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EDITORIAL

# Editor's Introduction to This Issue

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Next generation sequencing (NGS) has been facilitating the discovery and functional validation of causal variants in diverse rare diseases. However, for most of the 7,000 rare diseases, pathogenic mechanisms are still unknown. In this issue, Dr. Murim Choi's group (Seoul National University, Korea) comprehensively reviewed ultra-rare diseases and genomics-driven precision medicine. They presented the perspectives gained from recent studies on ultra-rare diseases which were diagnosed by NGS analysis and successfully treated. They also discussed the potential obstacles to wider application of NGS-based precision medicine. Dr. Young-Joon Kim's group (Yonsei University, Korea) reported classification of colorectal cancer patients based on promoter methylation patterns. They used colon cancer data sets from the Cancer Genome Atlas to observe CpG islands that showed significant aberrations in DNA methylation and also analyzed changes in DNA methylation patterns to assess the relationships between epigenetic changes and tumorigenesis. Dr. Jong Il Kim's group (Seoul National University, Korea) reported the genome-wide association study which evaluated the effects of genetic polymorphisms on the bone mineral density at the lumbar spine and femur in Korean men. Investigating the etiology of osteoporosis in males of East Asian descent can have high diagnostic and clinical implications. Dr. Thirumudi's group (Kamalnayan Bajaj Institute for Research in Vision and Ophthalmology, India) reported two potential inhibitors targeting the hydrophobic cleft of Toxoplasma gondii apical membrane antigen 1 (AMA1).

For further details, please visit G&I homepage (http://www.kogo.or.kr/webapp/kogo_publish/genomics_and_informatics/).

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