEMY OF MEDICINE IRELAND / IRISH JOURNAL OF MEDICAL SCIENCE (RAMI/IJMS) DOCTORS AWARD FOR EXCELLENCE IN CLINICAL RESEARCH 2001:

("Risk Factor Medicine" category)

Yap S*, Naughten ER, Wilcken B, Wilcken DEL, Boers GHJ.*
Vascular complications of severe hyperhomocysteinaemia in patients with homocystinuria: effects of homocysteine-lowering therapy.
Seminars in Thrombosis and Haemostasis 2000; 26 (3): 335-40

- Trophy and a bursary awarded

FINALIST FOR THE ROYAL ACADEMY OF MEDICINE IRELAND/IRISH JOURNAL OF MEDICAL SCIENCE (RAMI/IJMS) DOCTORS AWARD FOR EXCELLENCE IN CLINICAL RESEARCH 2002:

("Cardiology" category)

Yap S, *Boers GHJ, Wilcken B, Wilcken DEL, Brenton DP, Lee PJ, Walter JH, Howard PM, Naughten ER.*

Vascular outcome in patients with homocystinuria due to cystathionine β-synthase deficiency treated chronically: A multicentre observational study.

Arterioscl Thromb Vasc Biol 2001; 21(12): 2080-5

FINALIST FOR THE ROYAL ACADEMY OF MEDICINE IRELAND/IRISH JOURNAL OF MEDICAL SCIENCE (RAMI/IJMS) DOCTORS AWARD FOR EXCELLENCE IN CLINICAL RESEARCH 2002:

("Clinical Neurology" category)

Yap S, *Rushe H, Howard PM, Naughten ER.*

The intellectual abilities of early treated individuals with pyridoxine nonresponsive homocystinuria due to cystathionine β-synthase deficiency.

J Inher Metab Dis 2001; 24(4): 437-47

FINALIST FOR THE ROYAL ACADEMY OF MEDICINE IRELAND/IRISH JOURNAL OF MEDICAL SCIENCE (RAMI/IJMS) DOCTORS AWARD FOR EXCELLENCE IN CLINICAL RESEARCH 2003:

("Cardiology" category)

Yap S

Classical Homocystinuria: Vascular risks and its prevention

J Inher Metab Dis 2003; 26: 259-65

POSTER PRIZE WINNER FOR MEDICAL GENETICS SOCIETY MALAYSIA CONFERENCE 2010:

Varughese M, Chew HB, ***Yap S***, Thong MK

Glycogen Storage Disease Type III (GSD III) in Malaysian Children: Clinical and laboratory findings

NOMINATED FOR SHEIKH HAMDAN INTERNATIONAL AWARD FOR MEDICAL RESEARCH EXCELLENCE 2011-2012 (Category : Inborn Errors of Metabolism)

Yap S

Classical homocystinuria: Newborn screening with early treatment prevents complications

Travel Award for oral presentation at 4th Asian Congress for Inherited Metabolic Diseases 2015, Taiwan:

Yap S, Tisna H

The acute management of intercurrent illness requires adequate caloric intake in Medium Chain Acyl Co-A Dehydrogenase deficiency (MCADD)

Sheffield Children's Hospital Children's Star Award 2020:

Yap S

Highly commended individual star

CERTIFICATES OF TUITION AND ORGANISATION:

1. Advanced Course in Blood Transfusion for Medical Laboratory Technologists – presented by the National Blood Services Centre**,** Hospital Kuala Lumpur, Malaysia 1993.
2. Workshop on Haematology and AIDS - presented by the Ministry of Health, Malaysia 1993.
3. Speaker at Medical Genetics Conference 2010- 2nd National Dysmorphology Seminar-presented by Medical Genetics Society of Malaysia, 22-23rd April 2011.
4. Speaker at Postgraduate Short Course on Paediatric Neurology- presented by Dept of Paediatrics, Faculty of Medicine, University Malaya, 20-21st May 2010.
5. Co-Editor and member of the Teaching Faculty of the Inaugural Asia Pacific Metabolic Course-presented by Ministry of Health, Malaysia and Faculty of Medicine, University Malaya, 12-14th October 2011.

CERTIFICATES OF ATTENDANCE (SCIENTIFIC SEMINARS AND WORKSHOPS):

1. Symposium and Workshop on "*Update in Haemophilia Care*".
   Haemophilia Society of Malaysia, Blood Services Centre, Hospital Kuala Lumpur,
   Ministry of Health Malaysia – March 1991.
2. Academy of Medicine Congress in Kuala Lumpur – August 1993.
3. CD4/CD8 Immunophenotyping Workshop. Ministry of Health, Malaysia – September 1993.
4. Workshop on Haematology and AIDS. Blood Services Centre, Hospital Kuala Lumpur and Ministry of Health, Malaysia – November 1993
5. Haematology Seminar. Malaysian Society of Pathology – November 1993.
6. Biotechnology Seminar on "PCR-Based Molecular Diagnosis". Medical Faculty, National University of Malaysia – November 1993.
7. 3rd Annual JAPAC Fabry & Pompe Physician Training Workshop- Medical Genetics Society Malaysia, 23-25th April 2010
8. 4th Annual MPS & Pompe Physician Training Workshop – Medical Genetics Society Malaysia, 9-11 September 2011.

INVITED PROFESSIONAL BIOGRAPHICAL INCLUSION:

Marquis Who's Who in Medicine and Healthcare

- 4th Edition 2002 – 2003
- 5th Edition 2004 – 2005
- 6th Edition 2006 – 2007

Marquis Who's Who in Science and Engineering

- 7th Edition 2003 – 2004
- 8th Edition 2004 – 2005
- 9th Edition 2006 – 2007

Marquis Who's Who in the World

- 21st Edition 2004
- 22nd Edition 2005
- 23rd Edition 2006

INVITED LECTURES AND PRESENTATIONS (1994 TO PRESENT)

NATIONAL PRESENTATIONS:

INVITED SPEAKER:

1. Department of Neurology, Beaumont Hospital, Dublin
   Yap S, *Naughten E.*
   Classical Homocystinuria due to Cystathionine β-Synthase Deficiency.
   December 1998

2. Irish Society for Inherited Metabolic Disorders Study Day for Homocystinuria. The Children's Hospital, Temple Street, Dublin
   Yap S.
   Classical Homocystinuria.
   October 1999

3. MSc in Paediatrics Roundtable Discussion, Trinity College Dublin.
   Yap S.
   Severe Hyperhomocysteinaemia in Classical Homocystinuria.
   January 2000

4. Metabolic Workshops for Nurses. The Children's Hospital, Temple Street, Dublin, Ireland
   Yap S
   Classical Homocystinuria: Overview & Management.
   May 2000

5. National Maternity Hospital, Holles Street, Dublin
   Yap S
   Metabolic problems in the newborn
   Special and intensive care of the newborn course for nurses.
   March 2001

6. MSc in Paediatrics Roundtable Discussion, Trinity College Dublin
   Yap S
   Inherited Metabolic Disorders.
   March 2001

7. The Children's Hospital, Temple Street, Dublin
   Yap S, *Naughten E.*
   Phenylketonuria – Irish Experience
   July 2001

8. The Children's Hospital, Temple Street, Dublin
   Yap S, *Naughten E.*
   Classical Homocystinuria – Irish Experience
   July 2001

9. Metabolic Nursing Course
   Children's University Hospital, Temple Street, Dublin
   Yap S
   Homocystinuria and Galactosaemia
   October 2002

10. Metabolic Nursing Course
    Children's University Hospital, Temple Street, Dublin
    Yap S
    The Critically Ill Child: When to suspect a Metabolic Cause
    March 2003

11. Metabolic Nursing Course
    Children's University Hospital, Temple Street, Dublin
    Yap S
    Homocystinuria and Galactosaemia
    March 2003

12. Higher Diploma Neonatal Intensive Care Course (RCSI / Rotunda Hospital)
    Children's University Hospital, Temple Street, Dublin
    Yap S
    When to suspect a metabolic cause in a critically ill child
    May 2003

13. Paediatric Specialist Registrar's Course
    Children's University Hospital, Temple Street, Dublin
    Yap S
    Appropriate metabolic investigations and the interpretation of results
    May 2003

14. National Paediatric Nursing Study Day
    Children's University Hospital, Temple Street, Dublin
    Yap S
    When to suspect a metabolic cause in a critically ill child
    November 2003

15. Metabolic Nursing Course
    Children's University Hospital, Temple Street, Dublin
    Yap S
    The Critically Ill Child: When to suspect a Metabolic Cause
    November 2004

16. Metabolic Nursing Course
    Children's University Hospital, Temple Street, Dublin
    Yap S
    Homocystinuria
    November 2004

17. Medical Genetic Society Malaysia Conference, Kuala Lumpur
    Yap S
    Classical Homocystinuria – Natural History and Clinical Outcome
    April 2010

18. Medical Genetic Society Malaysia Conference, Kuala Lumpur
    Yap S
    Classical Homocystinuria – Treatment and Nutritional Management
    April 2010

19. Post-Graduate Paediatric Neurology Conference, University Malaya, Kuala Lumpur.
    Yap S

Metabolic Encephalopathy
May 2010

20. Medical Genetic Society Malaysia AGM – Mini-Symposium
Yap S
Asian Pacific Metabolic Course
June 2010

21. 2nd Intensive Course in Paediatrics for Master of Paediatrics Year 1, University Malaya, Kuala Lumpur
Yap S
Inborn Errors of Metabolism
September 2010

22. 32nd Malaysian Paediatric Association Congress, Kuala Lumpur.
Yap S
The Critically Ill Child – When to Suspect a Metabolic Cause
October 2010

23. 5th Safe Motherhood Congress, Kuala Lumpur. "Motherhood at the crossroad: Challenges ahead"
Yap S
IEM detection: How early is early?
December 2011

24. FOCUS 2013, The annual national meeting of the association of clinical biochemist, York, UK
Yap S
Expanded Newborn Screening: Homocystinuria
April 2013

