MOBILE ELEMENTS AND THE HUMAN GENOME

In recent years, data on human genome variability for SNPs and copy number variations have accumulated. Data on variations due to Mobile Elements Insertion (MEI) lagged slightly behind. An article in press in Genome Research¹ reports improved software to unveil MEIs (CloudMELT). The authors used this new tool to study 57,919 human genomes. They found 104,350 insertions and analyzed their mutagenic impact, such as disruption of genes, but also the potential consequences on gene expression. They also identified the active L1 elements that drive MEI mutagenesis.

Some insertions are very rare and probably arose relatively recently.

¹ https://genome.cshlp.org/content/31/12/2225.long