Hereditary Disease Risk Genetic Test 2.0 & Medication Response Genetic Test

What is the Hereditary Disease Risk Test?

The Color Hereditary Disease Risk Test 2.0 analyzes 60 genes to help you understand your potential for certain hereditary cancer, heart health, and other actionable health concerns that are recognized as medically actionable by the American College of Medical Genetics and Genomics (ACMG). This also includes the Medication Response Genetic Test that analyzes 20 genes that influence how your body may process certain medications, offering insights to support informed health planning with your care team. Additionally, it provides insights into genetic ancestry and traits, allowing you to discover how your genes may influence your preferences, appearance, and other fun genetic facts.

What Does This Test Look For?

Hereditary Disease Risk Test: Analyzes 60 genes for genetic variants across several health areas including cancer, heart health, metabolism, and neurocutaneous conditions. These genes include:

ACTA2, ACTC1, APC, APOB, ATP7B, BMPR1A, BRCA1, BRCA2, CACNA1S, COL3A1, DSC2, DSG2, DSP, EPCAM, FBN1, GLA, KCNH2, KCNQ1, LDLR, LMNA, MEN1, MLH1, MSH2, MSH6, MUTYH, MYBPC3, MYH11, MYH7, MYL2, MYL3, NF2, OTC, PCSK9, PKP2, PMS2, PRKAG2, PTEN, RB1, RET, RYR1, RYR2, SCN5A, SDHAF2, SDHB, SDHC, SDHD, SMAD3, SMAD4, STK11, TGFBR1, TGFBR2, TMEM43, TNNI3, TNNT2, TP53, TPM1, TSC1, TSC2, VHL, WT1

Medication Response Genetic Test: Analyzes 20 genes that affect how your body processes medications, helping guide safer, more effective treatments. These genes include:

ABCG2, CACNA1S, CYP1A2, CYP2C cluster, *CYP2C19, CYP2C9, CYP2D6, CYP3A4, CYP3A5, CYP4F2, DPYD, F5, G6PD, IFNL3, NUDT15, RYR1, SLCO1B1, TPMT, UGT1A1, VKORC1*

Discovery: Explores insights about your genetic ancestry and learn about how your genes influence unique characteristics such as lactose intolerance, alcohol flush response, cilantro preference, and more.

To learn more, see the Gene Tables on page 3.

Hereditary Disease Risk Test

What are the possible results?

Something important is found

A meaningful genetic change is identified that could impact your health.

Nothing significant is found

No increased risk for the hereditary conditions were found in the genes tested.

How can these results impact my healthcare?

Earlier interventions, at every step: Insights into your health can guide your healthcare providers in recommending regular screenings or lifestyle changes that suit your needs.

Medication choices: Your results can help guide your doctor in selecting the safest and most effective medications for you.

Family health insights: Your results may also provide useful health information for family members, as some genetic risks can be shared among relatives.

What happens after I get my results?

Color's care team will ensure you understand the result and the next steps associated with the result to take action on your health. We recommend that you share your Color test results with your healthcare provider. This can help you and your provider create a personalized healthcare plan. If you don't have your own healthcare provider, Color's care team can help get you connected to one in your area.

