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Plenary (October 7, 13:55-14:35)

Title: Rare Disease - Challenges in Diagnosis and Management

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Abstract:
To date molecular bases of 5,000 genetic diseases have been identified, while more than 3,000 genetic conditions still await to be elucidated. Since most of the genetic diseases are rare and present with obscure and/or unfamiliar combination of signs and symptoms, patients and their physicians often struggle to search for correct diagnosis for many years (i.e., a diagnostic odyssey). To this end we have initiated a national consortium, the Initiative on Rare and Undiagnosed Diseases in Pediatrics (IRUD-P) funded by the Japan Agency for Medical Research and Development (AMED). The aims of the project are to make diagnosis on patients with rare and undiagnosed diseases using next-generation sequencing, to construct their genome database with clinical information, to make banking system of precious specimens. The IRUD-P assigned 17 regional core clinical centers across Japan which evaluate clinical symptoms of patients who are referred from local hospitals/clinics and perform first-line laboratory examinations. The obtained data are carefully examined by experts specialized in rare diseases and selected patients are enrolled into IRUD-P. So far we have received DNA specimens from 1,542 patients and their parents. Our overall diagnostic rate was 33%. The diagnotic rate was as high as 45% when trio samples were available. Once the diagnosis is made, an appropriate treatment and management is provided to the patient. In some cases, molecular targeted therapies remarkably ameliorated patients' disease conditions. IRUD-P not only benefits patients with rare diseases, but also is expected to facilitate the discovery of important medical findings and the development of novel drugs for common diseases, as evidenced by the examples of PCSK9 inhibitors for hypercholesterolemia and SGLT2 inhibitors for diabetes.