Oxford Nanopore Technologies

Oxford Nanopore rapid whole-genome sequencing for paediatric rare disease research

Accelerating insights into rare and undiagnosed disorders

The implementation of rapid whole-genome sequencing in clinical research settings is enabling faster-than-ever sample-to-answer results in time-critical conditions. Achieve a turnaround time in as little as 24 hours with Oxford Nanopore rapid whole-genome sequencing (WGS), complete with 30 kb read lengths, to analyse SNVs, indels, SVs, repeat expansions, CNVs, and allele-specific methylation.

The Oxford Nanopore rapid whole-genome sequencing workflow delivers:

- A decentralised, flexible solution for rapid WGS
- Actionable results in 24 hours from sample collection
- Rapid analysis of SNVs, indels, SVs, CNVs, and methylation
- Comprehensive data fully compatible with tertiary analysis software for variant prioritisation and interpretation
- Simplified haplotype phasing to enable unambiguous characterisation of compound heterozygosity

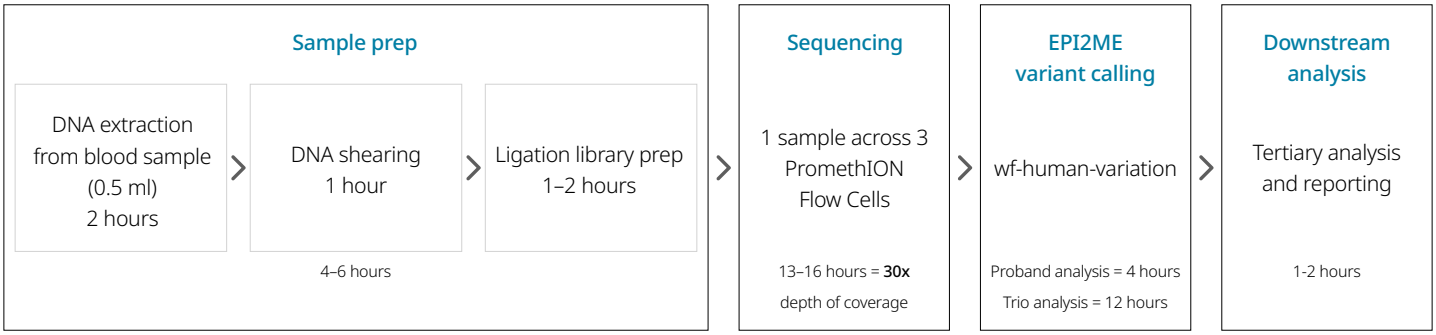
WGS with short-read sequencing	Rapid WGS with Oxford Nanopore sequencing
Turnaround time is typically ≥36 hours	Turnaround time is 24 hours
Cost penalty for running a single sample	Run 1–8 samples simultaneously, with no cost penalty for single samples
8% of the genome is challenging to sequence with short reads, including repetitive and complex areas, and some coding regions[1]	Recent studies show a 17–24% increase in diagnostic yield with long reads[2,3]
Does not include methylation data	Methylation data is included, enabling characterisation of imprinting disorders, some repeat expansion disorders, and uniparental disomy

Clinically used genetic methods leave gaps; reveal everything with Oxford Nanopore sequencing

	Oxford Nanopore	Sanger	PCR	Chromosomal microarray	Targeted methods	Short-read WGS
SNVs	✓	✓	✓		✓	✓
Indels	✓	✓	✓	✓	✓	✓
Complex SVs	✓			✓		
CNVs	✓		✓	✓	✓	✓
Genes/pseudogenes	✓					
Repeat expansions	✓		✓			
Mosaicisms	✓					
Mitochondrial variants	✓	✓				✓
Methylation haplotypes	✓					

Looking across all variant classes

The Oxford Nanopore rapid WGS workflow provides a fast and flexible method for the investigation of all variant classes to resolve challenging cases on a single platform.



Flexible, sample-to-prioritised-variants workflow

Prepare: extract DNA from human blood, then prepare libraries using the Ligation Sequencing Kit to retain epigenetic modifications and generate high output.

Sequence: process sample across three PromethION™ Flow Cells simultaneously to achieve >30x coverage (>100 Gb) in as little as 13 hours.

Analyse: after live basecalling, use the EPI2ME™ wf-human-variation workflow for all-in-one calling of SNVs, SVs, CNVs, STR expansions, and methylation — without the need for prior bioinformatics experience. The EPI2ME output is compatible with tertiary analysis partner solutions for variant prioritisation and interpretation.

Find out more about clinical research with Oxford Nanopore:
nanoporetech.com/clinical-research

References:

1. Wagner, J. et al. Curated variation benchmarks for challenging medically relevant autosomal genes. *Nat. Biotechnol.* 40(5):672–680 (2022). DOI: https://doi.org/10.1038/s41587-021-01158-1
2. Del Gobbo, G.F. and Boycott, K.M. The additional diagnostic yield of long-read sequencing in undiagnosed rare diseases. *Genome Res.* 35(4):559–571 (2025). DOI: https://doi.org/10.1101/gr.279970.124
3. Oxford Nanopore Technologies. Diagnosing the undiagnosed with long-read genome data. https://nanoporetech.com/resource-centre/diagnosing-the-undiagnosed-with-long-read-genome-data (2025) [Accessed 23 June 2025]

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