Building Reproductive Genetic Services from Bottom Up: Over 30-year Experience of a Major Prenatal Diagnostic Center in Guangdong Province

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INTRODUCTION
The Department of Medical Genetics of Guangdong Women and Children’s Hospital (GDWCH) was started in 1981. At that time, it was a small unit with only three staffs performing preliminary genetic tests in a 40 square meters laboratory. In 1994, aiming to provide better diagnostic services and genetic counseling for women and children, this small Department of Medical Genetics was expanded to a Prenatal Center for Genetic Disease Diagnosis and Treatment (PCGDDT). Continuous efforts in the past two decades have made the PCGDDT at GDWCH a major regional medical genetics center. As the public awareness of genetic diseases increased, there has been more and more governance support for prenatal, pediatric and reproductive genetic diagnosis in China. This report aims to present the spectrum of genetic services, the status of training programs and the scope of translational research projects developed in the PCGDDT. Our experience reflects a bottom-up model for the development of a hospital-based and institution-affiliated genetic services in China.

Key Words: bottom-up approach, genetic screening, prenatal diagnosis, biobanking

COMPREHENSIVE GENETIC SERVICES
To date, PCGDDT at GDWCH have equipped with a wide spectrum of technologies and occupied a space of approximately 3,600 square meters. The diagnostic laboratories are capable to perform fluorescent in situ hybridization (FISH), quantitative fluorescent polymerase chain reaction (QF-PCR), BACs-on-Beads (BoBs), multiplex ligation-dependent probe amplification (MLPA), array-based comparative genomic hybridization (aCGH), allele-specific PCR-based universal array, PCR-based Microsphere Hybridization Assay, DNA sequencing, next generation sequencing (NGS), flow cytometry (FCM), time-resolved fluorescence (TRF), enzyme-linked immunosorbent assay (ELISA), automatic chromosome scanning and karyotyping, etc. All these technologies have been validated and used in different genetic tests.

The CGDDT is composed of 76 clinical doctors and researchers, including nine with doctoral degree and 32 with master degree. Additionally, outstanding experts, including the Fellows of American College of Medical Genetics and Genomics and Diplomats of American Board of Medical Genetics from USA, are engaged as scientific advisers. The jointed efforts from internal staffs and external experts have kept us informed with the current advances and catch-up with the cutting edge technologies.

There are seven departments in the PCGDDT, including the Departments of Clinical Genetics, Molecular Genetics, Cytogenetics, Molecular Cytogenetics, Biochemical
The Department of Molecular Microbiology Genetics performed molecular microbiology genetic diagnosis of common infections in women and children, including UU, VT, NG, HSV, HPV, EV71 and CMV. Besides, the Department of Molecular Microbiology Genetics carry out the research of gut microbiota, host genetics and diet composition, for better nutritional consulting, diagnosis and therapy of overweight or obese subjects.

GENETIC TRAINING AND TRANSLATIONAL RESEARCH

The Department of Translational Medicine focuses on the course of predicting, preventing, diagnosing, and treating diseases. Translational Medicine also uses what can be gleaned in clinical studies to sharpen and improve what is done in preclinical efforts to discover new medicines. Translational medicine represents a paradigm shift in the biomedical research enterprise, and is highly valued by the Center and the Hospital.

The biobank has become a supportive resource for many types of research projects like genomics and personalized medicine. The biobank of Guangdong Women and Children Hospital contains more than 100,000 biological samples with related patient records. Researchers can access the samples and data for projects using large case series and disease cohort. Furthermore, samples in the biobank and the data derived from those samples can often be used by multiple researchers for multiple purposes.

SUMMARY

The PCGDDT has established cooperative relationship with over 130 women and children hospitals to provide comprehensive prenatal screening and genetic disease diagnosis. Every year, over 50,000 patients and 500,000 samples were transferred to our Center. Cooperative services are provided by every department in the Center to provide comprehensive screening and diagnosis of genetic diseases for women and children. Under comprehensive screening and diagnosis projects, we detect about 300 fetal chromosomal abnormalities and 400 moderate to severe thalassemia fetus each year, as well as other hereditary diseases. The experience of PCGDDT represents a bottom-up approach to build up sustainable hospital-based and institute-affiliated reproductive genetic services.

CONFLICT OF INTEREST

None.

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