

Medical Genetics in Hong Kong & Networking in Asia

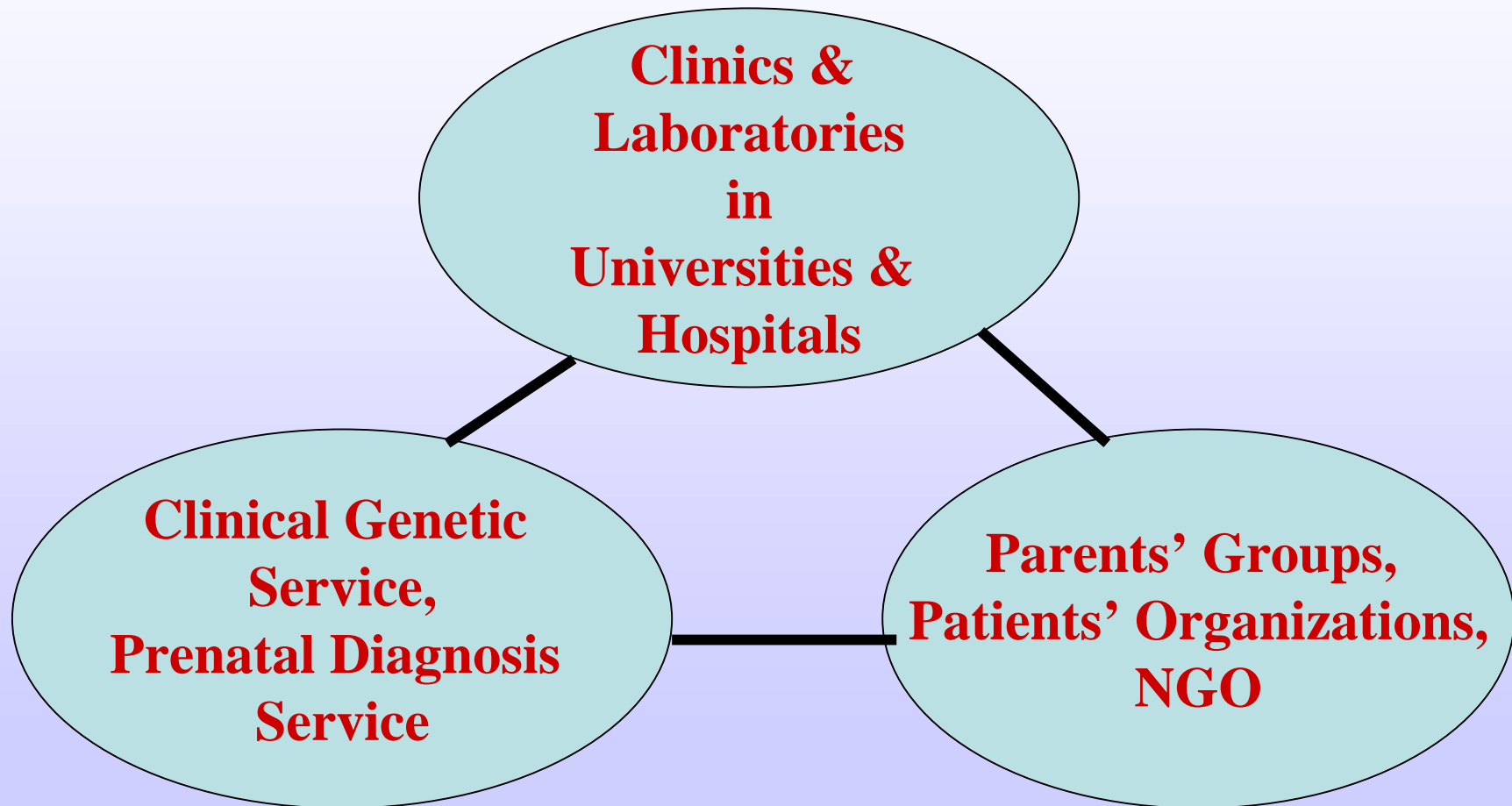
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Medical Genetics in Hong Kong



