PATHOGENICITY PREDICTION OF STRUCTURAL VARIANTS

Predicting the pathogenicity of structural variants (SV) is not an easy task, especially if they have been detected *de novo*. Several databases and software are available to help clinicians and researchers in this task. A paper in Am. J, Hum. Genet.[1] proposes a new supervised learning method, StrVCTVRE (available free at https://github.com/andrewSharo/StrVCTVRE). The authors state that this tool allows clinicians to eliminate about half of the SVs from consideration while maintaining a sensitivity of 90%. The improvement is mainly due to the inclusion of information about expression and evolutionary conservation among the analyzed parameters of the gene in question.

1 https://www.cell.com/ajhg/fulltext/S0002-9297(21)00462-6