# International Congress of Global Chinese Geneticists 2006

The "International Congress of Global Chinese Geneticists 2006 (ICGCG 2006)" was held in Beijing, China, on August 2-4, 2006 (http://pucmg.pku.edu.cn/icgcg2006.htm). The Congress was co-organized by Peking University, the Association of Chinese Geneticists in America (ACGA), the Chinese Society of Medical Genetics, the Taiwan Human Genetics Society, the Hong Kong Society of Medical Genetics, the National Genome Institute of Singapore, Fudan University, Zhejiang University, and Wenzhou Medical College. It was co-chaired by Professor Nanbert Zhong, who is the Director of Peking University Center of Medical Genetics and Director of Molecular Neurogenetic Diagnostic Laboratory of New York State Institute for Basic Research in Developmental Disabilties, and Professor Li Jin, the Dean of College of Life Sciences of Fudan University, with support of Secretary General Professor Marylin Li, who is the Director of Cytogenetics and Molecular Diagnostic Laboratory at Tulane University. The mission of the Congress is to promote global communication and collaboration among Chinese geneticists and geneticists around the world. Near 200 participants who came from different areas of the world attended the meeting.

A total of 106 abstracts were received. The presentations were designed in various formats including an invited symposium (chaired by Nanbert Zhong), platform presentations, and posters. The session of "Genetic resources, advanced technology and stem cell research" was moderated by Z. Chen and X. Zhang, "Community genetics and public health" by S. Lam, "Clinical cytogenetics" by Y. Fan and M. Li, "Monogenic inheritance and genomic medicine" by LJ Wong, "Immunogenetics and infectious diseases" by TJ Chen, "Molecular testing and prenatal diagnosis" by BL Wu and S. Huang, "Developmental genetics and experimental models" by C. Lau and YQ Zhang, "Population genetics and multigenic diseases" by JJ Liu and L. Jin, "Reproductive genetics and better-bearing" by WY Chan and P. Dai, "Birth defects and child development" by XR Wu and MX Guan, "Pharmacogenomics and proteomics" by TS Huang, "Cancer genetics and signal transduction" by G. Lu and Z. Chen, "Biochemical genetics and newborn screening" by XF Gu and CL Yu, and "Genomics and bioinformatics" by N. Zhong and P. Jin.

Among these sessions, the following topics which were quite unique at this Congress received the highest feedback and were the most attractive.

## Application of microarray in genetic studies

Dr. Caifu Chen, the Director of Gene Expression and Genotyping Research at Applied Biosystems (USA) presented genome-wide microRNA expression profiling in mouse ES and differentiated cells. Dr. Zugen Chen (UCLA, USA) presented pedigree-free identity-by-descent (IBD) mapping of disease genes using dense SNP genotyping, which is using the founder effect to locate the disease risk factor and detect co-inherited "disease" DNA fragments by high-density SNP genotyping. Dr. Huasheng Xiao from the National Engineering Center for Biochip in Shanghai introduced current biochip service in China and compared different biochip platforms. One exciting presentation, given by Dr. Yaoshan Fan, the Director of Cytogenetics laboratory at University of Miami Miller School of Medicine, introduced the application of array CGH in analyzing genome wide duplication/deletion, especially in diagnosis of unexplained mental retardation (MR). In Dr. Fan's presentation, he compared conventional cytogenetics and fluorescence *in situ* hybridization (FISH) with array CGH. Currently mental retardation accounts for 2%-3% of newborns and the underlying cause is unknown in most of cases. Conventional cytogenetic studies usually may detect chromosomal abnormalities in about 5% of patients with mental retardation. FISH may detect subtelomeric imbalance in 3% of patients with unknown reasons. Applying microarray-based comparative genomic hybridization may detect 10% of patients with unexplained MR. Array CGH may also be applied in epigenetic studies, and presenting a new direction in the cytogenetic daily operations, which were addressed by Dr. A. Beaudet.

