

Review

Enhancing Molecular Testing for Effective Delivery of Actionable Gene Diagnostics

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Table S1. Relevant data on human cell line samples processed by ONT.

Region of interest	Sample	DNA extraction	Library Prep	Sequencing platform	Results	Validation	Reference
<i>ABCB1</i>	THP-1 cell line	QIAamp DNA mini kit	LSK108 or LSK109	MinION	Structural variants, <i>ABCB1</i> translocation detected by nanopore sequencing	Chip seq, NGS	[62]
Chromosome 1	Lymphoblastoid cell line	Chromosome preparation	LSK-109	MinION	Highly continuous and selective assembly of chromosome 1	NA	[63]
<i>Gene fusion BCR-ABL1 PAX5-AUTS2 HAS2-PLAG1</i>	K562 cell line	RNA - RNeasy system	LSK-108	MinION	Fusions were detected	NGS - Illumina	[39]
Short tandem repeats	HiPSC line	HMW DNA preparation with a modified phenol-chlorophorm method	LSK-108, 109	MinION, PromethION	STR expansions	Southern-blot	[36]
NMD-targeted isoforms	HeLa cell line	Total RNA: GenElute Mammalian Total RNA Miniprep Kit	DCS-108	GridION	identification of many novel NMD-sensitive mRNAs	NGS - Illumina	[64]
Evaluate CpG methylation	GM12878 lymphoblast cell line	Phenol-chlorophorm	LSK-108	MinION	NS allows to generate human epigenome	Bisulfite sequencing	[65]
<i>TP53, BRAF, KRAS</i>	MDA-MB-231, MCF-7, MCF-10A, GM12878	Circulomics CBB kit	LSK-109	MinION	SNV, methylation	whole-genome bisulfite sequencing	[14]
<i>whole genome</i>	GM12878	QIamp DNA Mini kit (Qiagen)	LSK-108, RAD002	MinION	assembly of human genome 99,88%	Non-validated	[66]

NA—no available data, NGS—next generation sequencing.

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