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The earlier, the better – turning data into knowledge

Predisposition analyses of hereditary cancers

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This might interest you:

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The benefits of our applications for hereditary cancer predispositions

Automated analysis of hereditary cancer predispositions

Our applications for hereditary cancer predispositions are optimized for automated identification of clinically-significant germline variants associated with hereditary breast and ovarian cancers (HBOC), and other hereditary cancer predispositions.

How your laboratory benefits from our solutions:

- **Approved for diagnostic use**
  Our applications for HBOC and other hereditary cancers are modules of MH Guide, which is approved as an IVD medical device in the EU and intended for diagnostic use.
- **Faster results**
  Automatic access to relevant databases, as well as variant pre-classification according to ACMG criteria and genotype-phenotype correlations.
- **Easy to integrate**
  Flexible interfaces make it possible to analyze standard data formats from the sequencing of commercially-available or proprietary gene panels, independent of the platform used.
- **Customizable evaluation**
  The filtering and editing options allow quick access to the most important information as well as the integration of proprietary databases.
- **Audited quality**
  Molecular Health is certified to EN ISO 13485:2016. Users benefit from the safety and reliability of MH software applications.

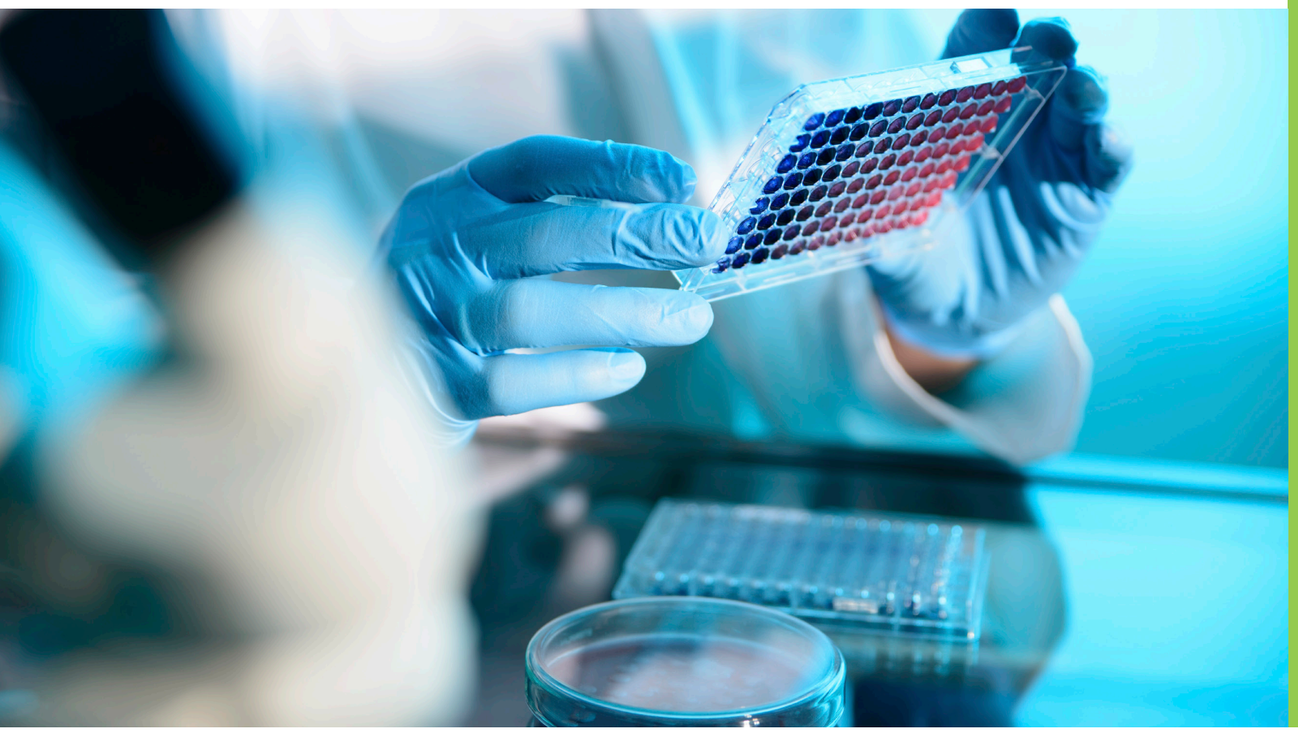
Questions? Please contact us on +49 6221 43851-150 or via CustomerService@molecularhealth.com

Identify hereditary cancer predispositions – quickly, accurately, and efficiently

Our software applications analyze gene variants in comparison with data from Dataome, one of the world's largest knowledge bases for biomedical information. The software provides annotation data from recognized databases such as ClinVar, BRCAExchange, and UTAH BRCA, which are updated regularly.

