TARGETED LONG-READ SEQUENCING AND GENETIC DISEASES

Short-read sequencing technology may occasionally fail to resolve suspected genetic diseases. The authors of a paper which appeared in Am J Hum Genet* employed targeted (151 Mb) long-read sequencing (T-LRS) in 40 of these cases using the Oxford Nanopore platform. In 8/8 individuals with complex structural rearrangements, T-LRS allowed more precise mutation resolution, leading to changes in clinical management in one case. In ten individuals with suspected Mendelian conditions, T-LRS identified pathogenic or probably pathogenic variants in six, and variants of uncertain significance in two others.

* https://www.sciencedirect.com/science/article/abs/pii/S0002929721002305?via%3Dihub