

# Hereditary Cancer Panel

Targeted sequencing via adaptive sampling

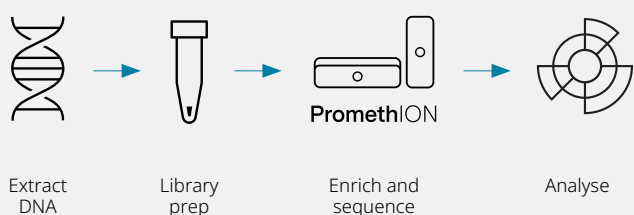
## Comprehensive characterisation of hereditary cancer targets with Oxford Nanopore sequencing

The **Oxford Nanopore Hereditary Cancer Panel** is a comprehensive sequencing assay targeting 258 full-length genes — covering exons, introns, and promoters — linked to inherited cancer risk. The panel uses **adaptive sampling**: a fast and flexible on-sequencer target enrichment methodology. No lengthy library preparation, baits, or primers required.

Leveraging Oxford Nanopore reads of unrestricted length — from short to long — the Hereditary Cancer Panel can accurately resolve complex structural variants, single nucleotide variants, and insertions/deletions. It enables haplotype phasing and can resolve pseudogenes. Additionally, direct DNA sequencing captures methylation patterns, offering a multiomic view that enables the discovery of key biological insights such as gene silencing and parent-of-origin effects.