25. Mid Eastland Paediatric Registrar Training Day, Queen Medical Center, Nottingham, UK
Yap S
The Critically Ill Child-When to suspect metabolic?
July 2013

26. MetBioNet Annual Training Meeting, Nottingham, UK
Yap S
Expanded Newborn Screening: Homocystinuria –from the bench to the bedside
November 2013

27. Sheffield Children's Hospital Grand Rounds, Sheffield, UK
Yap S
Glutaric Aciduria type 1: Newborn screened
February 2015

28. 2015 East Midlands and South Yorkshire Regional Neonatal Grid Training Day, Metabolic and Endocrine, Sheffield, UK.
Yap S
The Critically Ill Child- when to suspect a metabolic disorder?
July 2014

29. Neonatal Surgical Unit Mandatory Training Day, Sheffield, UK.
Yap S
The Critically Ill Child- when to suspect a metabolic disorder?
July 2015

30. Neonatal Surgical Unit Mandatory Training Day, Sheffield, UK.
Yap S
The Critically Ill Child- when to suspect a metabolic disorder?
July 2015

31. Sheffield Paediatric Neurology Masterclass, Sheffield, UK.
Yap S
Neonatal Metabolic Encephalopathies.
September 2015

32. Sheffield Children's Hospital Grand Rounds, Sheffield, UK
Yap S
Treatment update on hyperammonaemia
March 2016

33. Newborn Bloodspot Forum meeting, Long Eaton, UK
Yap S
Newborn screening: Card to Clinic
May 2016

34. MetBioNet Biomedical Scientist Annual Training Meeting, Manchester, UK
Yap S
Newborn screening: Card to Clinic
May 2016

35. Trent Paediatric Society Meeting, Doncaster, UK
Yap S
Metabolic Disorders in Children: Clinical Spectrum
October 2016

36. Trent Paediatric Society Meeting, Doncaster, UK
Yap S
Metabolic Disorders in Children: investigations, interpretation & management
October 2016

37. Sheffield Regional Newborn Screening Laboratory Open Day for Midwives, UK
Yap S
Referral of newborn screened positive cases: How important is timing for inherited metabolic diseases.
June 2017

38. Sheffield Community Paediatrics Training Session, UK
Yap S
Developmental delay: when and how to investigate for inborn errors of metabolism.
October 2017

39. Sheffield Children's Hospital Nurses Training Day for IMD
Yap S
What is and how to manage Inherited Metabolic Disorders
March and May 2018

40. Newborn Sceening Lab Open Day for Midwives, Sheffield.
Yap S
Referral of Newborn Screen Positive cases: how important is timing for inherited metabolic diseases
June 2018

41. MetBioNet BMS Training Group Annual Training, Manchester, UK
Yap S
Introduction to Inborn Errors of Metabolism from a clinical point of view
October 2018

42. Sheffield Children's Hospital PCCU Teaching.
Yap S
Hyperammonaemia: Recognising it as a metabolic emergency.
June 2020

43. Regional PCCU training: Time is brain
Yap S
Metabolic encephalopathy: What you really need to know.
September 2020

44. Sheffield Children's Hospital, Metabolic case meeting
Yap S
Acute Hyperammonaemia: Clinical diagnostic clues
November 2020

45. Sheffield Children's Hospital PCCU Teaching.
Yap S
Hyperammonaemia: Recognising it as a metabolic emergency and application in clinical cases.
July 2021

ORAL PRESENTATIONS:

1. The Children's Hospital, Temple Street, Grand Rounds:
   Yap S*, Naughten E.*
   Long chain hydroxyl-acyl dehydrogenase deficiency – presentation and treatment.
   August 1994

2. National Maternity Hospital, Neonatal Grand Rounds:
   Yap S*, Gorman WA, Murphy J, O'Brien N.*
   Nutritional needs of premature babies
   February 1995

3. Junior Irish Paediatric Association – Annual Neonatal Meeting
   Yap S*, Gorman WA, Murphy J, O'Brien N.*
   Juene Syndrome
   April 1995

4. The Children's Hospital, Temple Street, Grand Rounds
   Yap S*, Naughten E.*
   Diagnostic Approach to Fat Oxidation Defects
   December 1996

5. Irish Paediatric Association, Dublin
   Yap S*, Boers GHJ, Wilcken B, Wilcken DEL, Naughten ER.*
   A three centre study on the effects of long-term treatment on the vascular outcome of patients with homocystinuria due to cystathionine β-synthase deficiency.
   November 1999

6. Irish Paediatric Association Meeting, Dublin
   Yap S*, Annesley D, Pittock S, Ryan A, Brennan P, Hardiman O, Naughten ER.*
   Superior sagittal sinus thrombosis and homocystinuria: early anticoagulation is essential.
   November 1999

7. 1st Royal Academy of Medicine Ireland Paediatric Registrar Medal, Royal College of Physician, Dublin
   Yap S*, Boers GHJ, Wilcken B, Wilcken DEL, Naughten ER.*
   A three centre study on the effects of long-term treatment on the vascular outcome of patients with homocystinuria due to cystathionine β-synthase deficiency.
   January 2000

8. The Children's Hospital, Temple Street, Grand Rounds
   Yap S*, Boers GHJ, Wilcken B, Wilcken DEL, Naughten ER.*
   A three centre study on the effects of long-term treatment on the vascular outcome of patients with homocystinuria due to cystathionine β-synthase deficiency.
   May 2000

9. 2nd Royal Academy of Medicine Ireland Paediatric Registrar Medal, Royal College of Physician, Dublin
   Yap S*, Rushe H, Howard PM, Naughten ER.*
   Early dietary treatment prevents mental retardation in patients with homocystinuria.
   April 2001

10. Spring Irish Perinatal Society Meeting, Galway
    Yap S*, Barry-Kinsella C, Naughten ER.*
    The role of dietary management and anticoagulation in maternal pyridoxine nonresponsive homocystinuria with good maternal and fetal outcome
    April 2001

11. Spring Irish Perinatal Society Meeting, Dublin
    Wallace A, Reddy KM, Stenson C, McGovern C, Kellett M, ***Yap S***, Treacy EP, Naughten ER.
    Maternal Phenylketonuria
    March 2003

12. Spring Irish Perinatal Society Meeting, Dublin
    Tirupathi S, McGuinn F, Healy F, Sheridan M, White M, Treacy E, Naughten E, ***Yap S.***
    Transient Methylmalonic Aciduria - May be a deficiency and not a disease
    March 2003

13. Joint Irish Paediatric Association/ Junior Irish Paediatric Association Winter Meeting, Dublin
    Hughes J, Clark A, Geoghan O, O'Toole , Hendroff , Rogers Y, Walsh O, O'Regan M, Stenson C, Lambert D, ***Yap S***, Manning R, Treacy E.
    A Study on Outcomes in Siblings with Galactosaemia in Ireland.
    November 2005

14. British Inherited Metabolic Diseases Group (BIMDG) Trainees Meeting, London, UK
    AF Catchpole, CA Scott, SE Olpin, ***S Yap***, JR Bonham
    Missed opportunities for early diagnosis – Lysinuric Protein Intolerance
    July 2013

POSTER PRESENTATIONS:

1. 22nd Annual Conference of the Association of Clinical Biochemists in Ireland
   Yap S*, O'Donnell KA, O'Neill C, Mayne PD, Thornton P, Naughten E.*
   Factor V Leiden (A506G) and thrombosis in homozygotes and obligate heterozygotes for cystathionine β-synthase deficiency.
   October 1999

2. Irish Ophthalmology Meeting, Dublin
   Mulvihill A, ***Yap S****, Naughten ER, O'Keefe M.*
   Ocular findings and delay in diagnosis in patients with late diagnosed or poorly controlled Homocystinuria.
   June 2000

3. 22nd Annual Conference of the Association of Clinical Biochemists in Ireland
   Yap S*, Naughten ER, Corlett L, Howard P, Irranca M, Howard PM, O'Brien M, Thornton* P.
   The effects of treatment on bone mineral density (BMD) in patients with homocystinuria due to cystathionine β-synthase (CBS) deficiency.
   October 1999

4. Irish Society of Human Genetics Annual Scientific Meeting, Smurfit Institute of Genetics, Trinity College, Dublin
   Yap S*, O'Donnell KA, O'Neill C, Naughten ER.*
   Coexistence of Factor V Leiden, Prothrombin G20210A, Methylenetetrahydrofolate reductase C677T and A1298C polymorphisms in patients with cystathionine β-synthase deficiency: Effects of treatment on the vascular outcome.
   September 2001

5. Irish Society of Human Genetics Annual Scientific Meeting, Smurfit Institute of Genetics, Trinity College, Dublin
   Yap S*, O'Donnell KA, O'Neill C, Naughten ER.*
   Coexistence of Methylenetetrahydrofolate reductase C677T and A1298C polymorphisms, Factor V Leiden and Prothrombin G20210A in obligate heterozygotes for cystathionine β-synthase deficiency.
   September 2001

6. 24th Annual Conference of the Association of Clinical Biochemists in Ireland
   Yap S*, O'Donnell KA, O'Neill C, Naughten ER.*
   Coexistence of Factor V Leiden, Prothrombin G20210A, Methylenetetrahydrofolate reductase C677T and A1298C polymorphisms in patients with cystathionine β-synthase deficiency: Effects of treatment on the vascular outcome.
   October 2001

7. 24th Annual Conference of the Association of Clinical Biochemists in Ireland
   Yap S*, O'Donnell KA, O'Neill C, Naughten ER.*
   Coexistence of Methylenetetrahydrofolate reductase C677T and A1298C polymorphisms, Factor V Leiden and Prothrombin G20210A in obligate heterozygotes for cystathionine β-synthase deficiency.
   October 2001

8. Irish Paediatric Association Meeting, Royal College of Physicians Ireland, Dublin
   Tirupathi S, McGuinn P, Healy F, Sheridan M, White M, Treacy E, Naughten E, ***Yap S***
   Transient Methylmalonic Aciduria: May be a deficiency and not a disease
   November 2002

9. Annual Meeting of Haematology Association of Ireland, Limerick, Ireland.
   O'Shea DM, Desmond R, Nolan B, ***Yap S***
   A case of severe hyperhomocysteinaemia presenting with thromboembolisms.
   November 2003.