## Community genetics plays an important role in decreasing birth defects

On behalf of the March of Dimes Birth Defects Foundation, Dr. Chritopher P Howson, Vice President of March of Dimes Global Program, presented a topic of "Care and prevention of birth defects in low- and middle-income countries" in which he introduced the "March of Dimes Global Report on Birth Defects" that is now being translated into the Chinese language and will be published in China soon by the Chinese National Population and Family Planning Commission. He listed the most common five birth defects in the world nowadays, which are congenital heart defects, neural tube defects, the hemoglobin disorders, Down syndrome, and glucose-6-phosphate dehydrogenase (G6PD) deficiency. These five disorders account for 26% of total birth defects. Dr. Howson encouraged the global Chinese geneticists to work on birth defects because, based on a data released in 2001 from China, the prevalence rate of birth defects was as high as 51.2 per 1000 Chinese live births, the annual births with birth defects were 963 997 individuals, and the annual deaths due to birth defects were 402 682. He highly valued the Chinese geneticists in working on care and prevention of birth defects.

Two studies, the "Current status of thalassemia in minority populations in Guangxi" presented by Dr. HF Pan and "Exploring a feasible method to diagnose MR in the rural area of Shanxi province" by Dr. LW Wang, described their experience in dealing with birth defects in regional or rural areas. Nationwide molecular epidemiological investigation of hereditary hearing impairment in China, presented by Dr. P. Dai from PLA hospital, revealed that the idea molecular screening for hereditary deafness in Chinese population should be the loci of GJB2, SLC26A4, and mitochondrial genome. Dr. S. Lam, the Director of Hong Kong Clinical Genetic Service and the President of Hong Kong Society of Medical Genetics summarized their eight-year experience in dealing with congenital anomaly syndromes in Hong Kong area. Among 1043 cases they made clinical diagnosis, Down syndrome

accounts for 57.6%, Turner syndrome for 12.2%, Trisomy 18 for 11.3%, Klinefelter syndrome for 4.9%, Trisomy 13 for 4.0%, Noonan syndrome for 3.1%, hemifacial microsomia for 1.8%, Cornelia de Lange syndrome for 1.7%, Prader Willi syndrome for 1.7%, and Cri du Chat syndrome for 1.6%. These congenital anomaly syndromes were followed by Williams syndrome, Russell Silver syndrome, Potter sequence, Oro-facial-digital syndrome, VACTERL association, Marfan syndrome, Pierre Robin sequence, Fragile X syndrome, ADAM complex, and Rubinstein Taybi syndrome. Obviously, implementing of genetic diagnosis including molecular testing could make this order more accurate.

Dr. E. Hau, representing the Government of Hong Kong Special Administrative Region (HKSAR), concluded that nine elements are essential in dealing with public health genetics. These include genetics, public health, epidemiology, evaluation, ELSI (ethical, legal and social implications), communication and collaboration, health promotion and education, information handling and integration, and policy formulation and implementation.

## Translational research may open new avenues for genetic disorders

Dr. RE Chee, Deputy Director of Genome Institute of Singapore, presented "Host factor regulation of hepatitis B virus replication" in which he showed that knocking-down hnRNP K resulted in lowering HBV viral load and raised an open question whether hnRNP K is a target of drug for suppressing HBV replication. A similar study carried out by Ms. LR Huo, who is a Ph.D. student working on aging process in Dr. Nanbert Zhong's laboratory at the Peking University Center of Medical Genetics showed that knocking-down hnRNP E1 may alter global gene expression profiles. The alteration may be detected at both mRNA level with gene expression biochip and protein level with PF2D platform.

Dr. D. Wallace, a member of American Academy of Sciences at University of California (Irvine) had an overview of evolutionary medicine perspective on mitochondrial etiology for degenerative diseases, cancer, and aging. Dr. J. Qu showed their nice work on Leber's hereditary optic neuropathy in Chinese families. Dr. LJ Wong, the director of mitochondrial laboratory at Baylor College of Medicine, pointed out that majority of mitochondrial proteins are encoded by nuclear genes that take charge of mitochondrial diseases. Dr. MX Guan presented nuclear modifier genes modulate phenotypic expression of deafness associated with mitochondrial 12S rRNA mutation.