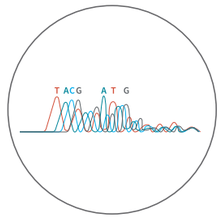
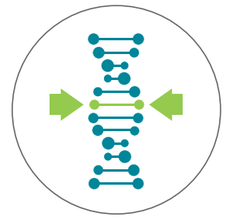
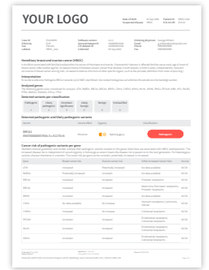
You can use any commercially-available or proprietary gene panels and have the data analyzed in VCF format with our applications. Likewise, raw data (FASTQ) from common Illumina platforms can be analyzed.

The software summarizes all of the relevant results in individual reports that provide users with clear, specific information on possible pathogenic gene mutations associated with HBOC and other hereditary cancers.



2

The analyses process

Genomic data from a blood sample	Interpretation of genetic variants	Reporting
		
Technology-independent data upload: • VCF, FASTQ files • Commercial panels • Proprietary panels	Automated database comparison and classification • Public databases • MH database • ACMG classification	Standardized, customizable report • Your summary • Summary of pathogenic variants • All reported variants with classification

3

Integration in lab workflows

Flexibility and data security in one

The web-based software applications can be easily integrated in the laboratory. They enable the annotation and interpretation of genetic variants from common NGS or other analysis platforms.

Approved for clinical use
Our applications for HBOC and other hereditary cancers are modules of MH Guide, a software application approved in Europe as an IVD medical device (according to Regulation (EU) 2017/746 (IVDR).

SaaS – individually scalable
Our applications are offered as scalable SaaS (Software as a Service) to suit small and large institutions alike.

Secure data transmission
Our applications provide secure transmission of patient data through advanced encryption standards (SSL/TLS, AES-256) and storage of patient data with controlled access authorization.

Guaranteed security of patient data
Our applications comply with GDPR in Europe, GenDG in Germany, and the Health Insurance Portability and Accountability Act (HIPAA) in the USA.

Customizable patient reports
The design, content, and format of analysis reports can be adapted to individual needs on request.

Flexible input and output formats
Our applications can process the standard data formats VCF and FASTQ. Output formats are PDF, JSON, and XML.

Efficient workflows in your lab
Our applications let you optimize your everyday processes. The cloud-based software automates the interpretation of germline variants and delivers high-quality analyses.