Hereditary Disease Risk Test - Cancer

Gene	Hereditary Condition	Associated Cancers
APC	Familial adenomatous polyposis (FAP), Attenuated FAP	Colon, thyroid, brain, stomach, small bowel
BMPR1A	Juvenile polyposis syndrome	Colon, stomach
BRCA1	Hereditary breast and ovarian cancer syndrome	Breast, ovarian, pancreatic, prostate
BRCA2	Hereditary breast and ovarian cancer syndrome	Breast, ovarian, melanoma, pancreatic, prostate
EPCAM	Lynch syndrome	Colon, uterine, ovarian, stomach, pancreatic, prostate
MEN1	Multiple endocrine neoplasia type 1	Thyroid
MLH1	Lynch syndrome	Colon, uterine, ovarian, stomach, pancreatic, prostate
MSH2	Lynch syndrome	Colon, uterine, ovarian, stomach, pancreatic, prostate
MSH6	Lynch syndrome	Colon, uterine, ovarian, stomach, prostate
MUTYH	MUTYH-associated polyposis	Colon, uterine, ovarian, stomach, prostate
PMS2	Lynch syndrome	Colon, uterine, ovarian, stomach
PTEN	PTEN hamartoma tumor syndrome	Breast, thyroid, uterine, kidney, colon
RB1	Retinoblastoma	Eye
RET	Multiple endocrine neoplasia type 2	Thyroid
SDHAF2	Paraganglioma-pheochromocytoma (PGL-PCC) syndrome	Endocrine, kidney, stomach
SDHB	Paraganglioma-pheochromocytoma (PGL-PCC) syndrome	Endocrine, kidney, stomach
SDHC	Paraganglioma-pheochromocytoma (PGL-PCC) syndrome	Endocrine, kidney, stomach
SDHD	Paraganglioma-pheochromocytoma (PGL-PCC) syndrome	Endocrine, kidney, stomach
SMAD4	Juvenile polyposis syndrome	Colon, stomach
STK11	Peutz-Jeghers syndrome	Breast, colon, stomach
TP53	Li-Fraumeni syndrome	Breast, colon, brain, pancreatic, sarcoma
VHL	Von-Hippel Lindau	Brain, kidney
WT1	WT1 disorder	Kidney

Hereditary Disease Risk Test - Heart

Gene	Hereditary Condition
ACTA2	Familial thoracic aortic aneurysm and dissection (FTAAD)
ACTC1	Cardiomyopathy
APOB	Familial hypercholesterolemia
COL3A1	Vascular Ehlers-Danlos syndrome, FTAAD
DSC2	Cardiomyopathy
DSG2	Cardiomyopathy
DSP	Cardiomyopathy
FBN1	Marfan syndrome, FTAAD
GLA	Fabry Disease, Cardiomyopathy
KCNH2	Arrhythmia, Long QT syndrome, Short QT syndrome
KCNQ1	Arrhythmia, Long QT syndrome, Short QT syndrome
LDLR	Familial hypercholesterolemia
LMNA	Cardiomyopathy
MYBPC3	Cardiomyopathy
MYH11	FTAAD
MYH7	Cardiomyopathy
MYL2	Cardiomyopathy
MYL3	Cardiomyopathy
PCSK9	Familial hypercholesterolemia
PKP2	Cardiomyopathy
PRKAG2	Cardiomyopathy
RYR2	Arrhythmia, Catecholaminergic polymorphic ventricular tachycardia
SCN5A	Arrhythmia, Brugada syndrome, Long QT syndrome
SMAD3	Loeys-Dietz syndrome, FTAAD
TGFBR1	Loeys-Dietz syndrome, FTAAD
TGFBR2	Loeys-Dietz syndrome, FTAAD
TMEM43	Cardiomyopathy
TNNI3	Cardiomyopathy
TNNT2	Cardiomyopathy
TPM1	Cardiomyopathy

Hereditary Disease Risk Test - Other

Gene	Hereditary Condition
ATP7B	Wilson disease
CACNA1S	Malignant hyperthermia susceptibility
NF2	*NF2*-related schwannomatosis
OTC	Ornithine transcarbamylas deficiency
RYR1	Malignant hyperthermia susceptibility
TSC1	Tuberous sclerosis complex
TSC2	Tuberous sclerosis complex