10. Annual Meeting of Haematology Association of Ireland, Limerick, Ireland.
    Flanagan M, McCauly M, White B, ***Yap S****, Nolan B.*
    Successful dietary treatment of severe hyperhomocysteinaemia in a man with recurrent thrombosis.
    November 2003.

11. Irish Society of Human Genetics Annual Scientific Meeting, Royal College of Surgeons in Ireland, Dublin.
    Chew HB, Kluijtmans LAJ, ***Yap S****.*
    A novel association of Pyridoxine (B6) responsive phenotype with Cystathionine β-Synthase (CβS) 919G→A (G307S) genotype.
    September 2004.

12. Irish Paediatric Association Meeting, Dublin.
    Chew HB, Kluijtmans LAJ, ***Yap S****.*
    A novel association of Pyridoxine (B6) responsive phenotype with Cystathionine β-Synthase (CβS) 919G→A (G307S) genotype.
    November 2004.

13. Medical Genetics Society Malaysia Conference, Kuala Lumpur, Malaysia:
    Varughese M, Chew HB, ***Yap S****, Thong MK*
    Glycogen Storage Disease Type III (GSD III) in Malaysian Children: Clinical and laboratory findings.
    May 2010.

14. British Inherited Metabolic Diseases Group (BIMDG) Meeting, Belfast, UK.
    Nesbitt IM, Kirk RJ, Leong HY, Croft J, Dalley J, Watkinson J, Olpin S, ***Yap S***
    Unexpected biochemical and genetics findings in a family with SCAD.
    June 2015

15. 30th Annual Paediatric Intensive Care Society (PICS) Conference, Southhampton, UK
    Riddell R, Thomas R, Jackman L, Watkinson J, Cherachathoor Joseph U, Blythe D, Hart A, ***Yap S***
    Newborn screening, nutritional management and emergent haemodiafiltration facilitates successful outcome in Maple Syrup Urine Disease (MSUD)
    October 2016

INTERNATIONAL PRESENTATIONS:

INVITED SPEAKER:

1. Biennial Scientific Meeting, Asia Pacific Association for the Study of Liver, Kuala Lumpur, Malaysia
   Duraisamy G, ***Yap S****, Zuraidah H.*
   HIV and Hepatitis B,C,D in IV Drug Users Screened in 1992
   January 1994

2. 1st. Dietitian's meeting at the Society for the Study of Inborn Errors of Metabolism, Cardiff, Wales
   Howard P, Irranca M, Kellett M, ***Yap S****, Naughten E.*
   Protein, Methionine, and Cystine Intakes of dietary treated patients with homocystinuria due to cystathionine β-synthase deficiency.
   September 1996

3. 2nd. International Conference on Homocysteine Metabolism, Nijmegen, The Netherlands
   Yap S*, O'Donnell KA, O'Neill, Mayne PD, Thornton P, Naughten E.*
   Factor V Leiden (Arg506Gln) and thrombosis in homozygotes and obligate heterozygotes for cystathionine β-synthase deficiency.
   April 1998

4. 4th Dietitian's meeting at the Society for Study of Inborn Errors of Metabolism, Genoa, Italy
   Naughten ER, Hunt D, ***Yap S****.*
   Homocystinuria – Diet, Management and Outcome.
   September 1999

5. British Inherited Metabolic Disease Group Workshop on "Homocystinuria – What should we be doing?", Council Chamber, Royal College of Physicians, London
   Naughten ER, Mayne PD, ***Yap S****.*
   Homocystinuria: Screening, problems, outcome. Ireland 1971-1999.
   October 1999

6. British Inherited Metabolic Disease Group Workshop on "Homocystinuria – What should we be doing?", Council Chamber, Royal College of Physicians, London
   Yap S*.*
   Vascular Outcome of Patients with Homocystinuria.
   October 1999

7. Annual German Paediatric Symposium on Inborn Errors of Metabolism.
   Molecular pathology of homocystinuria, Barcelona, Spain,
   Yap S*, Naughten ER.*
   Treatment of homocystinuria due to cystathionine β-synthase deficiency: State of the art.
   November 2000

8. National Metabolic Conference, Columbus, Ohio.
   Yap S*, Naughten ER*
   Phenylketonuria – Irish Experience
   May 2001

9. National Metabolic Conference, Columbus, Ohio.
   Yap S*, Naughten ER.*
   Homocystinuria : Screening, Treatment and Outcome
   May 2001

10. Biochemical Genetic Grand Rounds at The Montreal Children's Hospital, McGill University, Montreal, Canada
    Yap S*, Naughten ER*
    Phenylketonuria – Irish Experiences on screening, treatment, Maternal Phenylketonuria and outcome.
    May 2001

11. Biochemical Genetic Grand Rounds at The Montreal Children's Hospital, McGill University, Montreal, Canada
    Yap S*, Naughten ER*
    Classical Homocystinuria – Aspects of screening, treatment, biochemical control and outcome in Ireland
    May 2001

12. British Inherited Metabolic Disease Group Meeting, Dublin
    Yap S
    Homocystinuria – Outcome in chronically treated patients
    November 2001

13. The Association of Clinical Biochemists and British Inherited Metabolic Disease Group Meeting, Doncaster, U.K.
    "Metabolic Medicine, Adolescence to Adulthood"
    Yap S
    Experience of Homocystinuria into Adulthood
    March 2002

14. Department of Genetics and Metabolism, Hospital Necker Enfant Malades, Paris, France
    Yap S
    Homocystinuria - Irish experience from newborn to adulthood
    April 2002

15. State Laboratory Institute, New England Newborn Screening Program, University of Massachusetts and Amino Acid Laboratory/Metabolic Unit, Massachusetts General Hospital, Harvard School of Medicine, Boston, U.S.A.
    Yap S
    Homocystinuria - the Irish experience
    May 2002

16. 40th Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Dublin, Ireland
    Yap S
    Classical Homocystinuria: Vascular risk and its prevention
    September 2002

17. USA National Metabolic Conference, Orlando, Florida
Yap S
Homocystinuria: Irish management and treatment
October 2002

18. 4th International Conference on Homocysteine Metabolism, Basel, Switzerland
Yap S
Cystathionine β-synthase deficiency and vascular disease: Treatment and outcome
July 2003

19. 2003 PKU Parents Conference, Los Angeles, California.
Yap S
Keynote Lecture: Inherited Metabolic Disorders: The Irish Experience.
October 2003

20. Biochemical Genetics Grand-round, Los Angeles Children's Hospital, Los Angeles, California.
Yap S
Classical Homocystinuria: Screening, Treatment and Long Term Clinical Outcome.
October 2003

21. Paediatric Grand-round, Institute of Paediatrics, Hospital Kuala Lumpur, Malaysia.
Yap S
Classical Homocystinuria – From Basic Science to Clinical Medicine and Long Term Outcome.
June 2004

22. Paediatric and Neonatal Grand-round, Selayang Hospital, Kuala Lumpur, Malaysia.
Yap S
When to suspect a metabolic cause in a critically ill child
June 2004.

23. Medical Research Grand-round, University Putra Malaysia, Bangi, Malaysia.
Yap S
Classical Homocystinuria – From Basic Science to Clinical Medicine and Long Term Outcome.
June 2004

24. Paediatric and Neonatal Grand-round, Hospital Bandar Tun Razak, University Kebangsaan Malaysia.
Yap S
When to suspect a metabolic cause in a critically ill child
June 2004.

25. 12th Abbott Nutrition Metabolic Conference, Advances in Management of Inherited Metabolic Disorders, Austin, Texas, USA.
Yap S
Homocystinuria – Natural History and Clinical Outcomes with Treatment
May 2009.

26. 12th Abbott Nutrition Metabolic Conference, Advances in Management of Inherited Metabolic Disorders, Austin, Texas, USA.
Yap S
Homocystinuria – Medical and Nutritional Management
May 2009.

27. Inaugural Asia Pacific Metabolic Course 2011, Kuala Lumpur, Malaysia.
Yap S
Defects of Protein Metabolism: Treatment and Management
October 2011.

28. Inaugural Asia Pacific Metabolic Course 2011, Kuala Lumpur, Malaysia.
Yap S
The Role of the Paediatrician in the follow-up of patients diagnosed by Newborn Screening
October 2011.

