



Report – 21st century medical genetic and genomic medicine in China

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The First Hangzhou International Symposium on the Medical and Laboratory Applications of Medical Genetics and Genomics was held in Hangzhou from October 6~8, 2005. The symposium was organized and sponsored by the First Affiliated Hospital, School of Medicine, Zhejiang University and co-sponsored by the Zhejiang Medical Association; School of Medicine of Zhejiang University; the James Watson Institute of Genomics Sciences and Beijing Genomics Institute/Huada Genomics Center of the Chinese Academy of Science. Many internationally prominent geneticists were invited to the meeting, including Arno Motulsky, Peter Byers, Douglas Wallace, Judith Hall, and David Weatherall. The invited speakers presented discussions of recent advances in human and medical genetics, genomics, and in cutting-edge research.

Dr. Jun Zhu, the vice president of Zhejiang University attended and congratulated the conveners and participants on the success of the symposium. He met all invited speakers and announced the establishment of the Center for Genetic and Genomic Medicine of Zhejiang University. The center is supported by School of Medicine, Zhejiang University and the James Watson Institute of Genomics Sciences, and is affiliated with the First Affiliated Hospital of Zhejiang University. The mission of the center is to establish a first-class platform that will serve as a bridge with developed countries. The center will use

the advanced technology in genetics and genomics, based in China, to advance research and clinical practice. The center is among the first such platforms that combine research, education, and clinical service. The center is heavily supported by the 985 projects that are part of the Higher Education Improvement Project, which was launched by the Ministry of Education in China. The center will be physically located in the James Watson Institute of Genomics Sciences and School of Medicine, Zhejiang University and will connect biomedical scientists in China and abroad. Some connections have already been established, for example, with the University of Washington, the University of Rochester, Cold Spring Harbor, and the University of California, Irvine. It is expected that the center will serve as a national and international institute for research, education, and clinical services. The researchers plan to take advantage of the unique populations in China and the broad spectrum of disease and rich genetic materials. They will focus on the discovery of novel genetic disorders with Mendelian inheritance, the identification of the disease-causing genes, and will explore the interaction between genes and the environment. The center will be directed by Professor Ming Qi. Dr. Qi is board-certified in clinical molecular genetics in the United States, and he is also an inspector for the American College of Pathology (CAP), the organization that regulates the quality of all genetic laborato-

all genetic laboratories in the US.

At the meeting, the vice president of Zhejiang University, Dr. Zhu Jun, and the Associate Director for the James Watson Institute of Genomics Sciences, Dr. Huanming (Henry) Yang, appointed Drs. Arno Motulsky and Douglas Wallace as guest professors of Zhejiang University. Dr. Motulsky is currently a professor emeritus at the University of Washington and is considered one of the co-founders of the field of medical genetics. He is a member of the National Academy of Sciences in the United States, a fellow of the American Association of Arts and Sciences, and a member of the Institute of Medicine in the United States. Dr. Motulsky is also the founder of the field of pharmacogenomics. Dr. Motulsky and his postdoctoral fellow, Joseph Goldstein, elucidated the genetic mechanism of coronary arterial sclerosis and hyperlipidemia. Dr. Motulsky's research includes metabolic disease, epidemiology, statistical genetics, and molecular genetics. At the symposium, Dr. Motulsky presented a strategy to identify genetic causes of complex diseases, as well as the advancement of pharmacogenomics. Dr. Motulsky is the chair of the advisory committee of Zhejiang University's Center for Genetic and Genomic Medicine.