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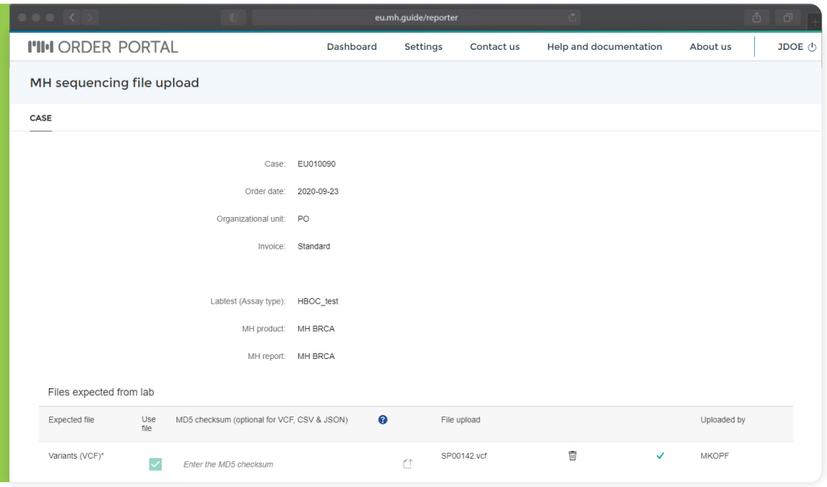
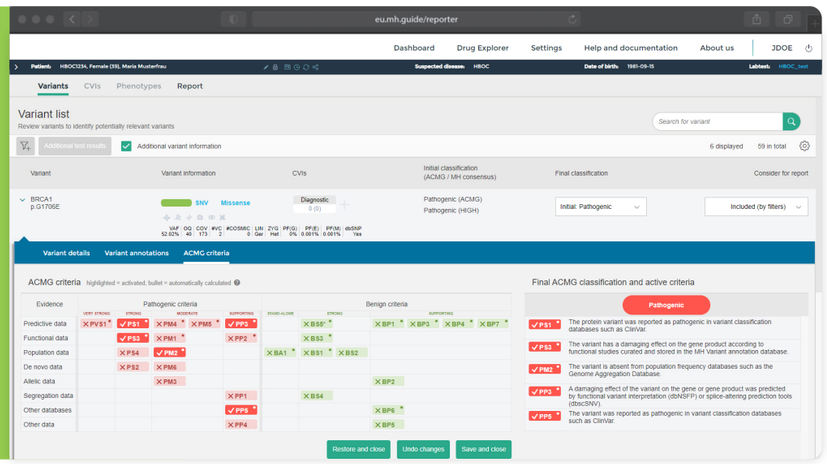
Generate the report: just a few, intuitive steps

How it looks on your screen: from raw data upload to final patient report

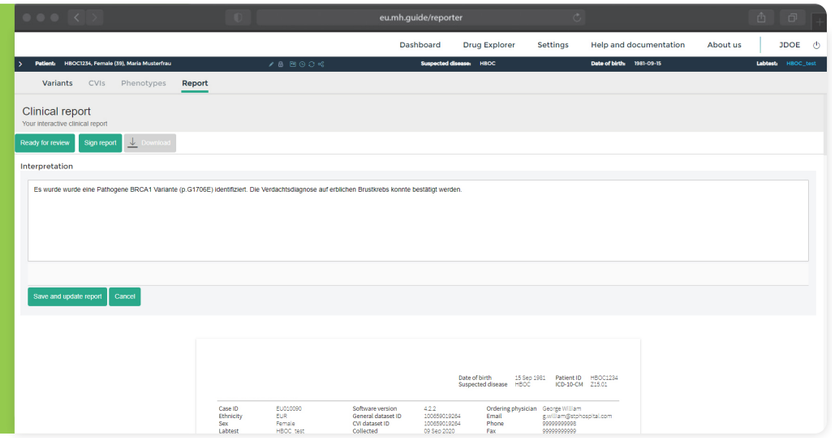
1. Upload sequence data easily, via the MH Order Portal



