EDITORIAL

Next-generation sequencing based molecular testing is an equalizer for diagnostic service of rare genetic disorders in China

The traditional approach of clinical genetic practice, known as the phenotype-first approach, usually starts with a thorough review of electronic medical records of the patient and a complete evaluation of patient's clinical presentation, as well as a survey of family history. Often more than one experienced clinical geneticists will order a variety of additional laboratory or imaging tests to obtain relevant information in order to reach a reasonable clinical diagnosis. Finally, molecular testing often helps to confirm the clinical diagnosis. This practice requires the availability of electronic medical records, well trained clinical geneticists who are able to perform relevant clinical evaluation, order appropriate tests and understand the molecular reports. These critical components of clinical genetics are mostly lacking in China today,[1] as a consequence, most of patients with genetic condition do not receive a proper evaluation by clinical geneticists, only a small percent of patients received a clinical diagnosis and the majority remains undiagnosed for life.

The advent of genome-wide tests for clinically relevant alterations, especially chromosomal microarray and exome tests, offered a new approach, which is genotype-first approach where a definitive diagnosis can be reached mainly based on molecular evidence. While electronic medical records, well trained clinical geneticists and genetic counselors are very important and desirable to have, we are appreciating more the value of the genotype-first approach that are currently helping the genetic diagnostic services of the tens of thousands of patients with undiagnosed genetic conditions.

The value of such a practice is exemplified in the paper authored by Hao et al[2] in this issue of Pediatric Investigation. Hao et al performed an exome analysis and successfully identified the causal variants for Antley-Bixler syndrome, an exceptionally rare genetic disorder with a feature of craniosynostosis in a Chinese child patient. While craniosynostosis is not a feature that could be easily missed, yet this is a feature known to be associated with a large number of clinically heterogeneous conditions with equally heterogeneous genetic basis. While a clinical diagnosis of this patient with this rare condition is possible by experienced clinical geneticist, the arrival of a definitive diagnosis may not be as quickly as it was demonstrated in this paper by an exome test. The fact that this patient had gone through different clinics (from Endocrinology to Otolaryngology to Orthopedics) before coming to Genetics demonstrated the current status of our ability for clinical evaluation and diagnosis for rare genetic disorders in China where genotype-first approach will continue to play an equalizing role for genetic diagnostics.

We all learn from practices, our clinicians will be able to arrive the right clinical diagnosis the next time when we encounter similar patients, thus the value of exome testing for this patient is more than getting the right diagnosis for this patient.

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CONFLICT OF INTEREST

I declare that I have no competing interests.

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Anhui Provincial Children's Hospital: On the fast track to development

The Anhui Provincial Children's Hospital (APCH) was established in 1982 and opened formally in 1990. Inspired by the hospital's motto of "Humanity, Devotion, Harmony, and Innovation", all the staff in APCH have made bold steps in making great achievements. As the only provincial-level tertiary pediatric hospital in Anhui Province, APCH has sophisticated medical equipment and complete disciplines that care for critically ill pediatric patients. It is a comprehensive base for clinical services, teaching, scientific research, disease prevention, health care, and rehabilitation. APCH currently is the vice-chairman unit of Anhui Children's Medical Association, director unit of Anhui Provincial Pediatric Professional Quality Control Center, etc.

The hospital covers an area of 50 000 square meters, and the floorage is 72 000 square meters. With 1 200 available beds, APCH has 28 wards, 11 medical technical departments, and 30 clinical subspecialties. It received over one million outpatients, discharged over 70 000 inpatients, and performed 16 000 operations in 2017. The hospital houses 1 815 highly trained staff, among whom 484 have an intermediate or higher professional title; 125 have senior title, and 32 are leaders of provincial or higher academic disciplines and academic groups. The hospital is fully equipped with a variety of modern medical and scientific research equipment including 1.5T nuclear magnetic resonance machine and 64-slice spiral CT.

The hospital has a wide range of disciplines. The internal medicine system includes the departments of neonatology, respiratory medicine, gastroenterology, nephrology, hematology, neurology, rheumatology, infection, and cardiology. The emergency center has departments of internal medicine, surgery, pre-hospital emergency transport service, emergency wards, and PICU. The surgical system is composed of departments of general surgery, cardiac surgery, thoracic surgery, neurosurgery, neonatal surgery, urology, orthopedics, burn and plastic surgery, and otorhinolaryngology.

With an attempt to assume the leading role of APCH in pediatrics in Anhui Province, the hospital initiated establishment of the first Children's Medical Association in China, which serves as a platform for scientific research, academic exchanges, and talent training for pediatricians in the province. Up to now, APCH has trained more than 8 500 pediatricians and other medical staff in primary maternal and child health care providers, which has rapidly improved pediatric care at the grass-roots level and made significant contributions to the decline of under-5 mortality rate in Anhui.

The hospital has successively won nearly 50 various honorary titles including "Survey on Residents' Satisfaction in the Top Ten Service Providers in Anhui Province", and APCH ranked first by the total score for four consecutive years (2014, 2015, 2016, and 2017) and won the "Golden Reputation Prize".

As a trusted center with a commitment to serving all the children in Anhui Province, APCH will keep up with the pace of advancements in science and technology and strive to improve the overall capacity and competitiveness of the hospital, so as to establish a first-rate children's hospital in China with complete disciplines and advanced diagnostic, therapeutic and preventive approaches.