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Oncogenetics - Hereditary cancer predispositions

The interest of exome sequencing

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5 to 10% of cancers are linked to the presence of constitutional genetic mutations which can be transmitted to descendants.

Certain clinical factors are particularly suggestive of a predisposition to an increased risk of cancer:

- family history
- multiple cancers in one patient
- onset of cancer at a young age
- rare cancers
- somatic analysis giving rise to suspicion of constitutional mutation
- knowledge of a common predisposition

To date, more than 80 genetic predisposition genes to cancer have been identified.

The identification of the causal mutation for the predisposition to cancer is essential:

- **For the patient:** because makes it possible to confirm a diagnosis, make a prognosis, guide the management of treatment and/or to set up a personalised follow-up.
- **And for relatives:** so they can be offered genetic counselling and specific monitoring can be arranged.

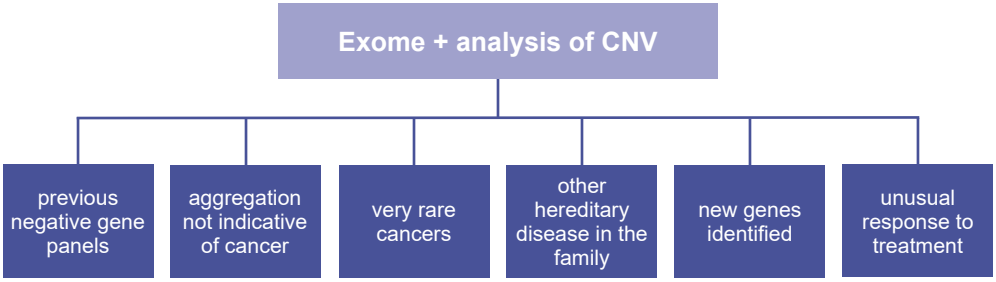
In a certain number of cases, the causes of hereditary cancers are not linked to known and validated predisposing genes included in the panels. **Exome sequencing makes it possible to analyse or identify new genes of predisposition.**

Exome sequencing in oncogenetics

Exome sequencing is an effective tool for studying diseases of genetic origin. Although exonic regions account for less than 2% of the genome, they can contain up to 85% of identified pathogenic variants.

Due to its exhaustive nature, exome sequencing particularly enables **the analysis of all known genes with validated implications in genetic predisposition to cancers** (in particular genes predisposing to cancers of the breast, ovaries, digestive tract, pancreas, skin or kidney) but also of **candidate genes** or even the identification of **new genes.**

Sequencing has therefore been integrated into the oncogenetics care pathway and specifically for the following indications:



This strategy offers the possibility of **retrospectively re-analysing the data** without the need for “resequencing”. The advancement of knowledge (new genes of interest or pathogenicity of variants) and the updating of databases will allow existing data to be re-interpreted.

Benefits of exome sequencing

- cost and time saving compared to working through a series of gene panels,
- provides a diagnostic test for patients whose clinical examination did not enable the clinician to identify the gene or panel of genes to be tested,
- retrophenotyping in atypical clinical pictures,
- generation of data that can be used in other clinical contexts,
- the establishment of a global database for interpretation, with all indications combined.

Oncogenetics by Eurofins Biomnis

Backed by multi-disciplinary expertise (clinical, genetic, scientific and bioinformatic), Eurofins Biomnis offers, post-sequencing the genetic material of the index patient (the proband/"solo") and potentially affected relatives ("duo"/"trio"):

- Provision of raw data (fastQ, VCF, BAM and quality report) via a secure interface (within 4 weeks),
- An interpretation based on the filtering and analysis of more than ten thousand variants in consultation with the clinician,
- Issue of report with detailed results within an optimised TAT of **4 weeks**.

Technique

All of the coding regions of the genes are analysed by next-generation sequencing on an Illumina NovaSeq sequencer. The data from the sequencing is then analysed and interpreted via the SeqOne bioinformatics solution.

Performance

- Whole exome coverage: ~22,000 genes and 37.5 Megabases targeted
- Depth > 30X for ~98% bases[2]
- > 99% of recall[3]

Why Eurofins Biomnis?

- Specialised clinical pathology laboratory
- ISO 15189 accreditation
- Authorisation to perform constitutional genetic testing
- Certified clinical pathologists
- Expertise in sequencing techniques in diagnostic practice
- Permanent biopathological support: implementation of the test, interpretation, participation in MDCMs, possibility of PNDs.

Choose the interpreting solution that's right for you

To actively involve partner clinicians and clinical pathologists in the interpretation of data, Eurofins Biomnis provides them with access to **SeqOne** a secure bioinformatics interface.

With this platform, you can either:

- interpret the genomic data of your patients **jointly** with Eurofins Biomnis (co-interpretation)
- or interpret the data **autonomously.**

SeqOne GENOMICS

Practical details

	Exome sequencing & CNV analysis	
Interpretation level	**Detailed** report	**Basic** report
Turnaround time	• **4 weeks** excluding any additional examinations	2 weeks
Indication	Hereditary predisposition to cancers	
Sample	**Solo:** 5 mL EDTA whole blood or DNA sample **Duo/Trio:** 5mL EDTA whole blood or DNA sample per relative	
Conservation & transport	Room temperature	
Required documents	**B67-INTGB** analysis request form available on **www.eurofins-biomnis.com** > Test guide > Analysis code **EXONC**	
Price	Contact us	
Associated tests	• Gene panels: Breast/Ovary and Prostate Panel **(EOSOP)**, Digestive System **(EODIG)**, Pancreas **(EOPAN)**, Kidney **(EOREI)**, Skin **(EOPEA)**, Lung **(EOPOU)**, Neuro-endocrine **(EONEN)**, "Extended Oncogenetics" **(EOETE)**, Retinoblastoma **(CUR10)** • Study of relatives by targeted screening technique (Sanger or qPCR) • "Reinterpretation" or "Opening of filters" of an analysis	
Associated document	**"Whole Exome Sequencing"** brochure **(code DS34-INTGB)** available on www.eurofins-biomnis.com > Specialities > Genetics > Prenatal constitutional genetics	

Key points

- **Exhaustive analysis** of known and validated genes implicated in genetic predisposition to cancer and of candidate genes or also identification of new genes.
- **Re-interpretation** of data retrospectively based on advancement in knowledge or in other clinical contexts.
- **Discussion** of the results at a staff meeting to confirm changes in points of interest before formalising the report.

References

INCa (Institut National du Cancer), Unicancer and Genetic and Cancer Group.

[1] Target CDS Refseq +/- 2 base pairs

[2] Data calculated from SNV's from NIST002 reference samples, for 40 million pairs of reads generated.

For more information

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