Medication Response Genetic Test

Gene	Star alleles and variants analyzed
ABCG2	rs2231142
CACNA1S	ENST00000362061: reference, c.520C>T, c.3257G>A
CYP1A2	*1, *30 (*1F)
CYP2C cluster	rs12777823
CYP2C9	*1, *2, *3, *4, *5, *6, *8, *9, *11, *12, *13, *14, *15, *16, *23, *24, *26, *29, *31, *33, *35, *39, *42, *43, *44, *45, *46, *55, *61
CYP2C19	*1, *2, *3, *4, *5, *6, *7, *8, *9, *10, *16, *17, *19, *22, *24, *25, *26, *35, *36 (whole gene deletion), *37 (partial gene deletion), *38
CYP2D6	*1, *2, *3, *4, *4N (hybrid, a.k.a. *4.013), *5 (whole gene deletion), *6, *7, *8, *9, *10, *11, *12, *13 (hybrid), *14, *15, *17, *18, *19, *21, *29, *31, *32, *35, *36 (hybrid), *40, *41, *42, *45, *49, *54, *55, *56, *59, *68 (hybrid), *69, *114, *119, *xN
CYP3A4	*1, *20, *22
CYP3A5	*1, *3, *6, *7
CYP4F2	*1, *2, *3, *4, rs2108622
DPYD	ENST00000370192: reference (*1), c.299_302del (*7), c.557A>G, c.703C>T (*8), c.868A>G, c.1129-5923C>G (HapB3), c.1156G>T (*12), c.1314T>G, c.1475C>T, c.1679T>G (*13), c.1774C>T, c.1898del (*3), c.1905+1G>A (*2A), c.2279C>T, c.2639G>T, c.2846A>T, c.2983G>T (*10), rs3918290, rs55886062.1 A>C, rs75017182, rs56038477, rs67376798, rs115232898
F5	rs6025
G6PD	"A- 202A_376G", "A- 968C_376G", "Asahi", "B (reference)", "Canton, Taiwan-Hakka, Gifu-like, Agrigento-like", "Chatham", "Chinese-5", "Gaohe", "Ilesha", "Kaiping, Anant, Dhon, Sapporo-like, Wosera", "Kalyan-Kerala, Jamnaga, Rohini", "Malaga", "Mediterranean, Dallas, Panama, Sassari, Cagliari, Birmingham", "Orissa", "Quing Yuan, Chinese-4", "Seattle, Lodi, Modena, Ferrara II, Athens-like", "Ube Konan", "Union, Maewo, Chinese-2, Kalo", "Viangchan, Jammu"
IFNL3	rs12979860
NUDT15	*1, *2, *3, *4, *6, *9, *14, rs116855232
RYR1	ENST00000359596: reference, c.38T>G, c.97A>G, c.103T>C, c.130C>T, c.131G>A, c.463C>A, c.487C>T, c.488G>T, c.529C>T, c.533A>G, c.742G>A, c.742G>C, c.982C>T, c.1021G>A, c.1021G>C, c.1201C>T, c.1202G>A, c.1202G>T, c.1565A>C, c.1565A>G, c.1589G>A, c.1597C>T, c.1615T>C, c.1615T>G, c.1630G>T, c.1654C>T, c.1655G>A, c.1840C>T, c.1841G>A, c.1841G>T, c.3166G>C, c.5183C>T, c.6349G>C, c.6387C>G, c.6487C>T, c.6488G>A, c.6488G>C, c.6488G>T, c.6502G>A, c.6612C>G, c.6617C>G, c.6617C>T, c.6628G>T, c.6757C>T, c.6838G>A, c.7007G>A, c.7035C>A, c.7036G>A, c.7042_7044del, c.7043A>G, c.7048G>A, c.7060G>A, c.7063C>T, c.7076G>A, c.7084G>A, c.7090T>G, c.7123G>A, c.7124G>C, c.7282G>A, c.7291G>A, c.7291G>T, c.7300G>A, c.7304G>A, c.7304G>T, c.7310C>T, c.7354C>T, c.7358T>C, c.7360C>T, c.7361G>A, c.7372C>T, c.7373G>A, c.7373G>T, c.7522C>T, c.7523G>A, c.7879G>C, c.8026C>T, c.9310G>A, c.11315G>A, c.11708G>A, c.11947C>T, c.11958C>G, c.11969G>T, c.12149C>A, c.12700G>C, c.12700G>T, c.14209C>T, c.14210G>A, c.14477C>T, c.14497C>T, c.14512C>G, c.14539G>C, c.14545G>A, c.14627A>G, c.14803G>A, c.14918C>T
SLCO1B1	*1, *5, *9, *14, *15, *20, *31, *46, *47, rs2306283, rs4149056
TPMT	*1, *2, *3A, *3B, *3C, *4, *8, *11, *14, *15, *23, *24, *29, *41, *42
UGT1A1	*1, *6, *27, *28, *36, *37
VKORC1	rs9923231, rs72547529, rs61742245

