Medical Genetics in Hong Kong & Networking in Asia

Stephen TS Lam
Head, Clinical Genetic Service
Department of Health, Hong Kong
BASIC EQUIPMENTS IN CYTOGENETICS
Medical Genetics in Hong Kong

- Clinics & Laboratories in Universities & Hospitals
- Clinical Genetic Service, Prenatal Diagnosis Service
- Parents’ Groups, Patients’ Organizations, NGO
Clinical Genetic Service (since 1981)

- Genetic diagnosis and counselling
- Genetic laboratory
- Genetic health promotion
- Genetic screening
- Treatment of genetic diseases
- Initiatives in community genetics
Genetic Counselling Unit

- 1981(Sept)-2006 (June), a total of 23,505 families referred for genetic diagnosis and counselling
- 1000 new families each year
- 70% of referrals from paediatric age group
- About 600 different disease classifications in register
Cytogenetic Investigations

1981-2004
(abnormalities detected from 21558 studies)

- G banding and metaphase (3722/14285, 26%)
- Prometaphase (994/3615, 27%)
- Fragile X (8/765, 1%)
- DEB fragility
- Sister Chromatid Exchange (2/245, 1%)
- Late replicating X
- C staining, G11, silver staining
- Q banding
- DAPI studies
- Fluorescence in-situ hybridization studies
  (424/1695, 25%)
Genetic Health Promotion

- Monthly genetic seminars in four major teaching hospitals
- Lectures for medical students and doctors
- Meeting the media
- Dialogue with patient groups
- Professional societies
- Networking with centers in neighboring countries and regions
Alma Ata – Kazakstan
Neonatal Screening as Part of a Comprehensive Genetic Service

- Territory – wide screening since 1984
  G6PD deficiency
    - Quantitative enzyme assay
      cord blood
    - Incidence: male 4.5%, female 0.3%
  Congenital hypothyroidism
    - Cord blood TSH assay
    - Incidence: 1 : 2,146
- Cover 99% of NB screened, about 50,000/yr
- Free of charge, funded by government
Molecular Genetic Studies

• Indications
  – For suspected single gene disorders affecting different systems / organs of body
  – Presently 80 different types of disorders can be investigated at Clinical Genetic Service

• Methodologies
  – DNA extraction
  – Gene amplification by PCR
  – Southern blotting
  – Protein truncation test
  – Screening (SSCP, DHPLC, MLPA)
  – Gene sequencing
Gaucher disease with pulmonary involvement in a 6-year-old girl: Report of resolution of radiographic abnormalities on increasing dose of imiglucerase

Shing Yan Robert Lee, MRCP(UK), Alex Wan Cheong Mak, MRCP(UK), Kwai Fun Huen, FRCP, Steven Tak Sum Lam, FRCP, and Chun Bong Chow, FRCPCH
Prenatal Diagnosis Service

• Started 1981 prenatal counselling
• Fetal diagnosis included
  – Imaging (from 2-D since 1981 to 3D 2002)
  – Amniocentesis, fetal blood sampling, chorionic villous sampling
  – TYH Prenatal Diagnostic Laboratory
    • Cytogenetic studies 1981
    • Molecular cytogenetics FISH 1997, qFPCR 1999
    • Molecular diagnostics on thalassaemia, hemophilia, DMD, SMA, Huntington
Prenatal Diagnosis Service

• Screening
  – Neural tube defect, thalassemia 1984
  – Fetal Down syndrome 1994
  – 18-20 week routine obstetric scanning 1995

• Fetal therapy
  – Since 1992, therapy for fetal arrhythmias, anemia, pleural effusion, obstructive uropathy, conjoint / acardiac twin, abnormal twinning, twin to twin transfusion
Public health genetics

• Provision & evaluation of genetic services
• Education of professional & public
• Advocacy
• Policy & regulatory
• Research
• ELSI
Second Sino-German Interdisciplinary Symposium about Medical Ethics in China
Shanghai, 19-23 October 1999
History of APSHG

• 1994  1st Asia Pacific Conference of Medical Genetics (Bangkok)
• 1995  2nd Conference (Jakarta), maintained a network
• 1997  3rd Conference (Kuala Lumpur), planned to start a Society
• Conjoint meetings with HUGO-Pacific Conference
• 2004  resume planning on forming Society
Objectives of APSHG

• To promote research in basic and applied human and medical genetics
• To integrate professional and public education in all areas of human genetics
• To provide a forum where scientists can share their research findings as well as increase or spread knowledge and understanding of human and medical genetics among the various professionals including health professionals, health policy makers, legislators and the general public
THE INAUGURAL MEETING OF THE ASIA PACIFIC
SOCIETY OF HUMAN GENETICS (APSHG)
NOVEMBER 12, 2005
ROYAL ORCHID SHERATON HOTEL
BANGKOK, THAILAND
Inaugural Meeting Bangkok Nov 2005

• Executive Committee elected
  – President            Pornswan Wasant
  – President Elect   Carmencita Padilla
  – Secretary            Stephen Lam
  – Treasurer            Ivy Ng

• Council members
  – MK Thong, Paul Hwu, ST Chen