Editorial

Single nucleotide polymorphisms (SNPs) may introduce premature termination codons. But do they really exist? In this issue of *Human Genomics*, Savas *et al*. publish their systematic search for such SNPs (known as X-SNPs) in the dbSNP database. Surprisingly, they found 28 of them, the majority of them representing commonly occurring SNPs. Furthermore, the population specificity of these X-SNPs is clearly notable. Are they related to any diseases or traits? Not yet, but soon they will be. Also from Ozcelik's group, Savas *et al*. report the results of a search of functional non-synonymous SNPs (nsSNPs) in a large number of carcinogenesis-related genes expressed in breast tissue. They suggest that these nsSNPs are likely to contribute to breast cancer susceptibility.

While association analysis provides clues for the location of the genes/mutations underlying diseases, functional assays are critical to nail them. Interrogation of the impact of a missense variation on the tertiary structure of a protein may shed light on the possible functional role of the mutation of interest. Shabbeer *et al*. did exactly that on 29 missense mutations underlying Fabry disease, as reported in this issue.

As a special feature of this Journal, we continue to publish reviews on analytical methods. In this issue, three such reviews are published. Dudbridge *et al*. conducted an in-depth review of the detection of multiple associations in genome-wide studies. Motsinger and Ritchie provide an introduction to multifactor dimensionality reduction for detecting and modelling gene–gene interactions. Onkamo and Toivonen contribute a comprehensive survey on data mining methods for linkage disequilibrium mapping.

In this issue, *Human Genomics* publishes its first ever Letter to the Editor since it was inaugurated two years ago. I am pleased to see that this Journal is becoming an attractive forum for constructive academic discussions. More will come in the next issue.

Li Jin
Managing Editor
Human Genomics