

TANDEM REPEATS, LONG-READ SEQUENCING AND NEUROLOGICAL DISEASES

Various neurological diseases, as well as other conditions, are caused by increase in the number of tandem repeat (TR) copies (Huntington's chorea, myotonic dystrophy, fragile X syndrome...). Characterizing these expansions using short-read sequencing is challenging, as this method struggles with repetitive regions. Long-read sequencing technologies, however, are far more suitable for this task. To enhance the utility of these technologies, Zhang et al. (1) developed an algorithm called **MotifScope** to identify and characterize TRs more accurately.

1. <https://genome.cshlp.org/content/early/2024/11/13/gr.279278.124>