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

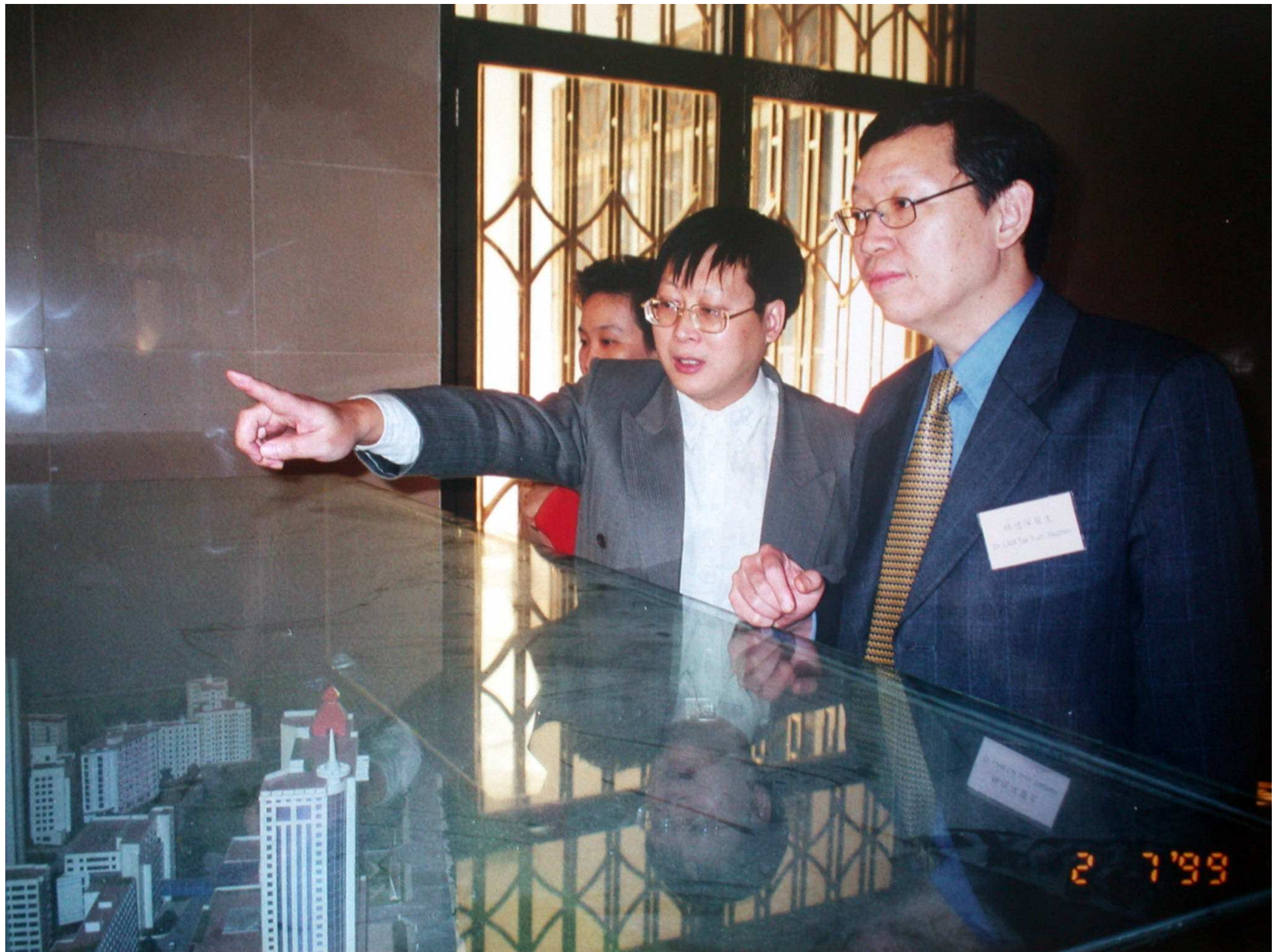
1981-2004

(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



2005

际研讨会



楨先生



上海市遗传学会
保健中心

Alma Ata – Kazakhstan







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% NB screened, about 50,000/yr
- Free of charge, funded by government