29. Symposium for Hyperammonaemia Research And Education (SHARE 2013): Hyperammonaemia in UCDs and OAs- the journey from the newborn to adult, Amsterdam, The Netherlands.
Yap S
Carglumic Acid in Neonatal Severe Hyperammonaemia
June 2013

30. Department of Paediatrics invited CME Lecture, University Malaya, Malaysia:
Yap S
Metabolic Encephalopathy
March 2015

31. 4th Asian Congress for Inherited Metabolic Diseases (ACIMD 2015), Taiwan:
Yap S
Treatment update on hyperammonaemia.
March 2015

32. Libyan Metabolic Workshop, Beirut:
Yap S
Homocystinuria: Natural History and Effects of Treatment
June 2015

33. Libyan Metabolic Workshop, Beirut:
Yap S
Homocystinuria: Medical and Nutritional Treatment
June 2015

34. Libyan Metabolic Workshop, Beirut:
Yap S
Management of Hyperammonaemia in Organic Acidaemia
June 2015

35. Satellite Symposium of the Society for the Study of Inborn Errors of Metabolism 2015, Lyon, France:
Yap S
Meet the Expert: Case Sharing on Hyperammonaemia in Organic Acidaemia
Sept 2015

36. 12th Middle East Metabolic Group Meeting (MEMG 2015), Muscat, Oman
Yap S
Orphan Europe Workshop: State of the art in the management of Organic Acidaemia
November 2015

37. 10th Latin American Congress of Inborn Errors of Metabolism and Newborn Screening, Santiago, Chile:
Yap S
Hyperammonaemia in Organic Acidaemia: perspective, acute and Chronic Treatment.
November 2015

38. Orphan Europe Advisory Board for Carbaglu, Paris, France.
Yap S
Management of Chronic Hyperammonaemia in Organic Acidaemia
December 2015

39. Iraqi Metabolic Workshop, Beirut, Lebanon
Yap S
Hyperammonaemia in Organic Aciduria: Acute & Chronic Management
April 2016

40. Iraqi Metabolic Workshop, Beirut, Lebanon
Yap S
Homocystinuria: Biochemical and clinical manifestation
April 2016

41. Iraqi Metabolic Workshop, Beirut, Lebanon
Yap S
Homocystinuria: Therapeutic goals, Treatment and Long term outcome
April 2016

42. Symposium on Hyperammaonaemia due to Urea Cycle Defects and Organic Acidurias, Stockholm, Sweden.
Yap S
Hyperammonaemia in Organic Acidurias: Acute and Chronic Management
May 2016

43. Satellite Symposium of the Society for the Study of Inborn Errors of Metabolism 2016, Rome
Yap S
Expert Viewpoints: Effective management of hyperammonaemia in organic acidaemia:
Clinical cases in the chronic use of carbaglu in propionic acidaemia
September 2016

44. Annual Conference of the Indonesian Paediatric Society, Makassar, Indonesia
Yap S
The critically ill infant: when to suspect & how to investigate for metabolic disorder
October 2016

45. Indonesian Metabolic Workshop, Makassar, Indonesia.
Yap S
Metabolic encephalopathy
October 2016

46. Secondary Urea Cycle Defect Workshop, Leipzig, Germany
Yap S
Chronic Treatment of Patients with Organic Acideamia
March 2017

47. 1st Paediatric Nutrition and Metabolic (NutriMet) Workshop, Jakarta, Indonesia
Yap S
Emergency treatment in Inherited Metabolic Disorders
April 2017

48. Nutrition Working Group (NutriMet) Workshop, Jakarta, Indonesia
Yap S
Metabolic Emergency Treatment
April 2017

49. Nutrition Working Group (NutriMet) Workshop, Jakarta, Indonesia
Yap S
Diagnostic Dilemma
April 2017

50. International Congress of Inborn Errors of Metabolism, Rio de Janeiro, Brazil.
Yap S
Exploring long term therapy for hyperammonaemia in organic aciduria: Clinical cases
September 2017

51. International Congress of Inborn Errors of Metabolism, Rio de Janeiro, Brazil.
Yap S
From the bench to the bedside: Functional test for NAGS deficiency
September 2017

52. International Congress of Inborn Errors of Metabolism, Rio de Janeiro, Brazil.
Yap S
From the bench to the bedside: Long term management of organic acidaemia
September 2017

53. Organic Acidaemia Treatment: Standards Today and Tomorrow, Frankfurt, Germany.
Yap S
European examples of organic acidaemia patient treatment
November 2017

54. Organic Aciademia in the Netherlands: Diagnostics and Treatment, Zeist, the Netherlands.
Yap S
Classical Organic Acidaemia: Carglumic acid in rapid ammonia detoxification and long term treatment.
November 2017

55. Global Advisory Board Meeting on Primary and Secondary NAGS deficiency, Amsterdam, the Netherlands.
Yap S
Carglumic acid in the long term treatment of OA: clinical examples
December 2017

56. 14th Symposium of the Portuguese Society of Metabolic Disorders, Porto, Portugal.
'Converging Paths in Inborn Errors of Metabolism'
Yap S
Carglumic acid: Lessons learnt from treating Organic Acidurias
March 2018

57. 2nd Paediatric Nutrition and Metabolic (NutriMet 2) Symposium, Surabaya, Indonesia.
Yap S
Critically Ill Child: When to suspect. how to investigate and how to manage.
April 2018

58. Nutrition Working Group (NutriMet 2) Workshop, Surabaya, Indonesia
Yap S
Comments on cases: Metabolic acidosis and hypoglycaemia -Glycogen storage disorders
April 2018

59. Nutrition Working Group (NutriMet 2) Workshop, Surabaya, Indonesia
Yap S
Hypoglycaemia: how to investigate and manage
April 2018

60. Nutrition Working Group (NutriMet 2) Workshop, Surabaya, Indonesia
Yap S
Hyperammonaemia: differential diagnosis and treatment modalities
April 2018

61. 13th Brazilian Medical Genetics Congress, Rio de Janeiro, Brazil
Hyperhomocysteinaemia: Diagnosis and Therapeutics
Yap S
Classical Homocystinuria: Pearls of Treatment
May 2018

62. 13th Brazilian Medical Genetics Congress, Rio de Janeiro, Brazil
Emergency Management in IEM
Yap S
Emergency Management in Urea Cycle Disprders
May 2018

63. 13th Brazilian Medical Genetics Congress, Rio de Janeiro, Brazil
Laboratory Clinical Approach of Organic Acidurias
Yap S
Treatment of organic acidurias : discussions of cases
May 2018

64. "Hereditary Metabolic Diseases" Patient/Parent-Expert Meeting, Rio de Janeiro, Brazil
Yap S
Back to basics with homocystinuria – what patients need to know!
May 2018

65. International Young Metabolicians Day, Vienna.
'Primary and Secondary Hyperammonaemia: identification, diagnosis and management.'
Yap S
Hyperammonaemia: Identification, Diagnosis and Management
June 2018

66. International Young Metabolicians Day, Vienna.
Yap S
Organic Acidaemia: PROTECT study & MMA case study
June 2018

67. International Young Metabolicians Day, Vienna.
Yap S
Organic Acidaemia: Case study on long term use of carglumic acid
June 2018

68. Central and East Europe Experts Day, Vienna.
'Primary and Secondary Hyperammonaemia: identification, diagnosis and management.'
Yap S
Hyperammonaemia: Identification, Diagnosis and Management
June 2018

69. Central and East Europe Experts Day, Vienna.
'Primary and Secondary Hyperammonaemia: identification, diagnosis and management.'
Yap S
Organic Acidaemia: PROTECT study & MMA case study
June 2018

70. Central and East Europe Experts Day, Vienna.
'Primary and Secondary Hyperammonaemia: identification, diagnosis and management.'
Yap S
Organic Acidaemia: Case study on long term use of carglumic acid
June 2018

71. Hyperammonaemia Masterclass, Russian Children's Clinical Hospital, Moscow.
Yap S
Hyperammonaemia: Evaluation, Differential Diagnosis and Emergency Treatment.
August 2018

72. Hyperammonaemia Masterclass, Russian Children's Clinical Hospital, Moscow.
Yap S
Primary Hyperammonaemia:
Urea Cycle disorders - diagnostics and management
August 2018

73. Hyperammonaemia Masterclass, Russian Children's Clinical Hospital, Moscow.
Yap S
Secondary Hyperammonaemia:
Organic Aciduria – pathophysiology, treatment modalities and research
August 2018

74. 1st Asian PACific (APAC) Masterclass, Kuala Lumpur
'Primary and Secondary Hyperammonaemia: Past, Present & Future.'
Yap S
Hyperammonaemia: Identification, Diagnosis and Management
August 2018

75. 1st Asian PACific (APAC) Masterclass, Kuala Lumpur
'Primary and Secondary Hyperammonaemia: Past, Present & Future.'
Yap S
Organic Acidaemia: Lessons from the Past to Vision for the Future
August 2018

76. 1st Asian PACific (APAC) Masterclass, Kuala Lumpur
'Primary and Secondary Hyperammonaemia: Past, Present & Future.'
Yap S
Organic Acidaemia: Case study on long term use of carglumic acid
August 2018

77. Satellite Symposium of the Society for the Study of Inborn Errors of Metabolism 2018, Athens
'Opitimsing long-term outcomes of patients with PA & MMA'
Yap S
Challenges with protein restrictions & malnutrition in the long-term management of Pa & MMA patients
August 2018

78. Satellite Symposium of the Society for the Study of Inborn Errors of Metabolism 2018, Athens
'Opitimsing long-term outcomes of patients with PA & MMA'
Yap S
Optimising long-term outcomes with Carbaglu: current and future data:
A 4 year follow-up case study & outlines of the PROTECT and CAMP studies
August 2018

79. International Paediatric Pathology Association, 40th Advanced Course on Paediatric Pathology, Sheffield
Yap S
Clinico-pathologic clues to diagnosis of Inherited metabolic diseases
August 2018

80. Meet-the-expert session at the 7th Congress of the European Academy of Paediatrics Societies (EAPS 2018), Paris
Yap S
Understanding hyperammonaemia: Practical pearls in diagnosis to optimising long-term outcomes in organic acidurias
November 2018

81. 60th Annual Meeting of the Japanese Society of Inherited Metabolioc Diseases (JSIMD)/ 16th Symposium of the Asian Inherited Metabolic Diseases (ASIMD), Chifu, Japan
Yap S
Hyperammonaemia: Clinical Experience of Carglumic Acid.
November 2018

82. Round Table Discussion on Hyperammonaemia in Organic Acidurias in Japan, Gifu, Japan
Yap S
Global practices on organic acidurias: Past, present and future.
November 2018

83. Orphan Europe European Cycle Meeting, Nice, France
Yap S
Organic Acidurias: Fundamentals and Treatment.
January 2019

84. 1st Royal College of Paediatrics and Child Health, MENA Conference, Cairo, Egypt
Yap S
Managing Hyperammonaemia
January 2019

85. International Masterclass on Organic Acidurias, Madrid
Yap S
Secondary Hyperammonaemia in Organic Acidurias: Biochemical Interactions
February 2019

86. International Masterclass on Organic Acidurias, Madrid
Yap S
Nutritional Treatment in Organic Acidurias: its Complexity and Associated Complications
February 2019

