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User-friendly automated analysis for diagnosis of repeat expansion disorders using targeted nanopore sequencing

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Standard diagnostic methods for tandem repeat expansion disorders, such as Southern blotting and modified PCR, cannot identify the size of long repeats accurately. We developed an efficient and accurate diagnostic platform using Cas9-targeted nanopore sequencing and RepeatLab, a new automated analysis pipeline. This program automatically estimates repeat length, repeat structure, and methylation profiles from sequencing data. Our platform's repeat expansion status was tested in thirteen myotonic dystrophy type 1 patients, four healthy individuals, and four cell line samples to determine its feasibility. Our method can diagnose repeat expansion diseases in less than a day for under $200 without computational skills or resources.