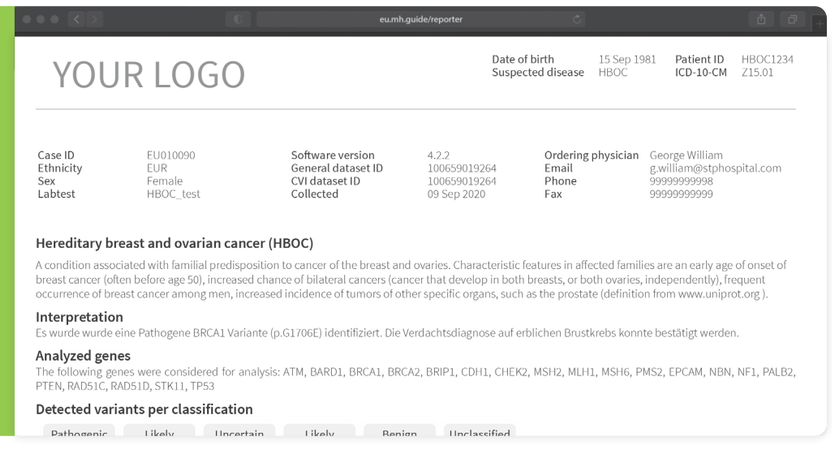
2. Automatic variant classification



3. Generate the report



4. Export the report (detailed description on following page)



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Everything at a glance:*

Patient data and suspected diagnosis

Summary of the disease, your interpretation of the findings, and analyzed genes

Number of detected variants per classification

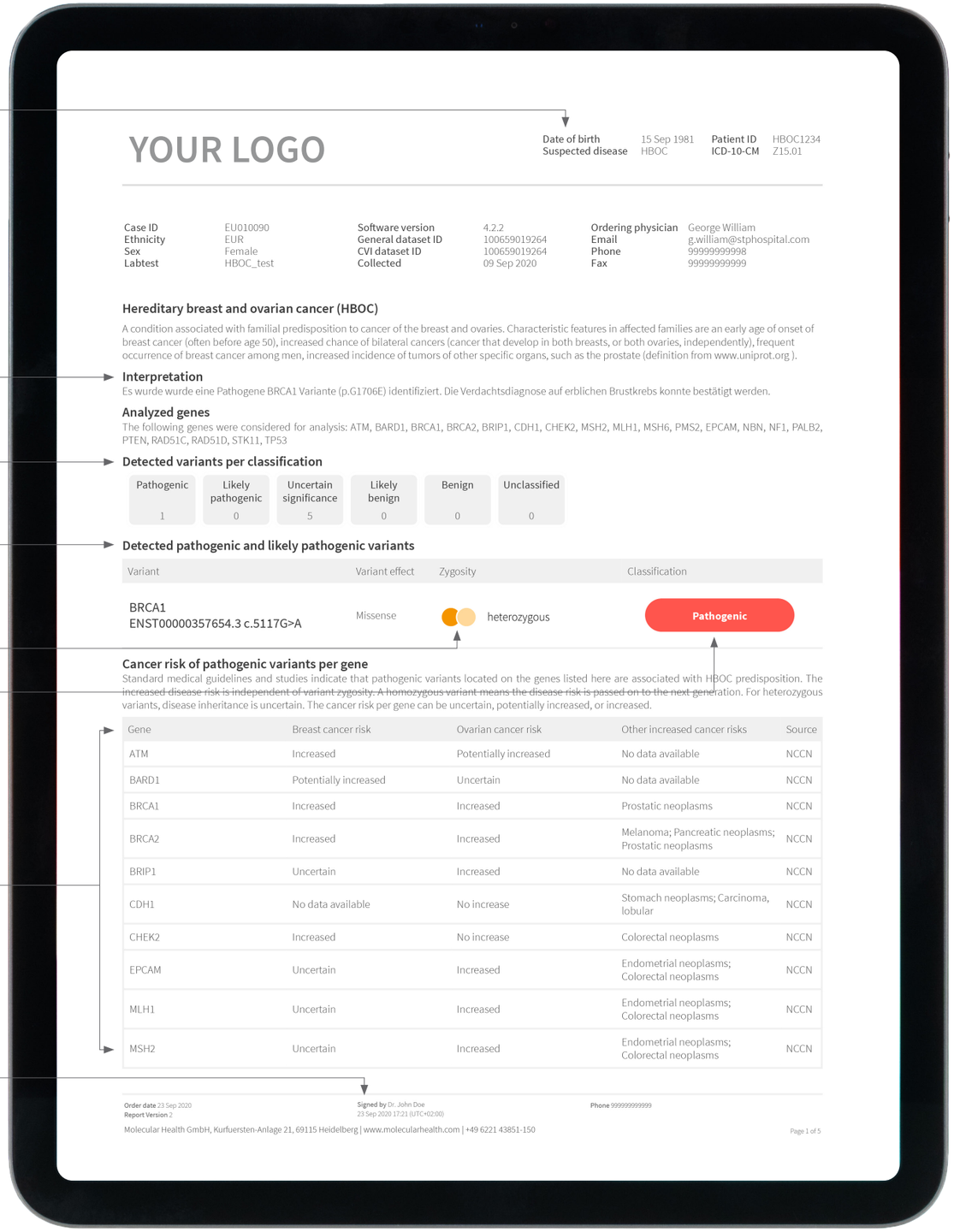
Summary of detected pathogenic variants

Zygosity

Variant classification according to ACMG

List of analyzed genes and inferred pathogenicity of the variant

Electronic signature of the human geneticist in charge



YOUR LOGO

Date of birth 15 Sep 1981 | Patient ID HBOC1234
Suspected disease HBOC | ICD-10-CM Z15.01

