Genes Analyzed (ACMG SF v3.2)

Genes related to cancer risk

Gene(s)	Condition associated with this gene	Associated cancers
APC	Familial adenomatous polyposis (FAP), Attenuated FAP	Colon, thyroid, brain, stomach, small bowel
BMPR1A	Juvenile polyposis syndrome	Colon, stomach
SMAD4	Juvenile polyposis syndrome, Hereditary hemorrhagic telangiectasia syndrome	Colon, stomach
BRCA1, BRCA2, PALB2	Hereditary breast and ovarian cancer syndrome	Breast, ovarian, pancreatic, prostate
PMS2, MLH1, MSH2, MSH6	Lynch syndrome	Colon, uterine, ovarian, stomach, prostate
MUTYH	MUTYH-associated polyposis	Colon, uterine, ovarian, stomach, prostate
PTEN	PTEN hamartoma tumor syndrome	Breast, thyroid, uterine, kidney, colon
RB1	Retinoblastoma	Eye
SDHAF2, SDHB, SDHC, SDHD, MAX, TMEM127	Paraganglioma-pheochromocytoma (PGL-PCC) syndrome	Endocrine, kidney, stomach
RET	Multiple endocrine neoplasia type 2	Thyroid
MEN1	Multiple endocrine neoplasia type 1	Thyroid
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

Genes related to heart condition risk

Gene(s)	Condition associated with this gene
ACTA2, MYH11	Familial thoracic aortic aneurysm and dissection (FTAAD)
FBN1	Marfan syndrome , FTAAD
SMAD3, TGFBR1, TGFBR2	Loeys-Dietz syndrome , FTAAD
COL3A1	Vascular Ehlers-Danlos syndrome, FTAAD
DSC2, DSG2, PKP2, TMEM43	arrhythmogenic cardiomyopathy
DSP	arrhythmogenic cardiomyopathy and dilated cardiomyopathy
CASQ2, RYR2, TRDN	Arrhythmia, catecholaminergic polymorphic ventricular tachycardia
BAG3, DES, FLNC, LMNA, RBM20, TNNC1	dilated cardiomyopathy
TNNT2	dilated cardiomyopathy and hypertrophic cardiomyopathy
MYH7	
ACTC1, MYBPC3, MYL2, MYL3, PRKAG2, TNNI3, TPM1	Hypertrophic cardiomyopathy
APOB, LDLR, PCSK9	Familial hypercholesterolemia
KCNH2, KCNQ1	Arrhythmia, long QT syndrome , Short QT syndrome
SCN5A	Arrhythmia, Brugada syndrome, long QT syndrome
CALM1, CALM2, CALM3	Long QT syndrome types 14-16
GLA	Fabry disease , Cardiomyopathy

Genes related to other disease risks

Gene(s)	Condition associated with this gene
ATP7B	Wilson disease
BTD	Biotinidase deficiency
CACNA1S, RYR1	malignant hyperthermia
GAA	Pompe disease
HFE	Hereditary hemochromatosis
ACVRL1, ENG	Hereditary hemorrhagic telangiectasia
HNF1A	Maturity-Onset of Diabetes of the Young
NF2	neurofibromatosis type 2
OTC	ornithine carbamoyltransferase (OTC) deficiency
RPE65	RPE65-related retinopathy
TSC1, TSC2	tuberous sclerosis complex
TTR	Hereditary TTR (transthyretin) amyloidosis

Methodology

Genes tested: 81 genes (ACMG Secondary Findings v3.2)
DNA sequencing: DNA was extracted from whole blood or saliva samples. Whole genome sequencing at 30x coverage using Illumina short reads on a NovaSeq X Series sequencer, performed by SAMPLED.
Sequence Alignment and Variant Calling: Reads from sequence data were aligned to a reference genome and bioinformatic tools were used to detect single nucleotide variants and small insertions/deletions.
Analysis: Focused on pathogenic/likely pathogenic variants reported in clinvar.com as of 04/21/2025; recessive carrier status not reported. This was performed by bioinformatic researchers of the ABCD study.
Limitations:

- In very rare cases recent transplant/transfusion may confound results.
- Large scale genetic changes (structural and copy number variants) were not tested in this analysis
- This analysis is limited to previously reported pathogenic and likely pathogenic variants in clinvar.com at the time of analysis.