

# Hereditary Disease Risk Test

## | What is the Hereditary Disease Risk Test?

The Color Hereditary Disease Risk Test 2.0 analyzes 59 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 test also includes 14 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 59 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, TNNT2, TNNT2, TP53, TPM1, TSC1, TSC2, VHL, WT1*



**Medication Response Genetic Test:** Looks at 14 genes that affect how bodies processes medications, helping guide safer, more effective treatments. These genes include:

*CYP1A2, CYP2C19, CYP2C9, CYP2D6, CYP3A4, CYP3A5, CYP4F2, DPYD, F5, IFNL3, NUDT15, SLCO1B1, TPMT, 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.**

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

# Hereditary Disease Risk Test - Heart

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

# Hereditary Disease Risk Test - Other

Gene	Hereditary Condition
<i>ATP7B</i>	Wilson disease
<i>CACNA1S</i>	Malignant hyperthermia susceptibility
<i>NF2</i>	Neurofibromatosis type 2
<i>OTC</i>	Ornithine transcarbamylase deficiency
<i>RYR1</i>	Malignant hyperthermia susceptibility
<i>TSC1</i>	Tuberous sclerosis complex
<i>TSC2</i>	Tuberous sclerosis complex

# Medication Response Genetic Test

Gene	Star alleles and variants analyzed
<i>CYP1A2</i>	*1, *1F, *1K
<i>CYP2C9</i>	*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
<i>CYP2C19</i>	*1, *2, *3, *4, *5, *6, *7, *8, *9, *10, *16, *17, *19, *22, *24, *25, *26, *35, *38
<i>CYP2D6</i>	*1, *2, *3, *4, *4N, *5, *6, *7, *8, *9, *10, *11, *12, *13, *14, *15, *17, *18, *19, *21, *29, *31, *32, *35, *36, *40, *41, *42, *45, *49, *54, *55, *56, *59, *68, *69, *114
<i>CYP3A4</i>	*1, *20, *22
<i>CYP3A5</i>	*1, *3, *6, *7
<i>CYP4F2</i>	*1, *2, *3, *4, rs2108622
<i>DPYD</i>	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
<i>F5</i>	rs6025
<i>IFNL3</i>	rs12979860
<i>NUDT15</i>	*1, *2, *3, *4, *6, *9, *14, rs116855232
<i>SLCO1B1</i>	*1, *5, *9, *14, *15, *20, *31, *46, *47, rs2306283, rs4149056
<i>TPMT</i>	*1, *2, *3A, *3B, *3C, *4, *8, *11, *14, *15, *23, *24, *29, *41, *42