87. International Masterclass on Organic Acidurias, Madrid
Yap S
Chronic Management of Organic Acidurias:
Literature Review, On-going Trials & Case Presentations
February 2019

88. 3rd Homocystinuria Patient-Expert Meeting, Rome
Yap S
Cystathionine B-Synthase Deficiency: Clinical Overview
February 2019

89. 3rd Homocystinuria Patient-Expert Meeting, Rome
Yap S
Cystathionine B-Synthase Deficiency: Interactive Panel Discussions
February 2019

90. Summit in Rare Diseases, Mexico
Yap S
Hyperammonaemia: evaluation, differential diagnosis and emergency treatment
March 2019

91. Summit in Rare Diseases, Mexico
Yap S
Primary Hyperammonaemia: Urea cycle disorders-diagnostics and management
March 2019

92. Summit in Rare Diseases, Mexico
Yap S
Secondary Hyperammonaemia: Organic aciduria – pathophysiology and treatment
March 2019

93. Summit in Rare Diseases, Mexico
Yap S
Classical homocystinuria: Clinical overview
March 2019

94. Summit in Rare Diseases, Mexico
Yap S
Classical homocystinuria: Treatment
March 2019

95. Masterclass on 'Optimising the long term outcome of patients with PA, MMA and IVA', Moscow.
Yap S
Hyperammonaemia: Differential diagnosis and acute treatment
May 2019

96. Masterclass on 'Optimising the long term outcome of patients with PA, MMA and IVA', Moscow.
Yap S
Clinical case studies on work-up and management of acute hyperammonaemia
May 2019

97. Masterclass on 'Optimising the long term outcome of patients with PA, MMA and IVA', Moscow.
Yap S
Acute management of Organic Acidurias: Literature review
May 2019

98. Masterclass on 'Optimising the long term outcome of patients with PA, MMA and IVA', Moscow.
Yap S
Clinical case studies on acute presentation and management of MMA
May 2019

99. Masterclass on 'Optimising the long term outcome of patients with PA, MMA and IVA', Moscow.
Yap S
Treatment of Organic Aciduria: Nutritional treatment and its associated complications
May 2019

100. Masterclass on 'Optimising the long term outcome of patients with PA, MMA and IVA', Moscow.
Yap S
Strategies for long term treatment of organic acidurias
May 2019

101. Masterclass on 'Optimising the long term outcome of patients with PA, MMA and IVA', Moscow.
Yap S
Clinical case studies on the long term treatment in organic acidurias.
May 2019

102. International Congress on 'Genetics of the XXI century' , Moscow, Russia
Yap S
Understanding Hyperammonaemia: Diagnosis to optimising long term treatment of organic acidurias.
May 2019

103. Abbott Metabolic Conference, Memphis, USA
Yap S
Classical homocystinuria: Clinical overview
May 2019

104. Abbott Metabolic Conference, Memphis, USA
Yap S
Understanding Hyperammonaemia: Diagnosis to optimising long term treatment of organic acidurias.
May 2019

105. Central Eastern European Metabolic Experts Meeting, Vienna
Yap S
Secondary Hyperammonaemia: Organic aciduria- acute and long term treatment
June 2019

106. International Masterclass on Acute and Chronic Management of PA, MMA & IVA, Berlin
Yap S
Nutritional management of OA: Its complexity and associated complications
June 2019

107. International Masterclass on Acute and Chronic Management of PA, MMA & IVA, Berlin
Yap S
Chronic management of Organic aciduria: literature review and on-going trials.
June 2019

108. International Masterclass on Acute and Chronic Management of PA, MMA & IVA, Berlin,
Yap S
Case studies on chronic management of organic acidurias.
June 2019

109. Asian Pacific Masterclass on 'Optimising the long term outcome of patients with primary and secondary hyperammonaemia', Taiwan.
Yap S
Primary hyperammonaemia: CPS 1 deficiency.
August 2019

110. Asian Pacific Masterclass on 'Optimising the long term outcome of patients with primary and secondary hyperammonaemia', Taiwan.
Yap S
Secondary hyperammonaemia: organic aciduria.
August 2019

111. Asian Pacific Masterclass on 'Optimising the long term outcome of patients with primary and secondary hyperammonaemia', Taiwan.
Yap S
Acute management of IVA, PA &MMA: recent publications & on-going trials
August 2019

112. Asian Pacific Masterclass on 'Optimising the long term outcome of patients with primary and secondary hyperammonaemia', Taiwan.
Yap S
Chronic management of PA & MMA: publications & on-going trials.
August 2019

113. SSIEM satellite symposium on 'Exploring long term strategies for patients with propionic and methylmalonic acidurias', Rotterdam
Yap S
Organic Acidurias: on going trials
September 2019

114. Australia Masterclass on 'Exploring treatment strategies for patients with organic acidurias', Sydney, Australia.
Yap S
Acute hyperammonaemia: differential diagnosis & management
November 2019

115. Australia Masterclass on 'Exploring treatment strategies for patients with organic acidurias', Sydney, Australia.
Yap S
Primary hyperammonaemia: NAGS & CPS 1 deficiencies.
November 2019

116. Australia Masterclass on 'Exploring treatment strategies for patients with organic acidurias', Sydney, Australia.
Yap S
Secondary hyperammonaemia: organic acidurias
November 2019

117. Australia Masterclass on 'Exploring treatment strategies for patients with organic acidurias', Sydney, Australia.
Yap S
Acute Management of organic acidurias: case studies and recent publications
November 2019

118. Australia Masterclass on 'Exploring treatment strategies for patients with organic acidurias', Sydney, Australia.
Yap S
Chronic management of organic acidurias: case studies, recent publications & on-going trials.
November 2019

119. Clinical insights into the long term management of organic acidurias, PA & MMA, Madrid
Yap S
PROTECT study: what we have learnt?
November 2019

120. PROTECT Trial Investigators Virtual Meeting – France, Paris.
Yap S
PA & MMA: disease background, diagnosis and treatment.
December 2019

121. Organic Aciduria Masterclass on Long term management of PA & MMA patients, Lisbon.
Yap S
Natural protein restriction, Diet & Growth, complications and development issues in PA & MMA.
Patients pharmacological long term management of PA & MMA patients: personal experience.
PROTECT Trial overview.
February 2020

122. Meet the Expert Webinar on 'Long term management of Organic Acidurias'
Yap S
Organic acidurias: Pathophysiology, Diagnosis, Treatment & Research
March 2020

123. REaSoN neonatal virtual meeting.
Yap S
Neonatal hyperammonaemia: how to manage this medical emergency?
June 2020

124. Colombian Virtual Masterclass on Hyperammonaemia
Yap S
Hyperammonaemia: how to manage this medical emergency.
PROTECT trial overview
July 10, 2020

125. Colombian Virtual Masterclass on Hyperammonaemia
Yap S
Hyperammonaemia: how to manage this medical emergency.
PROTECT trial overview
July 23, 2020

126. Colombian Virtual Masterclass on Hyperammonaemia
Yap S
Hyperammonaemia: how to manage this medical emergency.
PROTECT trial overview
July 24, 2020

127. PROTECT Trial Investigators Virtual Meeting – UK, Nordic & Germany
Yap S
Chronic management of PA & MMA: Disease background, Diagnosis and treatment, Diet and place of transplantation.
September 2020

128. Colombian Masterclass Webinar 1
Yap S
Hyperammonaemia: Evaluation, Differential diagnosis & Emergency treatment.
September 10, 2020.

129. Colombian Masterclass Webinar 2
Yap S
Urea Cycle Disorders: Diagnostics and Management
September 17, 2020.

130. Colombian Masterclass Webinar 3
Yap S
Organic Acidurais: Pathophysiology, Diagnosis, Treatment & Research (PROTECT trial)
September 24, 2020.

131. Virtual Expert Meeting on Chronic Hyperammonaemia Management in PA & MMA patients.
Yap S
Long term management of two cases of propionic aciduria.
PROTECT study protocol.
September 30, 2020

132. Metabolic Support UK Virtual Annual Conference.
Yap S
Update on PROTECT study.
October, 2021

133. PROTECT Trial Investigators Virtual Meeting – Spain
Yap S
Chronic management of PA & MMA: Disease background, Diagnosis and treatment, Diet and place of transplantation.
October 15, 2021

134. PROTECT Trial Investigators Virtual Meeting – Italy & France
Yap S
Chronic management of PA & MMA: Disease background, Diagnosis and treatment, Diet and place of transplantation.
October 22, 2021

135. SSIEM virtual satellite symposium on 'Long term management of organic academia patients with Carbaglu: The PROTECT trial overview & interim analysis.
Yap S
Organic Acidurias: on going trials
December 2020

136. PROTECT Trial Investigators Virtual Meeting
Yap S
Interim data.
June 2021

137. Webinar on PA & MMA dietary management and treatment recommendations/guidelines
Yap S
PA & MMA dietary management and treatment recommendations/guidelines and personal experience cases with clinical outcome
June 2021

138. China Webinar on Hyperammonaemia:
Yap S
Acute & chronic management of Urea Cycle Disorders and Organic Acidurias.
July 2021

139. Webinar on Homocystinuria: Discrrepancy between recommendations vs current clinical practice and Untreated vs Treated patients with Homocystinuria
Yap S
Clinical outcomes of untreated and treated patients with homocystinuria – CBS deficiency.
September 2021

140. SSIEM virtual satellite symposium on 'Long term management optimisation of patients with propionic and methylmalonic acidurias.
Yap S
Clinical experience with long term treatment of patients with propionic acidurias.
PROTECT trial – Overview and interim data.
September 2021

141. German APS 34th Annual Symposium – satellite symposium on 'Long term management optimisation of patients with propionic and methylmalonic acidurias.
Yap S
PROTECT trial - Overview and interim data
Clinical experience with long term treatment of patients with propionic and methylmalonic acidurias.
September 2021

ORAL PRESENTATION:

1. 37th Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Genoa, Italy
   Yap S*, Boers GHJ, Wilcken B, Wilcken DEL, Naughten ER.*
   A three centre study on the effects of long-term treatment on the vascular outcome of patients with homocystinuria due to cystathionine β-synthase deficiency.
   September 1999

2. VIII International Congress of Inborn Errors of Metabolism, Cambridge, UK. September
   Yap S*, Annesley D, Ryan A, Pittock S, Brennan P, Hardiman O, Naughten ER.*
   Identification of micro infarcts in patients with treated homocystinuria: Brain MRI and neurological findings.
   September 2000

3. 13th International Conference on Pterins, Folates and related Biogenic Amines, Maui, Hawaii.
   Yap S, O'Donnell KA, O'Neill C, Naughten ER.
   Inherited Thrombophilia in Classical Homocystinuria: Effects of treatment.
   April 2003

4. Spring Meeting for British Paediatrics Association, York, United Kingdom.
   Mohamed S, Mulhair P, O'Neill C, Naughten E, ***Yap S***, Treacy EP, Mayne PD.
   The outcome of high risk screening for galactosaemia in Ireland, 1989-2003
   April 2004.

