Genomic medicine in Changhua Christian Hospital, Taiwan: innovation, research and service

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Introduction
Genomic medicine involves using genomic information as a part of patient care, such as genetic diagnosis and principles for therapeutic decision-making. For genetic diagnosis, however, it is sometime difficult to identify the genetic defect for a condition because it is possibly caused by a variety of genetic changes in a genome, ranging from a point mutation in a single gene to a gross chromosome abnormality involving the addition or deletion of an entire chromosome or set of chromosomes. To overcome this dilemma, innovative researches exploring easy, reliable methods for facilitating genetic diagnosis and translation of novel findings/techniques into routine clinical service are important, and these activities are conducting in our laboratory (Department of Genomic Medicine, Changhua Christian Hospital). we report and share our experiences in the rapidly developing field of genomic medicine.

Methodology and achievements

Figure 1. (A) A serial of cytogenetic, molecular cytogenetic and molecular technologies was established and applied for genetic testing. Examples include the establishments of (B) array-based comparative genomic hybridization (aCGH) for automatic detection of DNA copy number changes, (C) cell-free circulating fetal DNA analysis for noninvasive prenatal testing (NIPT) of fetal anomaly, and (D) preimplantation genetic diagnosis/screening (PGD/PGS) for identification of genetic defects within embryos prior to implantation.

Conclusions
The development of diagnostic techniques is helpful for the genetic defect detections, and the diagnostic results improve our understanding for mechanisms underlying diseases and facilitate personalized medicine by selecting the most appropriate treatment, according to the specific circumstances of the patient.

- Center for Medical Genetics: http://www2.cch.org.tw/CMG/index.aspx