Rare Diseases Column (Message from the Column Editor)

Challenges and opportunities in the studies on rare diseases

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According to the estimation from the Global Genes project, there are about 300 million people are worldwide affected by a rare disease (Orphan disease).

Most rare diseases are genetic based, presented in early life, with chronic phase but frequently progressive, disabling and life threatening. About 30% of affected children will die at 5 years old.

Although there are some definitions about the rare diseases based on the number of people living with a disease including several factors such as the existence of adequate treatments or the severity of the disease, unfortunately, there is no single, widely accepted definition for rare diseases up to now. Evidences and facts from the accumulated indicate that rare diseases are not uncommon. Actually, the most accepted definition is no disease is rare when it affects someone you love.

Nowadays, the treatment of infectious and nutritional disorders is well established, pediatric onset genetic disorders and cancer in children constitute a substantial load in pediatric clinic and rare disease is the major one in it. The care to rare diseases have become to unprecedented internationally. To raise awareness of rare disease and their impacting on patient's lives, the last day of the February each year is recognized as the Rare Disease Day.

The concepts and practices on rare diseases have been renewing with the recent advancing studies. The study on rare diseases is the hallmark of genetic era in medicine from clinical symptomatic to pathological etiology phase. The applications of next generation sequencing technology have been bringing the benefits to patients with rare diseases first. About 50% of the estimated 7,000 rare disease responsible genes have been identified for the determination of their molecular etiology and it is predicted that the rest of 50% genes will be identified by 2020 and it will be speeded up by the next sequencing technology.

In addition, knowledge and experience obtained from the studies on rare diseases increased our understanding of the correlations between phenotype and genotype, enlightening the other areas particularly in personalized medicine and translating into the development in therapies. The most representative studies on rare diseases is the Fanconi anemia, an inherited rare disease which it is characterized with developmental abnormalities, progressive bone marrow failure and cancer predisposition. Studies found that the incidence of Fanconi anemia is 1 in 160,000 in the general population but it is 1 in 20,000 in some ethnic groups. It was the first successful example in cord blood stem cell transplantation (in 1988). So far, 16 responsible genes were identified and interestingly, 4 of them are genes found in breast cancer and some rest genes were found associated with other types of cancers as well. So the study on Fanconi anemia is named as a paradigm for the understanding of cancer and aging.

To meet more needs, this Journal launched a special column on rare disease studies from this edition and Dr. Kaustuv Bhattacharya from University of Sydney contributed his typical and classical rare disease article titled "Investigation and management of the hepatic glycogen storage diseases" with the discussion from pathological theories to clinical practices of the disease.

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Footnote

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