5. Spring Meeting for British Paediatrics Association, York, United Kingdom.
   Mohamed S, Reddy K, ***Yap S***, O'Neill C, Mayne PD, Treacy EP.
   Galactosaemia/Duarte (DG) individuals: Is dietary intervention necessary beyond the first few months of life?
   April 2004

6. 4th Asian Congress for Inherited Metabolic Diseases (ACIMD 2015), Taiwan:
   Yap S, Tisna H
   The acute management of intercurrent illness requires adequate caloric intake in Medium Chain Acyl Co-A Dehydrogenase deficiency (MCADD)
   Treatment update on hyperammonaemia.
   March 2015

POSTER PRESENTATION:

1. 37th Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Genoa, Italy
   Yap S*, Naughten ER, Corlett L, Howard P, Irranca M, Howard PM, O'Brien M, Thornton* P.
   The effects of treatment on bone mineral density (BMD) in patients with homocystinuria due to cystathionine β-synthase (CBS) deficiency.
   September 1999
2. 9th meeting of the European Neurological Society, Milan, Italy
   Yap S*, Annesley D, Pittock S, Ryan A, Brennan P, Hardiman O, Naughten ER.*
   Superior sagittal sinus thrombosis and homocystinuria: early anticoagulation is essential.
   June 1999
3. 26th Annual Meeting of the American Association of Paediatric Ophthalmology and Strabismus Meeting, San Diego
   Mulvihill A, ***Yap S****, Naughten ER, O'Keefe M.*
   Ocular findings and delay in diagnosis in patients with late diagnosed or poorly controlled Homocystinuria.
   May 2000
4. VIII International Congress of Inborn Errors of Metabolism, Cambridge, UK
   Yap S*, Barniville G, O'Brien M, Naughten ER.*
   Bone mineral density (BMD) of patients with treated Maple Syrup Urine Disease (MSUD).
   September 2000
5. VIII International Congress of Inborn Errors of Metabolism, Cambridge, UK.
   Yap S*, Barry-Kinsella C, Naughten ER.*
   Pregnancy in pyridoxine non-responsive homocystinuria with good maternal and fetal outcome.
   September 2000
6. VIII International Congress of Inborn Errors of Metabolism, Cambridge, UK
   Yap S*, Rushe H, Howard PM, Naughten ER.*
   Effects of treatment on psychometric parameters in patients with homocystinuria.
   September 2000
7. 5th European Federation of Neurological Societies Congress, Copenhagen, Denmark
   Yap S*, Annesley D, Ryan A, Pittock S, Brennan P, Naughten ER, Hardiman O.*
   MRI findings in patients with treated homocystinuria
   October 2000 (1 of 10 selected for Poster Chairman Discussion)
8. European Paediatric Ophthalmology and Strabismus meeting, Cambridge, U.K.
   Mulvihill A, ***Yap S****, Naughten ER, O'Keefe M.*
   Ocular findings and delay in diagnosis in patients with late diagnosed or poorly controlled Homocystinuria.
   October 2000
9. 27th Annual Meeting of the American Association of Paediatric Ophthalmology and Strabismus Meeting, Orlando, Florida
   Mulvihill A, ***Yap S****, Naughten ER, O'Keefe M.*
   Ocular axial length measurements in homocystinuria patients with and without ocular changes
   May 2001

10. 3rd International Conference on Homocysteine Metabolism, Sorrento, Italy,
***Yap S**, Boers GHJ, Wilcken B, Wilcken DEL, Brenton DP, Lee PJ, Walter JH, Howard PM, Naughten ER.*
The vascular outcome of long-term treated patients with cystathionine β-synthase deficiency: A multicentre observational study.
July 2001

11. 3rd International Conference on Homocysteine Metabolism, Sorrento, Italy
***Yap S**, O'Donnell KA, O'Neill C, Naughten ER..*
Coexistence of Factor V Leiden, Prothrombin G20210A, Methylenetetrahydrofolate reductase C677T and A1298C polymorphisms in patients with cystathionine β-synthase deficiency: Effects of treatment on the vascular outcome.
July 2001

12. 3rd International Conference on Homocysteine Metabolism, Sorrento, Italy
***Yap S**, O'Donnell KA, O'Neill C, Naughten ER.*
Coexistence of Methylenetetrahydrofolate reductase C677T and A1298C polymorphisms, Factor V Leiden and Prothrombin G20210A in obligate heterozygotes for cystathionine β-synthase deficiency.
July 2001

13. 3rd International Conference on Homocysteine Metabolism, Sorrento, Italy,
***Yap S**, Rushe H, Howard PM, Naughten ER.*
Effects of long-term treatment on the intellectual outcome of patients with cystathionine β-synthase deficiency.
July 2001

14. 3rd International Conference on Homocysteine Metabolism, Sorrento, Italy
***Yap S**, Barry-Kinsella C, Naughten ER.*
Successful management of maternal pyridoxine nonresponsive homocystinuria due to cystathionine β-synthase deficiency.
July 2001

15. 3rd International Conference on Homocysteine Metabolism, Sorrento, Italy
*Vyletal P, Kraus JP, Koch H, Kluijtmans LAJ, Blom H, Boers G, Skovby F, Gaustadnes M, Wilcken B, Wilcken DE, Andria G, Sebastio G, Naughten ER, **Yap S**, Ohura T, Pronica E, Laszlo A, Kozich V.*
Origin of CBS alleles that carry the common pathogenic mutation c.833T>C (I278T).
July 2001

16. 39th Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Prague, Czech Republic
***Yap S**, Mulvihill A, O'Keefe M, Howard PM, Naughten ER.*
The effects of early treatment and biochemical control on the long-term ocular outcome of patients with cystathionine β-synthase deficiency.
September 2001

17. 39th Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Prague, Czech Republic
***Yap S**, Mulvihill A, O'Keefe M, Howard PM, Lanigan B, Naughten ER.*
Ocular axial lengths in cystathionine β-synthase deficient patients with and without ocular changes: effects of treatment and control.
September 2001

18. 40th Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Dublin,

Ireland.
Yap S*, O'Donnell KA, O'Neill C, Naughten ER*
Coexistence of Factor V Leiden, Prothrombin G20210A, MTHFR C677T and A1298C polymorphisms in patients with homocystinuria (HCU): effects of treatment on the vascular outcome
September 2002

19. 40th Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Dublin, Ireland
Yap S*, O'Donnell KA, O'Neill C, Naughten ER*
Coexistence of MTHFR C677T and A1298C polymorphisms, factor V Leiden and prothrombin G20210A in obligate heterozygotes for cystathionine β-synthase deficiency
September 2002

20. 41st Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Amsterdam, The Netherlands.
Yap S, *Chew HB, Kluijtmans LAJ.*
Pyridoxine (B6) responsive phenotype associated with Cystathionine β-Synthase (CβS) 919G→A (G307S) genotype.
September 2004

21. 41st Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Amsterdam, The Netherlands.
Mohamed S, Reddy K, ***Yap S,*** O'Neill C, Rogers Y, Mayne PD, Treacy EP.
Long-term dietary management and outcome of Galactosaemia/Duarte (D/G) cases.
September 2004

22. 41st Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Amsterdam, The Netherlands.
Mohamed S, Mulhair P, O'Neill C, Naughten E, ***Yap S***, Treacy EP, Mayne PD.
Is high-risk screening for Classical Galactosaemia in Ireland effective?
September 2004

23. 10th Congress of the European Haematology Association, Stockholm, Sweden.
O'Donghaile D, Nolan B, McCauley M, Fowler B, ***Yap S.***
Severe hyperhomocystiainaemia resulting in recurrent venous thromboembolism, in a patient with MTHFR 677C>T homozygosity and combined vitamin B_{12} and folate deficiency.
June 2005

24. 5th International Meeting on Homocysteine Metabolism, Milan, Italy.
Yap S, Hogan B, Irranca M, Collins P, Ryan S.
Carotid artery ultrasonagraphic findings, lipoprotein (a), serum lipid and dietary fat intake profiles in Classical Homocystinuria: Effects of chronic treatment and biochemical control.
June 2005

25. 5th International Meeting on Homocysteine Metabolism, Milan, Italy.
Yap S, O'Donghaile D, Nolan B, McCauley M, Fowler B.
Severe hyperhomocystiainaemia resulting in recurrent venous thromboembolism, in a patient with MTHFR 677C>T homozygosity and combined vitamin B_{12} and folate deficiency..
June 2005

26. 42nd Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Paris, France.
Yap S, Hogan B, Irranca M, Collins P, Ryan S.
Carotid artery ultrasonagraphic findings, lipoprotein (a), serum lipid and dietary fat intake profiles in Classical Homocystinuria: Effects of chronic treatment and biochemical control.
September 2005

27. 42nd Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Paris, France.
Hughes J, Clark A, Geoghegan O, O'Toole J, Hendroff U, Rogers Y, Walsh O, O'Regan M, Stenson C, Lambert D, ***Yap S***, Manning R, Treacy EP.
A study on outcomes in siblings with classical galactosaemia in Ireland.
September 2005

