Rare Diseases Column (Commentary)

Chromosomal instability syndromes—the most classical and representative types of rare diseases

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As defined, rare diseases including rare childhood cancers are any disorder or condition that is life-threatening or chronically debilitating and is statistically rare with an estimated prevalence of 5 in 10,000 people and the rare diseases are about 80% genetic based in their pathological cause. According to the announcement from national organisation for rare diseases (NORD), 1 in 10 Americans live with a rare disease and they are at high level of symptom complexity so they need special diagnostic and therapeutic strategies.

Many types of rare genetic diseases associate with chromosome instability so that they are named as chromosome instability syndromes and they share many clinical features and associated with an increased risk of cancer. These diseases are characterized with a high level of spontaneous chromatid/chromosome breakage, chromosome rearrangements and/or a hypersensitivity to clastogens.

Rare diseases have been becoming the major task in pediatric and also increased interestings practices worldwide especially in the molecular level with the molecular advances and on the improvement in methods for genomic diagnosis of rare disease, analysis of large genomic data genetic variation to validate drug targets.

Fanconi anemia, Ataxia telangiectasia, Ataxia telangiectasia-like disorder, Bloom syndrome and Nijmegen breakage syndromes are the typical rare diseases characterised with complex clinical presentations, chromatid/chromosome fragility, defects of DNA repair and cancer predisposition. Inherited cancer can be the early presentation of the disease as a reason to clinic.

Due to the diseases nature, diagnosis/differential diagnosis on rare diseases can be difficulty especially under the condition in lack of the specific techniques. Delay, misdiagnosis and even mistreatment were not uncommon under the condition to treat such syndromes with the normal dosage for radiation/chemotherapy.

Cooperation of Multi-disciplines including genetics, hematology, endocrinology, immunology, microbiology, oncology and surgical is required as a team in the diagnosis and treatment. In the laboratory testing, from cellular, protein and molecular levels and it is generally performed in a qualified and experienced laboratory.

To deal with such challenges in the need of pediatric practice, we published an article titled "Phenotypes and Genotypes of Chromosomal Instability Syndromes" (1) including the pathological causes, molecular mechanisms, clinical presentations, cancer predisposition laboratory diagnosis in their similarities and differences to help to make a correct and timing diagnosis for the early therapies in this rare disease column of this edition in this group of rare diseases including Fanconi anemia, Nijmegen breakage syndrome, Bloom syndrome, Ataxia telangiectasia and Ataxia telangiectasia-like disorder.

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Footnote

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References

1. Wu ZH. Phenotypes and genotypes of the chromosomal instability syndromes. Transl Pediatr 2016;5:79-83.

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