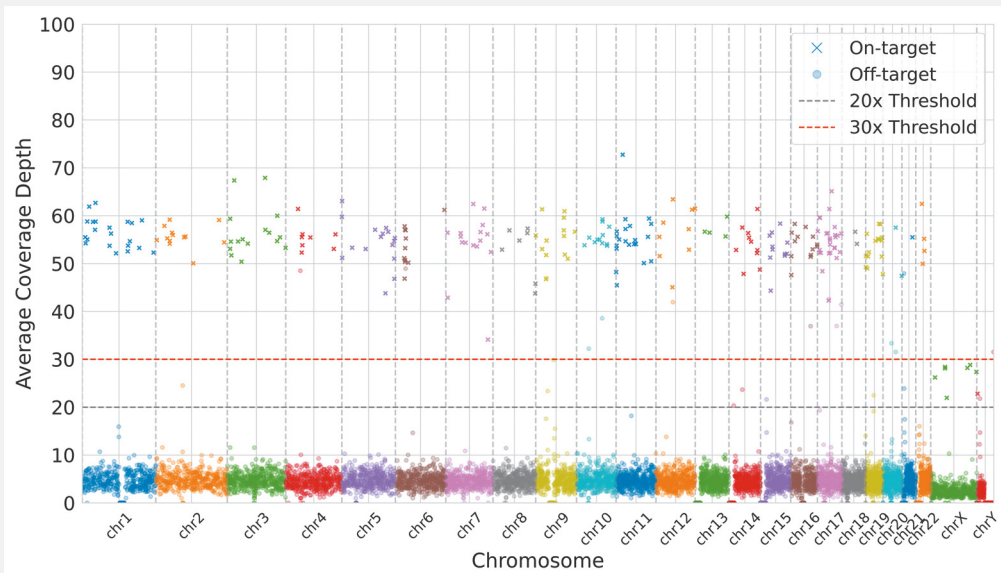
### Hereditary Cancer Panel gene list

|          |         |          |          |          |          |        |        |        |       |        |         |         |          |         |
|----------|---------|----------|----------|----------|----------|--------|--------|--------|-------|--------|---------|---------|----------|---------|
| ABRAXAS1 | ACD     | AIP      | AKT1     | ALK      | ANKRD26  | APC    | ARID5B | ATG2B  | ATM   | ATR    | AXIN2   | BAP1    | BARD1    | BLM     |
| BMPR1A   | BRAF    | BRCA1    | BRCA2    | BRIP1    | BUB1B    | CBL    | CDC73  | CDH1   | CDK12 | CDK4   | CDKN1B  | CDKN1C  | CDKN2A   | CEBPA   |
| CFTR     | CHEK2   | COL7A1   | CSF3R    | CTC1     | CTNNA1   | CTR9   | CYLD   | DDB2   | DDX41 | DICER1 | DIS3L2  | DKC1    | DNAH9    | DNAJC21 |
| DOCK8    | DPYD    | EFL1     | EGFR     | ELANE    | ELP1     | EPCAM  | ERCC1  | ERCC2  | ERCC3 | ERCC4  | ERCC5   | ERCC6L2 | ETV6     | EXT1    |
| EXT2     | FAAP100 | FAN1     | FANCA    | FANCB    | FANCC    | FANCD2 | FANCE  | FANCF  | FANCG | FANCI  | FANCL   | FANCM   | FAS      | FH      |
| FLCN     | G6PC3   | GALNT12  | GATA1    | GATA2    | GBA1     | GFI1   | GPC3   | GPR161 | GRB10 | GREM1  | GSKIP   | H19     | HAX1     | HLA-A   |
| HLA-B    | HLA-C   | HLA-DPA1 | HLA-DQA1 | HLA-DQB1 | HLA-DRB1 | HMBS   | HOXB13 | HRAS   | IGF2  | IKZF1  | ITK     | KCNQ1   | KCNQ1OT1 | KIF1B   |
| KIT      | KLLN    | KRAS     | L3MBTL1  | LIG4     | LZTR1    | MAD2L2 | MAP2K1 | MAP2K2 | MAX   | MBD4   | MC1R    | MECOM   | MEN1     | MEST    |
| MET      | MITF    | MLH1     | MLH3     | MPL      | MRE11    | MSH2   | MSH3   | MSH6   | MTAP  | MUTYH  | NAF1    | NAPRT   | NBN      | NF1     |
| NF2      | NHP2    | NLRP2    | NLRP5    | NLRP7    | NOP10    | NPM1   | NRAS   | NSD1   | NTHL1 | PADI6  | PALB2   | PALLD   | PARN     | PAX5    |
| PAX6     | PDGFRA  | PDGFRB   | PHOX2B   | PIK3CA   | PMS2     | PMS2CL | POLD1  | POLE   | POLH  | POT1   | PPM1D   | PRF1    | PRKAR1A  | PRSS1   |
| PTCH1    | PTEN    | PTPN11   | RAD21    | RAD50    | RAD51    | RAD51B | RAD51C | RAD51D | RAF1  | RB1    | RECQL4  | REST    | RET      | RFXD3   |
| RHBF2    | RINT1   | RMRP     | RNF43    | RPA1     | RPL11    | RPL15  | RPL23  | RPL26  | RPL27 | RPL31  | RPL35A  | RPL36   | RPL5     | RPS10   |
| RPS17    | RPS19   | RPS20    | RPS24    | RPS26    | RPS27    | RPS27A | RPS28  | RPS29  | RPS7  | RTEL1  | RUNX1   | SAMD9   | SAMD9L   | SBDS    |
| SDHA     | SDHAF2  | SDHB     | SDHC     | SDHD     | SETBP1   | SH2B3  | SH2D1A | SHOC2  | SLX4  | SMAD4  | SMARCA4 | SMARCB1 | SMARCE1  | SOS1    |
| SPINK1   | SPRED1  | SQSTM1   | SRP72    | SRY      | STAT3    | STK11  | STN1   | SUFU   | TBXT  | TERC   | TERF2IP | TERT    | TINF2    | TMEM127 |
| TP53     | TRIM28  | TRIM37   | TRIP13   | TSC1     | TSC2     | TSR2   | UBE2T  | UROD   | VHL   | WAS    | WRAP53  | WRN     | WT1      | XPA     |
| XPC      | XRCC2   | ZCCHC8   |          |          |          |        |        |        |       |        |         |         |          |         |



### Panel specifications

|                                   |                    |
|-----------------------------------|--------------------|
| Enrichment method                 | Adaptive sampling  |
| Genes                             | 258                |
| Reference genome                  | hg38               |
| Recommended DNA input             | 1 µg               |
| Sample type                       | Whole blood, DNA   |
| Coverage uniformity (fold-80)     | 1.2                |
| Samples per PromethION™ Flow Cell | 3                  |
| Total assay time (hands-on time)  | <5 days (<2 hours) |



**Figure 1.** Sequencing coverage across the genome, depicting low-pass whole-genome coverage and >30x enrichment for target loci.



**Figure 2.** Representative large >1 kb deletion with long reads spanning the full deletion in *POLH*, a gene mutated in the hereditary disorder xeroderma pigmentosum, along with hypomethylation of the promoter region.



Find out more: [nanoporetech.com/hereditary-cancer-panel](https://nanoporetech.com/hereditary-cancer-panel)



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