28. 42nd Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Paris, France.
McGovern L, Hovey J, O'Toole J, Clark A, Stenson C, O'Regan M, Rizvi S, ***Yap S***, O'Donnell, O'Neill C, Treacy EP.
Genotype and early education predicts dietary compliance in adults with PKU.
September 2005

29. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Birmingham, UK
Yap S, Abdullah IS, Ngu LH, The SH, Ong SY, Chong SY, Boey CCM, Lee WS, Mohamed Z.
Molecular spectrum of Glycogen Storage Disease Type III in Malaysian patients.
September 2012

30. 14th Asian Pacific Congress of Paediatrics and 4th Asian Pacific Congress of Pediatric Nursing , Kuching, Malaysia.
A Treatable Cause Of Global Developmental Delay And Hypotonia Due To Cobalamin C Defect
Fadzlina AA, Ong SY, Haszlin H, Sthaneswar P, Abdullah IS, Teh SH, Mohamed Z, ***Yap S***
September 2013

30. 14th Asian Pacific Congress of Paediatrics and 4th Asian Pacific Congress of Pediatric Nursing, Kuching, Malaysia.
Acute Management of Metabolic Encephalopathy and Early Diagnosis of Isolated Methylmalonic Aciduria (MMA) in a Sick Neonate.
Ong SY, Tay CG, Sthaneswar P, Fadzlina AA, Arliena MA, Karmila AB,
Haszlin H, Siti Hawa MT, ***Yap S***.
September 2013

31. RCPCH 2013 Annual Conference, Trainee Poster Session, Glasgow, UK
An Interesting Case of Developmental Delay and Seizures
E Binham, C Vas, C Rittey, S Olpin, N Manning, E Nagmeldin, ***S Yap***
June 2013

32. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain.
Diagnostic Difficulties in Glutaryl CoA-Dehydrogenase Deficiency
Croft J, Clark S, Manning NJ, Bohnam JR, Cleary M, Scott C, Alfadhel M, AlBalwi M, Allen KE, Kirk R, Catchpole A, Hind H, Sharrard M, ***Yap S***, Olpin SE.
September 2013.

33. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain
Trimethylaminuria (TMAU): the apparent prevalence of the secondary/non-genetic form presenting in the UK population.
Manning NJ, Allen KE, Bohnam JR, Croft JM, Edwards LA, Gillett GT, Kirk RJ, Olpin SE, Sharrard MJ, Smith EJ, Watkinson JM, ***Yap S***, Lachmann R.
September 2013.

34. British Paediatric Neurology Association Conference, Winchester, UK
Neonatal molybdenum cofactor deficiency mimicking hyperekplexia and mitochondrial (MELAS) disorders.
Rittey L, ***Yap S***, Alix J, Sarrigiannis PG, Hart AR.
January 2014.

35. British Paediatric Neurology Association Conference, Winchester, UK
MRS – a valuable diagnostic modality for nonketotic hyperglycinaemia (NKH)
Sharma R, Thomas R, Connolly DJA, Olpin S, ***Yap S***, Desurkar A
January 2014

36. 9th International Society for Newborn Screening (ISNS) European Regional Meeting, Birmingham, UK.
Markedly Elevated C6OH Acylcarnitine, Interfering with C5DC Acylcarnitine in a Newborn Bloodspot Screening Sample in the Screen for GA1
Shakespeare L, Baston J, Bonham J, Dibden C, Downing M, Manning N, ***Yap S***.
October 2014

37. 9th International Society for Newborn Screening (ISNS) European Regional Meeting, Birmingham, UK.
Case Report - Baby survives cardiac surgery pre-diagnosis of MCADD: a novel mutation
Dibden C, Downing M, Shakespeare L, Maloney M, Kirk R, Allen L, Qureshi I, ***Yap S***, Bonham J
October 2014

38. 4th Asian Congress for Inherited Metabolic Diseases (ACIMD 2015), Taiwan:
The acute management of intercurrent illness requires adequate caloric intake in Medium Chain Acyl Co-A Dehydrogenase deficiency (MCADD)
Yap S, Tisna H
March 2015

39. RCPCH Annual Conference: British Paediatric Respiratory Society and Association for Paediatric Palliative Medicine and Paediatric Intensive Care Medicine, Birmingham, UK.
Mind the gap! elevated anions secondary to paracetamol and sepsis.
Hulley S, Perring J, Maning N, Olpin S, ***Yap S***.
April 2015

40. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Lyon, France.
Serine deficiency – serial monitoring and response to treatment.
Croft JM, Leong HY, Desurkar A, Watkinson J, Smith L, ***Yap S***.
September 2015

41. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Lyon, France.
Early developmental outcome in Glutaric Aciduria type 1 following diagnosis in UK newborn screening.
Hart AR, Bohnam J, ***Yap S***
September 2015

42. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Lyon, France.
Fatty acid oxidation flux data from 304 symptomatic patients diagnosed with a range of fatty acid oxidation disorders facilitates the prediction of phenotype in screen positive babies from Newborn Screening program.
Olpin SE, Webb JM, Clark S, Dalley J, Hind H, Croft J, Coyler S, Manning N, Scott C, Kirk R, Bohnam J, Dowling M, ***Yap S***, Glamuzina E, Sharrard M
September 2016

43. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Lyon, France.
Biochemical and molecular study of carnitine-acylcarnitine translocase deficiency in a neonate with hyperammonaemic encephalopathy.
Leong HY, Ngu LH, Abd Azize NA, Yakob Y, Habib A, Md Yunus Z, Catchpole A, ***Yap S***, Olpin S.
Sepetmber 2015

44. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Lyon, France.
The value of combined biochemical and genetic testing in suspected SCAD deficiency: case reports of three families.
Kirk RJ, Leong HY, Croft JM, Gregersen N, Watkinson J, Nesbitt IM, Olpin SE, ***Yap S***
September 2015

45. 38th Annual Meeting of European Society of Neuroradiology (ESNR), Naples, Italy
An unusual cause of Stroke in Young patients: MRI findings in cerebrovascular disease in Patients with treated homocystinuria.
Yap S, Ryan A, Gallagher R, Srivastava S, Pittock S, Brennan P, Hardiman O, Annesley-Williams, D.
September 2015

46. 14th International Child Neurology Congress (ICNC), Amsterdam, The Netherlands
Serine Deficiency Disorders – Case series from a Tertiary Pediatric Neurology Center, UK
Ong MT, Kaur S, Wiame E, Croft J, Hart A, Baxter PS, ***Yap S***, Sharrard, M, Desurkar A
May 2016

47. World Congress on Paediatric Intensive and Critical Care, Toronto, Canada
Newborn Screening, nutritional management and emergent haemodiafiltration facilitates successful outcome in Maple Syrup Urine Disease.
Riddell R, Jackman L, Watkinson J, Thomas R, Joseph UC, Blythe D, Hart A, ***Yap S***
June 2016

48. 30th Annual Paediatric Intensive Care Society (PICS) Conference, Southampton, UK
Newborn screening, nutritional management and emergent haemofiltration facilitates successful outcome in Maple Syrup Urine Disease
Riddell R, Thomas R, Jackman L, Watkinson J, Cherachathoor Joseph U, Blythe D, Hart A, ***Yap S***

49. Indonesian Annual Paediatric Society Conference, Makassar, Indonesia.
Poor feeding and excessive weight loss leading to a false positive newborn screening for Medium Chain Co-A Dehydrogenase deficiency (MCADD)
Gultom L, ***Yap S***

BOOK CHAPTER AUTHORSHIP

1. *Boers GHJ,* ***Yap S****, Naughten E, Wilcken B.*
   The treatment of high homocysteine concentrations in homocystinuria. Biochemical control and their vascular outcome.
   In Robinson K. ed. Homocysteine and vascular disease. Kluwer Publications 2000, Chapter 22, pp 387-409.

2. ***Yap S****, Naughten E.*
   Treatment of homocystinuria: State of the Art. In III SHS Symposium on Inborn Errors of Metabolism, Molecular Pathology of Homocystinuria.
   SHS Publications 2001, Chapter 7, pp 79-88.

3. ***Yap S***
   Classical Homocystinuria. An Information Booklet for Adult Patients, Families and Carers of patients.
   Orphan Europe Publication June 2008.
   - Translated into Arabic, German and French.

4. Cohen MC, ***Yap S***, Olpin SE.
   Deaths: Inherited Metabolic Disease and Sudden Unexpected Death - Pathology. In Jason PJ and Roger B (eds). Encyclopaedia of Forensic and Legal Medicine 2nd edition, vol 2: pp. 85-95. Oxford: Elsevier 2016.

5. ***Yap S***
   Inherited Metabolic Disorders. In Lee WS, Tay CG and Lum SH (eds). Textbook of Paediatrics and Child Health, Chapter 73. University Malaya Press 2019.

6. ***Yap S***
   Hyperammonaemia: A Clinician's Perspective on Diagnosis and Management.
   Springer Health Care 2019

AUTHORSHIP FOR ONLINE DATABASE

1. ORPHANET

   - A relational database on rare diseases funded by The European Commission.
   - The Orphanet encyclopedia was launched in May 2003

Abstract:
Yap S
Homocystinuria due to cystathionine β-synthase deficiency
Updated: July 2007
Orphanet number: ORPHA394
http://www.orpha.net//consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=394

Full Article:
Yap S
Homocystinuria due to cystathionine β-synthase deficiency
Scientific editor: Professor Jean-Marie Saudubray
Creation date: May 2003
Updated: February 2005
http://www.orpha.net/data/patho/GB/uk-CbS.pdf

SCIENTIFIC PUBLICATIONS – ORIGINAL ARTICLES

1. ***Yap S***, *Duraisamy G.*
   The Clinical Use of Leucocytes Depleting Filters in the Multiply Transfused Patients - A Case Report.
   Medical Journal Malaysia, Vol.47, No.2, June 1992.