Case ID EU010090 | Software version 4.2.2 | Ordering physician George William
Ethnicity EUR | General dataset ID 100659019264 | Email g.william@stphospital.com
Sex Female | CVI dataset ID 100659019264 | Phone 99999999998
Labtest HBOC_test | Collected 09 Sep 2020 | Fax 99999999999

Hereditary breast and ovarian cancer (HBOC)

A condition associated with familial predisposition to cancer of the breast and ovaries. Characteristic features in affected families are an early age of onset of breast cancer (often before age 50), increased chance of bilateral cancers (cancer that develop in both breasts, or both ovaries, independently), frequent occurrence of breast cancer among men, increased incidence of tumors of other specific organs, such as the prostate (definition from www.uniprot.org).

Interpretation
Es wurde wurde eine Pathogene BRCA1 Variante (p.G1706E) identifiziert. Die Verdachtsdiagnose auf erblichen Brustkrebs konnte bestätigt werden.

Analyzed genes
The following genes were considered for analysis: ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MSH2, MLH1, MSH6, PMS2, EPCAM, NBN, NF1, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53

Detected variants per classification

Pathogenic	Likely pathogenic	Uncertain significance	Likely benign	Benign	Unclassified
1	0	5	0	0	0

Detected pathogenic and likely pathogenic variants

Variant	Variant effect	Zygosity	Classification
BRCA1 ENST00000357654.3 c.5117G>A	Missense	heterozygous	Pathogenic

Cancer risk of pathogenic variants per gene
Standard medical guidelines and studies indicate that pathogenic variants located on the genes listed here are associated with HBOC predisposition. The increased disease risk is independent of variant zygosity. A homozygous variant means the disease risk is passed on to the next generation. For heterozygous variants, disease inheritance is uncertain. The cancer risk per gene can be uncertain, potentially increased, or increased.

Gene	Breast cancer risk	Ovarian cancer risk	Other increased cancer risks	Source
ATM	Increased	Potentially increased	No data available	NCCN
BARD1	Potentially increased	Uncertain	No data available	NCCN
BRCA1	Increased	Increased	Prostatic neoplasms	NCCN
BRCA2	Increased	Increased	Melanoma; Pancreatic neoplasms; Prostatic neoplasms	NCCN
BRIP1	Uncertain	Increased	No data available	NCCN
CDH1	No data available	No increase	Stomach neoplasms; Carcinoma, lobular	NCCN
CHEK2	Increased	No increase	Colorectal neoplasms	NCCN
EPCAM	Uncertain	Increased	Endometrial neoplasms; Colorectal neoplasms	NCCN
MLH1	Uncertain	Increased	Endometrial neoplasms; Colorectal neoplasms	NCCN
MSH2	Uncertain	Increased	Endometrial neoplasms; Colorectal neoplasms	NCCN

Order date 23 Sep 2020
Report Version 2
Signed by Dr. John Doe
23 Sep 2020 17:21 (UTC+02:00)
Phone 99999999999

Molecular Health GmbH, Kurfuersten-Anlage 21, 69115 Heidelberg | www.molecularhealth.com | +49 6221 43851-150

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*Sample report of an HBOC predisposition analysis

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How to reach Molecular Health

Molecular Health GmbH
Kurfuersten-Anlage 21
69115 Heidelberg
Germany, Europe

Tel. +49 6221 43851-150
CustomerService@molecularhealth.com

We develop and deliver innovative technologies for in silico medicine and precision medicine

Our solutions enable the conversion of large amounts of data into evidence-based, medically relevant results for the actors in the healthcare sector. Therewith, we provide molecular pathologists, geneticists, physicians, and patients with better information on diagnoses and therapy options. We support pharmaceutical and health organizations by optimizing clinical studies in the development of promising active ingredients and meaningful disease models.