COMPREHENSIVE GENOMIC PROFILING (CGP)– Test Information Sheet

Indications for ordering

This test detects genomic abnormalities with a diagnostic, predictive or prognostic role for patients with the following tumour types (requests for any other type of tumour will not be accepted):

- Cholangiocarcinoma (CCA)
- High-grade serous ovarian cancer (HGOSC)
- Metastatic castration-resistant prostate cancer (mCRPC)

For CCA, only cases for which a prior tumour analysis of the *FGFR* gene has not been performed are eligible.

For HGOSC and mCRPC, only cases with no known *BRCA* gene alteration are eligible. For this test to be performed, the results of any prior *BRCA* testing must be provided with/on the requisition.

Test Methodology

The comprehensive genomic profiling targeted test is a targeted next generation sequencing assay for analysis of 523 genes with known relevance to solid tumours. The panel enables the simultaneous analysis of genes associated with several common cancers. The DNA and RNA analysis can detect multiple alterations including: (i) single nucleotide variants (SNVs), (ii) small insertion/deletions (indels), (iii) copy number gains/losses or amplifications (iv) gene fusions in RNA samples and some tumour signatures. See below for the list of actionable genes reported. For more information on the hotspot regions tested, please contact the laboratory.

Tumour Signatures reported

- **Tumour Mutation Burden (TMB)**
- **Microsatellite Instability (MSI)**
- **Homologous Recombinant Deficiency (HRD) (only reported for HGOSC)**

List of reported genes

Actionable genes : SNVs/Small indels

AKT1	*CDKN2A*	*FGFR2*	*MRE11A*	*PIK3CA*
AR	*CDKN2B*	*FGFR3*	*MSH2*	*PIK3R1*
ATM	*CHEK2*	*IDH1*	*MSH6*	*PTEN*
ATR	*EGFR*	*IDH2*	*MTOR*	*RAD51C*
BRAF	*ERBB2*	*KIT*	*NBN*	*RET*
BRCA1	*ESR1*	*KRAS*	*NRAS*	
BRCA2	*FANCA*	*MET*	*PALB2*	
CDK12	*FGFR1*	*MLH1*	*PDGFRA*	

Actionable genes: CNVs

ATM	*CCNE1*	*CHEK2*	*MLH1*	*NBN*
ATR	*CDK12*	*ERBB2*	*MRE11A*	*PALB2*
BRCA1	*CDKN2A*	*FANCA*	*MSH2*	*PTEN*
BRCA2	*CDKN2B*	*MET*	*MSH6*	*RAD51C*

Actionable genes: Fusions (RNA)

ALK	*FGFR2*	*MET*	*NTRK2*	*RET*
FGFR1	*FGFR3*	*NTRK1*	*NTRK3*	*ROS1*

Test Interpretation

Only variants with a recognized diagnostic, predictive or prognostic clinical significance are interpreted and reported (Tier I and II; PMID: 27993330). In specific circumstances, additional results could be made available upon request.

Specimens accepted

- Tumour cell content (TCC) ≥ 20% is required. CNV detection cannot be carried out if the tumour cellularity is <40%. This information is mandatory to assess the validity of the provided sample.
- Formalin-fixed paraffin-embedded (FFPE) specimens (cytology or histology), frozen or fresh tissues, and DNA are accepted. Please see the requisition and the information below for more details.
- FFPE specimen: only scrolls or unstained slides are accepted. Blocks and specimens decalcified with formic acid will not be accepted.
- FFPE unstained slides: a minimum of 10 sections of 10 µm thickness non-baked and uncharged (tumour area should be outlined with permanent marker if macrodissection is needed).
- One H&E stained slide must be provided with every request.
- **For HGOSC, primary surgery samples or diagnostic biopsy samples are recommended. Debulked samples are not accepted.**
- For shipping instructions, please refer to the requisition.

Important Information

Comprehensive genomic profiling of the tumor may identify variants suggestive of a hereditary cancer predisposition. Depending on the result, post-test genetic counselling could be recommended.

The ordering physician is responsible to ensure referral for germline confirmation and genetic counselling when a potential hereditary variant is identified.

This test is not intended to replace comprehensive germline genetic testing or genetic counselling and should not be used as the sole basis for clinical decision-making related to inherited cancer risk.

The laboratory assumes no responsibility for downstream clinical actions or omissions by the clinical team related to result interpretation, patient disclosure, or follow-up.

Limitations

Results must be interpreted in the context of clinical, radiological and histological findings. If the results obtained do not match other clinical or laboratory findings, or if you have novel relevant information, please contact the laboratory as soon as possible for an updated interpretation.

Results may be compromised if the recommended procedures (tissue fixation and preparation) have not been followed.

A negative result does not rule out the presence of an alteration and may be due to the limitations of this assay (i.e. insufficient % of tumour cell content or poor fixation).

Rare polymorphisms may be present that could lead to false-negative or false-positive results.

Turnaround time: 15 working days

Revised and approved by Dr. Andrea Gomez 2026-07-16