GENOMIC MOSAICISM AND DISEASE

Mosacism for aneuploides and DNA variants is quite common in early embryogenesis. Some variants are incompatible with cellular growth and are counterselected. Other variants can persist and can be detected in adult individuals, using deep sequencing. Depending on the extension of mosaicism and the tissue affected, these variants can contribute to an abnormal phenotype.

Truty et al. (2023)[1] analyzed 1 million individuals referred for genetic testing and searched for mosaicism of 1,900 disease-related genes. They found “5,939 mosaic sequence or intragenic copy number variants distributed across 509 genes in nearly 5,700 individuals, constituting approximately 2% of molecular diagnoses in the cohort”.

This large study expands our understanding of the phenotypic implications of mosaicism.

1-https://www.cell.com/ajhg/fulltext/S0002-9297(23)00083-6