BANQUET & THAILAND NIGHT

3rd APRM OF ISOIS

NOVEMBER 17, 1998

AN

Regional Meeting of International Society for Neonatology

September 14-17, 2004

Shanghai, China



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

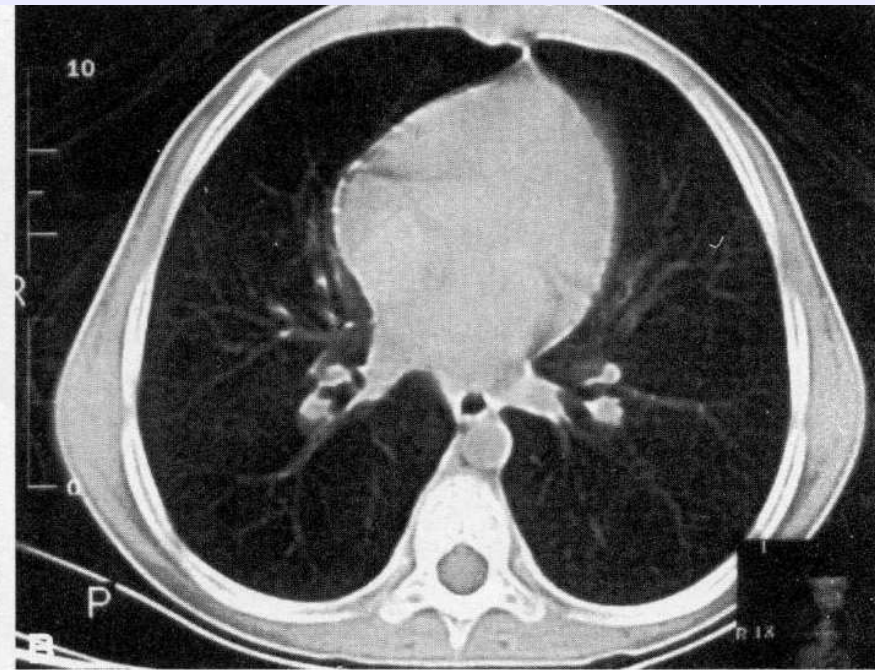
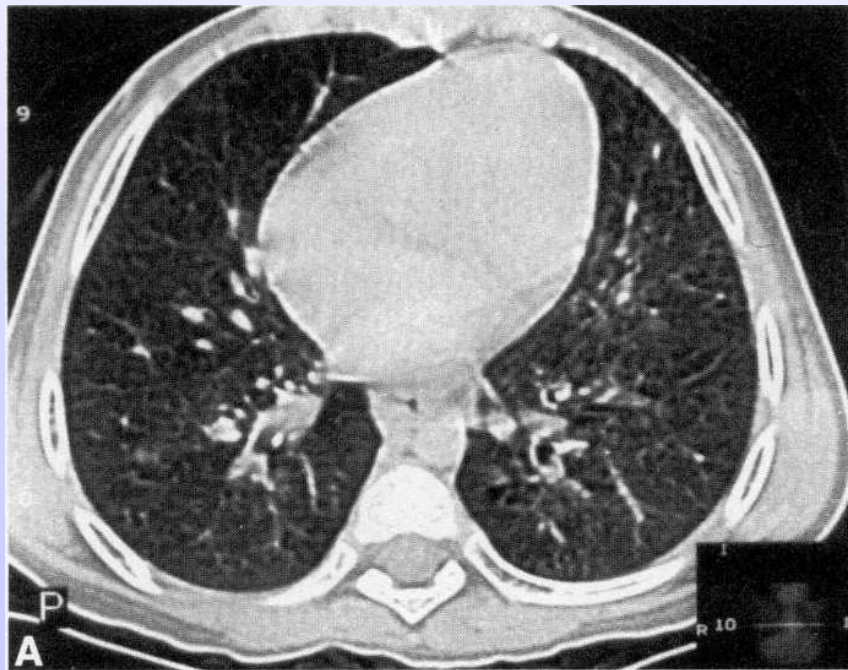
- **Methodologies**

- 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, hemophilias, 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

中欧医学伦理学国际学术研讨会

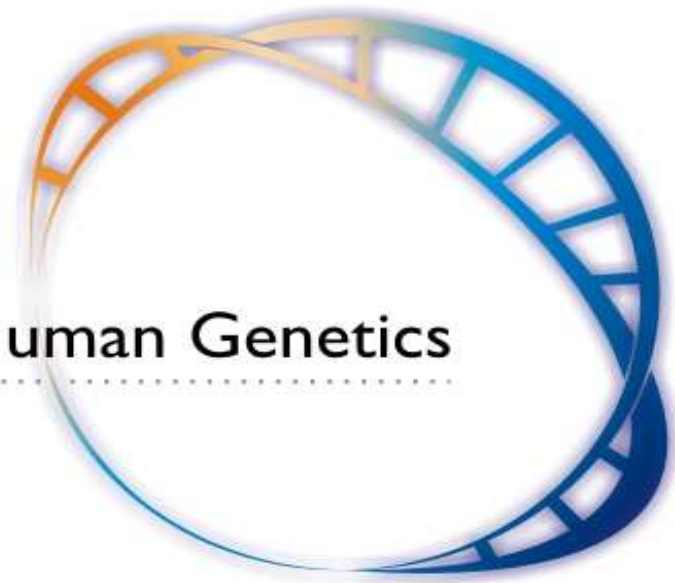
上海 1999.10.19-23

'Second Sino-German Interdisciplinary
Symposium about Medical Ethics in China
Shanghai, 19-23 October 1999





Asia Pacific Society of Human Genetics



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

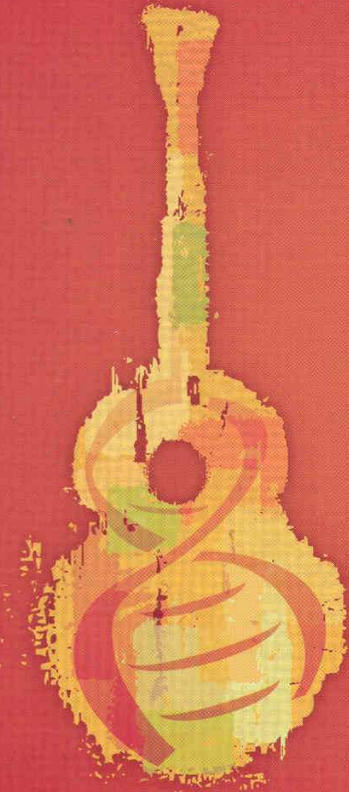


NEW PARADIGMS IN MEDICAL GENETICS
DEPARTMENT OF HEALTH & HKSMG



7th Human Genome Organization (HUGO)-Pacific Meeting
and 8th Asia-Pacific Conference on Human Genetics

www.HUGO-AP2008.ph



HUGO | AP
2008 



Shangri-La's Mactan
Island Resort
Cebu, Philippines

April 2008