Dr. Douglas Wallace is a Donald Bren Professor of Biological Science and Molecular Medicine and the director of the Center for Molecular and Mitochondrial Medicine and Genetics at the University of California, Irvine. He is a member of the National Academy of Sciences, and a fellow of the American Association of Arts and Sciences. He is considered the major innovator in the study of disorders of the mitochondrial genome. His research includes human evolution, degenerative diseases, cancer, and aging. In 1980, Dr. Wallace first demonstrated that mitochondrial inheritance was strictly maternal in humans and has subsequently identified the mitochondrial genome as the home for a number of complex human genetic disorders that show strictly maternal inheritance. Many molecular genetics and genetics textbooks contain specific chapters on his research. His group established the mitochondrial genetic markers that could be followed in different populations and on the basis of these studies proposed that humans originated in Africa. Currently, his group continues to lead research in degenerative diseases, diabetes, epilepsy, and mental retardation. Most recently, his

group proposed that the accumulation of mutations in the mitochondrial genome is one of the mechanisms of aging. During this symposium, Dr. Wallace launched collaborations with physicians and researchers at Zhejiang University, and he is planning a local research lab at Zhejiang University where he will study: (a) mitochondria and eye disease in the Chinese population; (b) how economic development affects mitochondrial mutations and increases susceptibility to cancer; (c) the relationship between the accumulation of mutations in the mitochondrial genome and aging and degenerative diseases; and (d) the mitochondrial mutations of diabetes.

Other renowned attendees included Dr. Wilson Lo, the father of medical genetics in China and currently a professor at Peking Union Hospital; the vice president of the Chinese Medical Genetics Association, Dr. Shanzhi Huang, Chairman of Medical Genetics at Peking Union Hospital, Dr. Xue Zhang, Emeritus President of the Chinese Pediatrics Association, Dr. Xiru Wu, and the Director of the Center for Medical Genetics at Beijing University, Dr. Nanbert Zhong. The president of the American Chinese Human Genetics Association, Dr. Zhong Chen, sent a letter of congratulations to the symposium. The leader of Zhejiang Province's Science and Technology Commission, Lian Yu, also attended the opening ceremony.

The president of the American Society of Human Genetics and the former editor of the *American Journal of Human Genetics*, Dr. Peter Byers, talked about disease associated with collagen gene mutations. He discussed the past, present, and future of human genetics and emphasized the application of the Human Genome Project. He pointed out that after the completion of human genome sequencing, the following will be the directions of the human genetics field: (a) genetic variation in the population; (b) phenotypic variations in populations and the relationships to genetic variables; and (c) prediction of behavior, particularly in environment and gene interaction. At the meeting, Dr. Byers was invited to be one of two editors-in-chief of *Journal of Zhejiang University Science B*.

Sir David Weatherall, Emeritus Professor and an honorary director of the Weatherall Institute of Molecular Medicine at Oxford University, a member of the Royal Academy of Sciences, and a foreign

member of the National Academy of Sciences in the United States was a major presence at the meeting. He was appointed as a Fogarty Resident Scholar by the United States National Institute of Health. Dr. Weatherall is an outstanding physician-scientist, who has made tremendous contributions in medical genetics, hematology, pathology, and in clinical medicine. At this symposium, he presented on the human genome and human health. In his talk, he pointed out that developing countries should benefit from the Human Genome Project. Tuberculosis, AIDS, malaria, and malnutrition are still major health issues. Researchers should combine epidemiology, population genetics, and genes and environment interaction and address those major medical issues. China will benefit from the Human Genome Project through improved diagnoses and more effective therapy.

Other experts from the United States and Canada also attended the symposium, including Drs. Judith Hall, Daniel Pinkel, Donna Albertson, Ronald Scott, Edith Cheng, Taosheng Huang, Marilyn Li, and Chunli Yu. They presented discussions of "Congenital Anomalies, Developmental Genetics, and Non-traditional Mendelian Inheritance"; "Cancer Genetics"; "Human Mutations and Disease"; "Genomics and Human Reproduction: Prenatal Diagnosis in the 21st Century"; "Clinical Genetics, and Molecular Cytogenetics and Metabolic Diseases". At the symposium, Huanming Yang, the principle investigator of the Human Genome Project in China disclosed the achievements of the James Watson Institute of Genomics Sciences during the past two years.

