iBio DIAGNOSTICS

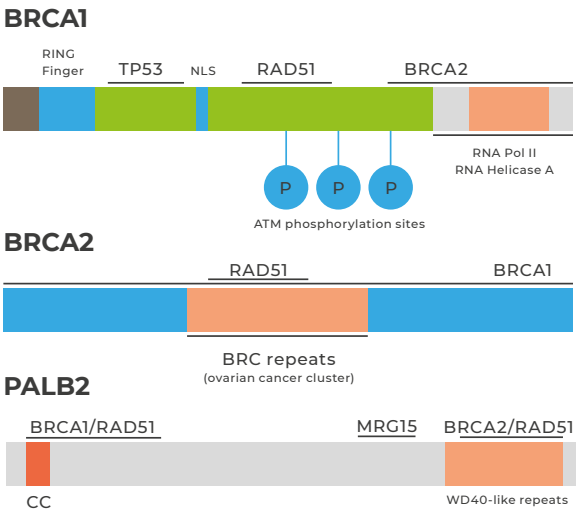
iBio - BRCA PANEL

EN

BRCA1, BRCA2 AND *PALB2* GENES

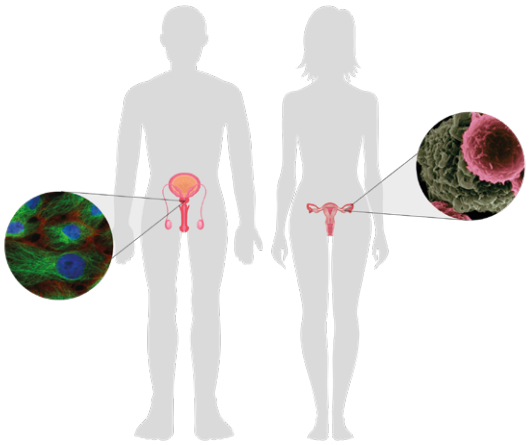
- *BRCA1, BRCA2* and *PALB2* are tumor suppressor genes that encode proteins involved in DNA repair (homologous recombination).
- Mutations appear in 4-6% of breast cancer, 10-15% of ovarian cancer and 6-8% of prostate cancer cases.
- Over 2000 mutations are recognized that result in loss of protein function.
- Mutations predict response to poly (ADP)ribose polymerase inhibitor (PARPi) therapy.
- **ESMO* recommends testing for *BRCA1* and *BRCA2* mutations in tumor tissue in non-mucinous ovarian cancer and metastasizing prostate cancer cases.**

* European Society for Medical Oncology



OVARIAN CANCER

- Ovarian cancer has the highest mortality among cancers of the female genital tract.
- There are approximately 300.000 cases of ovarian cancer globally each year, almost 200.000 are fatal.
- 70% of all ovarian cancer is high grade serous carcinoma; mutations of *BRCA1* and *BRCA2* genes are most common in this subtype.



PROSTATE CANCER

- Prostate cancer is the second most common cancer in men.
- Mean survival of patients with hormone therapy-resistant prostate cancer is only 2-4 years.
- Mutations of DNA-repair genes predict response to PARPi treatment as in ovarian cancer.

METHOD

- Single test to reveal somatic as well as germline mutations.
- Formalin fixed paraffin embedded (FFPE) tissue block or >10 slides may be used.
- Tumor cell ratio of ≥20% is needed.
- Sequencing of total coding regions as well as 3'/5' UTRs of *BRCA1, BRCA2* and *PALB2* genes.
- Bioinformatic identification of single nucleotide variants (SNV), short insertions and deletions.
- Variant classification and annotation (e.g. ClinVar, COSMIC, HGMD, BRCA Exchange).

COVERAGE: >95% (>500×)

AVERAGE SEQUENCING DEPTH: >1000×

TURNAROUND TIME: 2-3 WEEKS

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iBio SCIENCE

UNIVERSITY OF PÉCS
SZENTÁGOTHAI RESEARCH CENTRE

WORKFLOW

Our mission is advancing scientific research in the fields of **BIOTECHNOLOGY** and **MEDICINE** as well as applying the latest innovative technologies in diagnostics. **IBIOSCIENCE LTD.** in collaboration with **UNIVERSITY OF PÉCS SZENTÁGOTHAI RESEARCH CENTER** provides state-of-the-art next generation sequencing services and expertise for the Hungarian scientific community.

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02

SAMPLE DISPATCH
FFPE block or slides,
cell blocks or smears,
or isolated DNA

03

SAMPLE PROCESSING
microscopical control
of tumor cell ratio,
DNA isolation

04

GENETIC ANALYSIS
bioinformatic analysis,
variant identification
and annotation

05

REPORT
categorization of variants
based on guidelines
(pathogenic, likely
pathogenic, VUS etc.)

REFERENCES

Neff RT et al. Ther Adv Med Oncol. 2017 Aug;9(8):519-531. PMID: 28794804
Colombo N, et al. Ann Oncol. 2019 May 1;30(5):672-705. PMID: 31046081
Moschetta M et al. Ann Oncol. 2016 Aug;27(8):1449-55. PMID: 27037296
Mosele F et al. Ann Oncol. 2020 Nov;31(11):1491-1505. PMID: 32853681
Richards S et al. Genet Med. 2015 May;17(5):405-24. PMID: 25741868
Li MM et al. J Mol Diagn. 2017 Jan;19(1):4-23. PMID: 27993330

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