STRUCTURAL VARIATIONS (SVs) AND GENE EXPRESSION

In 2012 a Science comment[1] on the results of the ENCODE project was paradigmatic. It reads "ENCODE Project Writes Eulogy For Junk DNA ". The project had found that 80% of the human genome has a purpose. The term "junk DNA" was no longer used. Since then, more articles have highlighted the importance of DNA once considered junk, such as the contribution of transposable elements to gene regulation (for example, Diehl et al. 2020[2], in Nature Communications).

An article in press in Genome Research by Scott et al.[3] has analyzed the impact on gene expression of 61,668 Structural Variations in 613 individuals. They found that duplications and deletions were the variant types with the most impact, while the contribution of transposable element insertions was small. They also found that the effect often extends to more than one gene and, occasionally, it can reach genes up to 1 Mb apart.

[1] https://www.science.org/doi/10.1126/science.337.6099.1159?url_ver=Z39.88-2003&rfr_id=ori:rid:crossref.org&rfr_dat=cr_pub%20%200pubmed

[2] https://www.nature.com/articles/s41467-020-15520-5

[3] https://genome.cshlp.org/content/early/2021/09/20/gr.275488.121.long