On the evening of October 6, all the attendees and invited speakers discussed medical genetics and genomics in China. Dr. Wilson and Dr. Huang examined the history of medical genetics in China and discussed its problems and future perspective. The symposium's organizers expressed their appreciation of all the speakers from other countries willingness to attend the symposium and provide guidance for medical genetics in China. Many attendees pointed out the gap in medical genetics between China and developed countries, and expressed hope that the Chinese government would fully support medical genetics and genomics in China.

During the symposium, many scholars and physicians at Zhejiang University took the opportunity to establish collaborations. The president of

Zhejiang Children's Hospital invited Dr. Hall, a professor at British Columbia University, and Dr. Scott from the University of Washington to be on the members' editorial board of the *World Journal of Pediatrics*. Dr. Hall is a former president of the American Society of Human Genetics, and President Emeritus of the American Academy of Pediatrics.

Many of the invited scholars are excellent researchers as well as outstanding physicians. They specialize in cardiovascular genetics, ob/gyn, cancer genetics, etc. On October 8, at the invitation of Dr. Ming Qi, many physicians, including Drs. Byers, Hall, Scott, Huang, Li, Yu, and Chen, volunteered to see patients at the First Affiliated Hospital at Zhejiang University. The physicians were well received by the Chinese patients.

The symposium became the focus of newspapers, TV reports, and electronic media and increased awareness of medical genetics in China. The attendees discussed and exchanged ideas on human genetics and genomes. A few days after the establishment of the Center of Genetics and Genomics at Zhejiang University, many patients and physicians from different parts of the country wrote, e-mailed, and called the center. Many invited speakers felt that the symposium was extremely important for medical genetics in China and expressed their willingness to attend similar meetings again. Similar meetings should be held periodically to promote genetics and genetic medicine in China, increase international exchange, and promote the development and application of medical genetics and genomics.

Important Lectures in Symposium

Keynote speeches:

1. Peter Byers:
Past, Present and Future of Genomic Medicine
2. David Weatherall:
Genomics and World Health: Hopes and Realities

Special topic reports:

1. Huanming Yang/Taosheng Huang:
Genome Structure, Variation, and Mutation
 - (1)The structure of the genome
 - (2)Genetic variations and polymorphisms
 - (3)Human mutations & Diseases
2. Marilyn Li/Daniel Pinkel:
Medical Cytogenetics

- (1) Types and Frequency of chromosome aberrations
 (2) Clinically important cytogenetic disorders
 (3) Diagnostic methods (conventional karyotypes, FISH, comparative genomic hybridization)
3. Edith Cheng:
 Prenatal Diagnosis and Screening
 Genetic counseling
 Preimplantation diagnosis
4. Ronald Scott/Chunli Yu:
 Human Biochemical Genetics
 (1) Representative disorders
 (2) Carrier screening (conventional and new approaches)
 (3) Therapy (including gene therapy)
5. David Weatherall:
 Hemoglobin Abnormalities (the thalassemias)
 (1) Population genetics, pathogenesis
 (2) Screening and prenatal diagnosis
 (3) Therapy
6. Peter Byers:
 Molecular Mechanisms of Autosomal Dominant Disorders
 Heritable Disorders of Connective Tissue
7. Judith Hall:
 Developmental genetics and diseases. "Unusual" Modes of Inheritance
- Epigenetics (X-inactivation, imprinting)
8. Marilyn Li/Donna Albertson:
 Human Cancer Genetics
 (1) Major cancers, including those relevant to China
 (2) Cancer cytogenetics
 (3) Presymptomatic testing for BRCA1, BRCA2, HNPCC mutations
 (4) Cancer classification by microarray analysis
9. David Weatherall:
 Genetics and Infectious Disease
10. Arno Motulsky:
 Genetics of Complex Disease
 (1) The interaction of genetic and environmental factors in disease causation
 (2) Coronary artery disease, hypertension, and diabetes
 (3) Susceptibility testing
11. Douglas Wallace:
 Mitochondrial Genetics
 (1) Inheritance, markers and disease
12. Arno Motulsky/Ming Qi:
 Pharmacogenetics and Ecogenetics
 (1) Principles, polymorphisms and personalized medicine
 (2) Role of molecular testing



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