Dr. HM Yang, the Director of Genetic Institute at the Chinese Academy of Sciences, overviewed the progress of human genome project in China. Genomic medicine has opened a new avenue in medical practice. Knowing his/her genotype in an individual surely may help in designing a specific clinical management for this person. This individualized clinical management has been referred as personalized medicine. With this concept, a few institutes that are recently offering risk assessment for cancer(s) and cardiovascular disorders in China have been noticed by the meeting attendance, although whether the "risky" genotype would eventually lead to the development of clinical symptom(s) is controversial. The argument is that it seems this type of offer did not pay attention to the scientific issues of the gene-environmental interactions and simply ignored the ethical concerns that "bad" genotype may not develop a serious "bad" disease but could cause psychological burden for the person who participated in the assessment.

Research on lysosomal storage diseases (LSD) has been at the leading position in protein replacement treatment. Dr. YH Chien from Taiwan University Hospital presented their observation on infantile Pompe disease after enzyme replacement therapy (ERT) and concluded that although cardiac hypertrophy responses well to ERT, damages left may still bother the outcome. Therefore, early treatment should be required for a complete therapeutic effect for infantile-onset Pompe disease. Compared to the genotype-driven mouse disease model, Dr. JY Wu from Institute of Biomedical Sciences at Taiwan Academia Sinica presented their phenotype-driven approach with NEU mutagenesis, following biochemical screening with tandem mass spectrometry (TMS). Applying TMS, Dr. XF Gu who is from Shanghai Xin-Hua Hospital has detected 23 disorders in neonatal screening.

Following the format of problem based learning discussion we developed in ICMG 2005 (International conference of medical genetics 2005), which was adjunct to a clinical training course on dysmorphological syndromes organized by the Peking University Center of Medical Genetics and held in Beijing by the summer of 2005, a case presentation on cleidocranial dysplasia (CCD) was provided by Dr. Shaohua Tang in a session of discussing the clinical diagnosis of difficult case(s), chaired by Dr. Taosheng Huang who is a clinical geneticist from UCI (University of California at Irvine).

## Bundle of education with academic research is a very special issue at this Congress

Nearly 50 undergraduate and graduate students participated in this Congress. They were either presenting their research studies or volunteering in the meeting organization. Both researchers who are also professional educators and students communicated actively. Professor A. Beaudet, the chairperson of Dept. Human Genetics at Baylor Medical College, a member of American Academy of Science, introduced graduate training program in his department. Professor LC Tsui, President of Hong Kong University, spent most of his free time in talking with students. He instructed the young generation to open their mind in learning and study. Presidential panel discussion, the last session of the Congress co-chaired by Y. Ke and LC Tsui has been focused on "Genetics/genomics education in Chinese universities/ colleges". Professor Y. Shen, academician and Director of Institute of Basic Medical Sciences in the Peking Union Medical College, encouraged students and young researchers to devote their life in genetics. Finally, Dr. Y. Ke expressed her appreciation to the students who have participated in the ICGCG 2006.

As the first time of effort, ICGCG 2006 was a successful meeting that has brought the global Chinese geneticists together and promoted communication among them. On behalf of the organizer committee, I would like to express our appreciation to all who participated in the meeting, to local organizers who did a beautiful job, and to the financial sponsors who made generous contributions. We are looking forward to getting together in the near future, especially communicating with the Chinese geneticists from Austria and European countries.

*Nanbert Zhong, MD,*
*Peking University Center of Medical Genetics, Beijing, China (pucmg@pku.edu.cn);*
*New York State Institute for Basic Research in Developmental Disabilities, Staten Island, NY, USA*