Extended Genetic Test

What is the Extended Genetic Test?

The Extended Genetic Test looks at three important health areas: hereditary cancer, heart health, and how one processes medicine. This test checks 79 genes to give a broad view of how genes may affect one's health. It can help you and your healthcare providers plan for prevention, early screening, and care that's right for you. 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 Cancer Genetic Test: Looks at 29 genes for genetic variants that may increase risk for the most common hereditary cancer types. These genes include:

APC, ATM, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A (p14ARF and p16INK4a)*, CHEK2, EPCAM, GREM1, MITF, MLH1, MSH2, MSH6, MUTYH, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SMAD4, STK11, TP53*

Hereditary Heart Health Genetic Test: Looks at 30 genes linked to hereditary heart conditions, which may affect the heart's structure or how it functions. These genes include:

ACTA2, ACTC1, APOB, COL3A1, DSC2, DSG2, DSP, FBN1, GLA, KCNH2, KCNQ1, LDLR, LMNA, MYBPC3, MYH11, MYH7, MYL2, MYL3, PCSK9, PKP2, PRKAG2, RYR2, SCN5A, SMAD3, TGFBR1, TGFBR2, TMEM43, TNNI3, TNNT2, TPM1

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*

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

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.

Extended Genetic 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 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 Cancer Genetic Test

Gene	Hereditary Condition	Associated Cancers
APC	Familial adenomatous polyposis (FAP), Attenuated FAP	Colon, thyroid, brain, stomach, small bowel
ATM	Hereditary breast and pancreatic cancer	Breast, pancreatic
BAP1	BAP1 tumor predisposition syndrome	Melanoma, kidney, lung
BARD1	Hereditary breast cancer	Breast
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
BRIP1	Hereditary ovarian cancer	Ovarian
CDH1	Hereditary diffuse gastric cancer	Breast, stomach
CDK4	Familial atypical mole-malignant melanoma syndrome	Melanoma, pancreatic
CDKN2A (p14ARF and p16INK4a)	FAMMM, Melanoma and neural system tumor syndrome	Melanoma, pancreatic, nervous system
CHEK2	Hereditary breast cancer	Breast, prostate
EPCAM	Lynch syndrome	Colon, uterine, ovarian, stomach, pancreatic, prostate
GREM1	Hereditary mixed polyposis syndrome	Colon
MITF	Hereditary melanoma and kidney cancer	Melanoma, kidney
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
PALB2	Hereditary breast and ovarian cancer	Breast, ovarian, pancreatic
POLD1	Polymerase proofreading-associated polyposis	Colon
POLE	Polymerase proofreading-associated polyposis	Colon
PMS2	Lynch syndrome	Colon, uterine, ovarian, stomach
PTEN	PTEN hamartoma tumor syndrome	Breast, thyroid, uterine, kidney, colon
RAD51C	Hereditary breast and ovarian cancer	Breast, ovarian
RAD51D	Hereditary breast and ovarian cancer	Breast, ovarian
SMAD4	Juvenile polyposis syndrome	Colon, stomach
STK11	Peutz-Jeghers syndrome	Breast, colon, stomach
TP53	Li-Fraumeni syndrome	Breast, colon, brain, pancreatic, sarcoma

Hereditary Heart Health Genetic Test

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

Medication Response Genetic Test

Gene	Star alleles and variants analyzed
ABCG2	rs2231142
CACNA1S	ENST00000362061: c.520C>T, c.3257G>A
CYP1A2	*1, *1F, *1K
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, *36 (whole gene deletion), *37 (partial gene deletion), *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, *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", "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", "Sierra Leone", "Ube Konan", "Union, Maewo, Chinese-2, Kalo", "Viangchan, Jammu"
IFNL3	rs12979860
NUDT15	*1, *2, *3, *4, *6, *9, *14, rs116855232
RYR1	ENST00000359596: 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