LONG-READ SEQUENCING AND MISSING DISEASE-CAUSING VARIATION

Short-read sequencing in cases of suspected genetic disease has proved to be crucial in about half of the cases studied. This approach, however, is not very efficient in detecting pathogenic structural variants such as repeated expansions, insertions, deletions or rearrangements. Long-read sequencing is much more efficient in this respect, but whole genome sequencing is currently too expensive. The authors of an article that appeared in Am J Hum Genet (Miller et al, 2021) found a way to use one of these technologies (Oxford Nanopore Technologies) for appropriate targeted sequencing (up to 151 Mb in this case). The results suggest that this approach can be used efficiently in cases where whole genome sequencing based on short reads has failed to reveal the etiology of the clinical condition.