# my GeneticRisk

Genetic test to assess the risk for common diseases





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### my GeneticRisk

#### What is myGeneticRisk?

myGeneticRisk is the preventive genetic test to determine the hereditary risk of cardiovascular disease and cancer, the most common conditions worldwide. Knowing the predisposition to these diseases allows the specialist to establish medical management strategies to prevent them or detect them in early stages, when treatment is typically more effective.

#### Who is it intended for?

The test is indicated for adults who are proactive in their healthcare.

#### Why is it important?



5-20%

Between **5-20%**<sup>1</sup> of cancer cases are hereditary in origin, the percentage varies depending on the type of cancer.



About **30%**<sup>2</sup> of sudden deaths are due to genetic abnormalities related to the structure of the heart muscle or heart rhythm.



More than **5%**<sup>3</sup> of people have a variant in genes recommended to be analysed by international genetics societies (ACMG), as they are related to actionable diseases.

\*American College of Medical Genetics and Genomics

75%<sup>4</sup> of people with a risk variant related to cancer or familial hypercholesterolemia do not have a known family history.

#### What are the advantages of the test?

- It is carried out **once in a lifetime** and allows preventive medicine strategies to be established.
- Based on Whole Exome Sequencing (WES), myGeneticRisk analised 162 genes related to hereditary risk of cancer and cardiovascular disease, including the genes recommended by the ACMG related to actionable diseases.
- If a variant is detected, it can be used **to screen family members who may be at risk.**
- Veritas provides a **differential service offering counselling** to the physician or the patient for result interpretation, when needed.

#### Why is genetic information key? The new era of preventive medicine

The incorporation of genetic information into patient care and health check-ups provides key information before the onset of disease symptoms, allowing a truly preventive approach moving from "sick care" to "healthcare".



#### Which genes are included?

#### 100 Genes related to inherited cardiovascular disease

- Cardiomyopathies
- Arrhythmias

- Syndromes with vascular involvement
- Other syndromes linked to cardiac pathology
- RASopathies

Familial hypercholesterolemia

ABCC9	CACNA1C	CSRP3	FHL1	KCNE1	LIPA	MYLK	PRKG1	SLC2A10	TGFBR2
ABCG5	CALM1	DES	FKTN	KCNE2	LMNA	NEXN	PTPN11	SMAD3	TMEM43
ABCG8	CALM2	DSC2	FLNA	KCNH2	LOX	NF1	RAF1	SMAD4	TNNC1
ACTA2	CALM3	DSG2	FLNC	KCNJ2	MAP2K1	NOTCH1	RBM20	SOS1	TNNI3
ACTC1	CASQ2	DSP	FXN	KCNQ1	MAP2K2	NRAS	RIT1	SOS2	TNNT2
ACTN2	CAV3	EFEMP2	GAA	KRAS	MYBPC3	PCSK9	RYR2	TAZ	TPM1
APOB	CBL	ELN	GLA	LAMP2	MYH11	PKP2	SCN5A	TCAP	TRDN
APOE	COL3A1	EMD	HRAS	LDB3	MYH7	PLN	SHOC2	TGFB2	TTN
BAG3	COX15	FBN1	JPH2	LDLR	MYL2	PPP1CB	SKI	TGFB3	TTR
BRAF	CRYAB	FBN2	JUP	LDLRAP1	MYL3	PRKAG2	SLC25A4	TGFBR1	VCL

#### 40 Genes related to the most frequent hereditary cancers

<ul><li>Breast cancer</li><li>Gynaecological</li><li>Prostate cancer</li></ul>			<ul><li>Colored</li><li>Gastric</li></ul>	ctal cancer cancer	• P • S	<ul> <li>Pancreatic cancer</li> <li>Skin cancer</li> </ul>			
APC*	ATM*	AXIN2	BAP1	BARD1	BMPR1A	BRCA1*	BRCA2*	BRIP1	CDH1
CDK4	CDKN2A	CHEK2*	EPCAM*	FLCN	GREM1*	HOXB13	MITF	MLH1*	MLH3
MSH2*	MSH3	MSH6*	MUTYH	NBN	NF1	NTHL1	PALB2*	PMS2*	POLD1
POLE	POT1	PTCH1	PTEN*	RAD51C	RAD51D	SMAD4	STK11*	SUFU	TP53*

\* Genes including analysis of copy number variations.

#### Other genes recommended by the ACMG <sup>5</sup>

myGeneticRisk includes the analysis of a heterogeneous group of actionable diseases such as haemochromatosis, malignant hyperthermia or Maturity Onset Diabetes of the Young (MODY).

ACVRL1	ATP7B	BTD	CACNA1S	ENG	HFE	HNF1A	MAX
MEN1	NF2	OTC	RB1	RET	RPE65	RYR1	SDHAF2
SDHB	SDHC	SDHD	TMEM127	TSC1	TSC2	VHL	WT1

## Actionable disease: a medical approach exists to prevent its development or detect it early.

#### Opportunistic screening for common inherited diseases:

why family history is not enough

Recent studies show that the current elegibility criteria to use genetic testing for cardiovascular and cancer risk screening exclude a significant percentage of the population with risk variants.<sup>4</sup>

The graphs show the total number of people in the study who have mutations related to a certain pathology. The percentage of them who meet the criteria for genetic testing is indicated.



#### References

1. Nielsen FC, et al. Hereditary breast and ovarian cancer: new genes in confined pathways. Nat Rev Cancer. 2016 Sep;16(9):599-612.

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3. Internal data

4. Grzymski JJ, et al. Population genetic screening efficiently identifies carriers of autosomal dominant diseases. Nat Med. 2020;26(8):1235-1239.

5. Miller DT, et al. ACMG SF v3.0 list for reporting of secondary findings in clinical exome and genome sequencing: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021 Aug;23(8):1381-1390.

#### **About Veritas**

Veritas Genetics, a LetsGetChecked company, is one of the world leaders in advanced genetic sequencing and clinical interpretation of the exome and whole genome, driving the transition to personalized and preventive medicine.

Using state-of-the-art technologies and the highest safety standards, Veritas Genetics helps individuals, healthcare professionals and institutions worldwide to understand and anticipate genetic risks, enabling more informed and proactive health decisions.

With a focus on innovation and accessibility, Veritas Genetics transforms the way we understand and care for health at every stage of life.



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