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Editor's Introduction to This Issue (G&I 16:2, 2018)

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This issue covers two original articles on mutation analysis and one original article on the identification of a rare disease gene. In the article by Dr. Dong-Uk Kim and Dr. Kwang-Lae Hoe, the authors report the frequency and type of mutations in synthetic DNA barcodes that are incorporated into the genome of fission yeast. They had previously reported the construction of a gene deletion library of fission yeast. Mutations in DNA barcodes might cause serious problems in microarray experiments. The comprehensive characterization of mutations by this study will guide the proper application of the library. Dr. Yeun-Jun Chung's group compared mutations in primary colorectal tumors and the corresponding xenograft tumors. The patient-derive xenograft (PDX) has been recognized as an important tool for finding the most appropriate personalized treatment for each cancer patient. Its biggest issue is how often the mutation in the cancer tissue disappears or how frequently new mutations occur during the process of PDX generation.

In this paper, the authors show that most somatic mutations and copy number alterations are maintained in the PDX. These results are important data that support the implementation of PDX in precision medicine. Dr. Jong-Keuk Lee's group and the Korean Kawasaki Disease Genetics Consortium show that Kawasaki disease in children aged younger than 6 months, which is not a common age of onset, is significantly associated with a rare nonsynonymous single-nucleotide polymorphism in the lymphoid enhancerbinding factor 1 (LEF1) gene in a genome-wide association study and a follow-up replication study. It is interesting that similar patterns, although not statistically significant, were observed in the Japanese patient group. This study may provide new insights into the pathogenesis of Kawasaki disease if further association studies using more patient samples and gene function analysis are performed.

For further details, please visit the G&I homepage (https://genominfo.org/).

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