Technical details

Technology

Color uses next generation sequencing (NGS) and custom bioinformatics pipelines to analyze genes related to hereditary disease risk and medication response. DNA is extracted from the sample and enriched for specific regions using a hybridization-based method. Sequencing is performed on Illumina platforms, and data are processed to identify a wide range of genetic changes, including single nucleotide variants, small insertions/deletions, copy number variants, and select structural variants. More than 99% of targeted gene regions are sequenced at high depth (≥50X), ensuring accurate and reliable variant detection. For medication response, additional analysis is performed to assign star alleles and predict drug metabolism phenotypes based on established pharmacogenetic guidelines.

Sequencing is performed in a Clinical Laboratory Improvement Amendments (CLIA)-certified and College of American Pathologists (CAP)-accredited laboratory (CAP #8975161 - CLIA #05D2081492).

All variants are interpreted using guidelines from the American College of Medical Genetics and Genomics (ACMG) and reviewed by qualified clinicians. Reports focus on pathogenic and likely pathogenic variants with potential clinical relevance. Benign and likely benign variants are not reported. Variants of uncertain significance (VUS) are noted but details are typically not reported unless clinically relevant or requested.

Limitations

While highly sensitive, the test does not detect all possible genetic changes. Some complex variant types, such as large rearrangements, certain duplications, or variants in difficult-to-sequence region, may not be detected or may be reported with reduced confidence. Phase (whether two variants are inherited on the same or different copies of a gene) is generally not determined.

For Hereditary Disease Risk Test, limitations apply to specific genes:

- *APOB*: Analysis limited to codon 3527 (chr2:g.21229159_21229161).
- *ATP7B*: Only biallelic pathogenic and/or likely pathogenic variants are reported; single heterozygous variants are not. VUSs are not reported.
- *EPCAM*: Analysis limited to large deletions involving exons 8 and/or 9.
- *KCNH2*: Exons 4 and 14 are not analyzed.
- *KCNQ1*: Exon 1 not analyzed.
- *MUTYH*: Only biallelic pathogenic and/or likely pathogenic variants are reported; single heterozygous variants are not. VUSs are not reported.
- *MYBPC3*: Exon 11 not analyzed.
- *MYH7*: VUSs are not reported for exon 27.
- *NF2*: Exon 12 not analyzed.
- *PMS2*: VUSs are not reported for exons 12-15. Analysis excludes five variants commonly observed in the pseudogene *PMS2CL*: c.2182_2184delinsG, c.2243_2246del, c.2444_2445insTT, c.2523G>A, and deletion of exons 13-14 (chr7:g.6015768_6018727del).
- *PRKAG2*: Exon 5 not analyzed.
- *RYR1*: Exon 91 not analyzed.
- *SMAD4*: Structural variant detection may be impacted by pseudogene.
- *TGFBR1*: Exon 1 not analyzed.
- *WT1:* Exon 1 not analyzed

For Medication Response Genetic Test, not all star alleles or gene variations are analyzed. Specific limitations include:

- *CYP2D6*: Sensitivity to detect copy number >3 is limited; some hybrid alleles not reported.
- *CYP2C19*: Copy number not always determined; partial deletions and duplications may be missed.
- *G6PD*: Chromosome X aneuploidies are not reported.

Additional limitations include reduced sensitivity in regions of high or low GC content, repetitive sequences, and insertions/deletions between 40–250 base pairs. For medication response genetic test, not all possible allele combinations or phasing between variants can be resolved. In cases where results are ambiguous, the diplotype may be reported as “indeterminate.”

Recent blood transfusions, bone marrow or organ transplants, or certain cancer treatments may affect test accuracy.