2. Consensus Statement for the GUIDELINES on the use of Blood and Blood Products
   Consensus Statement Group, Ministry of Health, Malaysia 1993.

3. ***Yap S***, *Monavari AA, Thornton P, Naughten E.*
   Late infantile form of 3-Methylcrotonyl CoA –Carboxylase Deficiency presenting as Global Developmental Delay.
   J Inher Metab Dis 1998; 21: 175-6.

4. ***Yap S***, *Naughten E.*
   Homocystinuria due to cystathionine β-synthase deficiency in Ireland: 25 years' experience of a newborn screened and treated population with reference to clinical outcome and biochemical control.
   J Inher Metab Dis 1998; 21: 738-47.

5. *Naughten ER,* ***Yap S***, *Mayne PD.*
   Newborn screening for homocystinuria: Irish and worldwide experience.
   Eur J Pediatr 1998: 157(Suppl 2): S84-7.

6. *Gallagher PM, Naughten E, Hanson NQ, Schwichtenberg K, Bignell M, Yuan M, Ward P,* ***Yap S***, *Whitehead AS, Tsai M.*
   Characterisation of mutations in the cystathionine β-synthase gene in Irish patients with homocystinuria.
   Mol Genet Metab 1998; 65(4): 298-302.

7. ***Yap S***, *O'Donnell KA, O'Neill C, Mayne PD, Thornton P, Naughten E.*
   Factor V Leiden (Arg506Gln), a confounding genetic risk factor but not mandatory for the occurrence of venous thromboembolism in homozygotes and obligate heterozygotes for cystathionine β-synthase deficiency.
   Thromb Haemost 1999; 81: 502-5.

8. ***Yap S***, *Naughten ER, Wilcken B, Wilcken DEL, Boers GHJ.*
   Vascular complications of severe hyperhomocysteinaemia in patients with homocystinuria: effects of homocysteine-lowering therapy. In GHJ Boers, JJ Michiels,eds. Hyperhomocysteinaemia as a risk factor in arterial disease and venous thromboembolism.
   Seminars in Thrombosis and Haemostasis 2000; 26 (3): 335-40 Review.

9. ***Yap S***, *Barry-Kinsella C, Naughten ER.*
   Maternal pyridoxine nonresponsive homocystinuria: The role of dietary management and anticoagulation.
   Br J Obstet Gynaecol 2001; 108: 425-8.

10. ***Yap S***, *Rushe H, Howard PM, Naughten ER.*
The intellectual abilities of early treated individuals with pyridoxine nonresponsive homocystinuria due to cystathionine β-synthase deficiency.
J Inher Metab Dis 2001; 24(4): 437-47.

11. *Mulvihill A,* ***Yap S****, Naughten ER, Howard PM, O'Keefe M.*
Ocular findings among patients with late diagnosed or poorly controlled homocystinuria compared to a screened well-controlled population.
J Am Assoc Pediat Ophthal & Strab 2001; 5(5): 311-5.

12. ***Yap S****, Boers GHJ, Wilcken B, Wilcken DEL, Brenton DP, Lee PJ, Walter JH, Howard PM, Naughten ER.*
Vascular outcome in patients with homocystinuria due to cystathionine β-synthase deficiency treated chronically: A multicentre observational study.
Arterioscl Thromb Vasc Biol 2001; 21(12): 2080-5.

13. ***Yap S***
Classical Homocystinuria: Vascular risks and its prevention
J Inher Metab Dis 2003; 26: 259-65

14. *Mulvihill A, O'Keefe M,* ***Yap S,*** *Naughten E, Howard P, Lanigan B*
Ocular axial length in homocystinuria patients with and without ocular changes: Effects of early treatment and biochemical control.
J Am Assoc Pediat Ophthal & Strab 2004; 8(3): 254-8

15. *Vyletal P, Sokolova J, Cooper DN, KrausJP, Krawczak M, Pepe G, Rickards O, Koch HG, Linnebank M, Kluijtmans LAJ, Blom H, Boers GHJ, Gaustadnes M, Skovby F, Wilcken B, Wilcken DEL, Andria G, Sebastio G, Naughten ER,* ***Yap S****, Ohura T, Pronica E, Laszlo A, Kozich V.*
Diversity of Cystathionine β-Synthase Haplotypes bearing the Most Common Homocystinuria Mutation c.833T>C: A Possible Role for gene Conversion
Human Mutation 2007; 28(3): 255-264

16. ***Yap S***
Classical Homocystinuria: Newborn Screening with Early Treatment Effectively Prevents Complications.
Hamdan Medical Journal 2012; 5: 351-362. (http://dx.doi.org/10.7707hmj.v5i3.191)

17. ***Yap S***
Varied Phenotype of Homocystinuria: Possible Diagnostic Error.
Indian J Ophthalmol. 2014 Jul;62(7):835. doi: 10.4103/0301-4738.138637.

18. Tay CG, Ariffin H, ***Yap S***, Rahmat K, Sthaneshwar P, Ong LC.
Succinic Semialdehyde Dehydrogenase Deficiency in a Chinese Boy: A Novel ALDH5A1 Mutation With Severe Phenotype.
J Child Neurol. 2015; 30(7): 927-931 [DOI: 10.1177/0883073814540523]

19. Calpena E, Deshpande AA, ***Yap S***, Kumar A, Manning NJ, Bachhawat AK, Espinós C.
New insights into the genetics of 5-oxoprolinase deficiency and further evidence that it is a benign biochemical condition.
Eur J Pediatr. 2014 Aug 17. [Epub ahead of print. DOI: 10.1007/s00431-014-2397-0]
Eur J Pediatr. 2015; 174 (3): 407-11

20. Hulley SL, Perring J, Manning N, Olpin S, ***Yap S***
Transient 5-oxoprolinuria: unusually high anion gap acidosis in an infant.
Eur J Pediatr 2015; 174: 1685-1688 [DOI 10.1007/s00431-015-2585-6]

21. ***Yap S***, Leong HY, Fadzlina AA, Haszlin H, Sthaneshwar P, Teh SH, Ili SA, Ngu LH, Zulqarnian M
N-Carbamylglutamate is an effective treatment for acute neonatal hyperammonaemia in a patient with methylmalonic aciduria.
Neonatology 2016; 109: 303-307 – [DOI: 10.1159/000443630]

22. Tanjung C, Watkinson J, Olpin S, ***Yap S***
Lysinuric Protein Intolerance: A treatable cause of developmental delay with multiple complications.
Pediatr Neonat Bio 2016; 1(1): 000103

23. Morris AA, Kožich V, Santra S, Andria G, Ben-Omran TI, Chakrapani AB, Crushell E, Henderson MJ, Hochuli M, Huemer M, Janssen MC, Maillot F, Mayne PD, McNulty J, Morrison TM, Ogier H, O'Sullivan S, Pavlíková M, de Almeida IT, Terry A, ***Yap S***, Blom HJ, Chapman KA.
Guidelines for the diagnosis and management of cystathionine beta-synthase deficiency
J Inherit Metab Dis. 2017 Jan;40(1):49-74. DOI:10.1007/s10545-016-9979-0. Epub 2016 Oct

24. Martinello K, Hart A, ***Yap S***, Mitra S, Robertson NJ
Management and investigation of neonatal encephalopathy – 2017 update
Arch Dis Child Fetal Neonatal Ed 2017 Apr 6. pii: fetalneonatal-2015-309639.
DOI: 10.1136/archdischild-2015-309639. [Epub ahead of print]

25. Piercy H., Machaczek K., Ali P., ***Yap S***.
Parental experiences of raising a child with medium chain acyl-CoA dehydrogenase deficiency (MCADD)
Global Qualitative Nursing Research 2017; vol 4: 1-8. (DOI: 10.1177/2333393617707080)

26. ***Yap S***, Annesley-Williams D, Hardiman O
Cerebral venous sinus thrombosis in homocystinuria: Dietary intervention in conjunction with anticoagulation.
SAGE Open Medical Case Reports 2017; vol 5: 1-4. (DOI: 10.1177/2050313X17722289)

27. ***Yap S***, Gougeard N, Hart AR, Barcelona B, Rubio V
N-Carbamoylglutamate-responsive carbamoyl phosphate synthase 1 (CPS1) deficiency: A patient with a novel CPS1 mutation and an experimental study on the mutation's effects.
JIMD reports 2019; 1-9, https://doi.org/10.1002/jmd2.12034

28. Abdullah IS, Teh SH, Khaidizar FD, Ngu LH, Keng WT, ***Yap S***, Mohamed Z
Intron retention is among six unreported AGL mutations identified in Malaysian GSD III patients.
Genes Genomics 2019; Apr 26. doi: 10.1007/s13258-019-00815-9.

29. Piercy H, Yeo M, ***Yap S***, Hart AR.
What are the information needs for a parent caring for a child with Glutaric Aciduria type 1
BMC Pediatrics 2019; 19: 349. Doi.org/101186/s12887-019-1742x

30. ***Yap S***, Vara R, Morais A**.**
Post-transplantation outcomes in patients with PA or MMA: A review of the literature
Adv Ther 2020; 37: 1866-1896. Doi.org/10.1007/s12325-020-01305-1

31. ***Yap S***, Garcia-Cazorla A, Vara R, Alfadel MA, Burlina A, Cazzorla C**.**
Exploring long-term strategies for patients with propionic and methylmalonic acidurias.
Key Opinions in Medicine –metabolic disorders 2020; Vol 1 Issue 2: 1-16

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2. ***Yap S****, Naughten ER, Annesley D, Ryan A, Pittock S, Brennan P, Hardiman O.*
   Successful management of superior sagittal sinus thrombosis in a patient with homocystinuria.
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3. ***Yap S****, O'Donnell KA, O'Neill C, Naughten ER.*
   Coexistence of Factor V Leiden A506G, Prothrombin G20210A, Methylenetetrahydrofolate reductase C677T and A1298C mutations in homozygotes of cystathionine β-synthase deficiency: Effects of treatment on the vascular outcome.
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