arcensus

Incidental Findings

Incidental findings are unexpected results that are not related to the primary reason for testing. These pathogenic or likely pathogenic genetic variants increase the patient's risk of developing a disease that is medically actionable.

The American College of Medical Genetics and Genomics (ACMG) has published guidelines that provide a standardized framework and a curated list of genes associated with actionable conditions where established interventions or treatments can mitigate the risk or severity of the condition. Arcensus reports incidental genetic findings that are based on the recommendations of the ACMG v3.3 guidelines and can have medical benefits for patients undergoing clinical sequencing and their families.

The consent form provides all patients with the choice to opt-out of receiving incidental findings.

PHENOTYPE	GENE	INHERITANCE	VARIANTS
Genes Related to Cancer Phenotypes			
Familial adenomatous polyposis	*APC*	AD	All P and LP
Familial medullary thyroid cancer / multiple endocrine neoplasia 2	*RET*	AD	All P and LP
Hereditary breast and / or ovarian cancer	*BRCA1* *BRCA2* *PALB2*	AD	All P and LP
Hereditary paraganglioma-pheochromocytoma syndrome	*SDHD* *SDHAF2* *SDHC* *SDHB* *MAX* *TMEM127*	AD	All P and LP
Juvenile polyposis syndrome	*BMPR1A*	AD	All P and LP
Juvenile polyposis syndrome/ hereditary hemorrhagic telangiectasia syndrome	*SMAD4*	AD	All P and LP
Li-Fraumeni syndrome	*TP53*	AD	All P and LP
Lynch syndrome (hereditary nonpolyposis colorectal cancer)	*MHL2* *MSH2* *MSH6* *PMS3*	AD	All P and LP
Multiple endocrine neoplasia type 1	*MEN1*	AD	All P and LP
MUTYH-associated polyposis	*MUTYH*	AR	P and LP
Peutz-Jeghers syndrome	*STK11*	AD	All P and LP
PTEN hamartoma tumor syndrome	*PTEN*	AD	All P and LP
Retinoblastoma 1	*RB1*	AD	All P and LP
Tuberous sclerosis complex	*TSC1* *TSC2*	AD	All P and LP

PHENOTYPE	GENE	INHERITANCE	VARIANTS
Von Hippel-Lindau syndrome	*VHL*	AD	All P and LP
WT1-related Wilms tumor	*WT1*	AD	All P and LP
Genes related to cardiovascular phenotypes			
Aortopathies	*FBN1* *TGFBR1* *TGFBR2* *SMAD3* *ACTA2* *MYH11*	AD	All P and LP
Arrhythmogenic right ventricular cardiomyopathy	*PKP2* *DSP* *DSC2* *TMEM43* *DSG2*	AD	All P and LP
Catecholaminergic polymorphic ventricular tachycardia	*RYR2* *CASQ2* *TRDN*	AD AR AR	All P and LP P and LP P and LP
Dilated cardiomyopathy	*TNNT2* *LMNA* *FLNC* *TTN* *BAG2* *DES* *RBM20* *TNNC1* *PLN*	AD	All P and LP*
Ehlers-Danlos syndrome, vascular type	*COL3A1*	AD	All P and LP
Familial hypercholesterolemia	*LDLR* *APOB* *PCSK9*	AD / AR AD AD	All P and LP
Hypertrophic cardiomyopathy	*MYH7* *MYBPC3* *TNNI3* *TPM1* *MYL3* *ACTC1* *PRKAG2* *MYL2* *TNNT2* *FLNC*	AD AD/AR AD AD AD/AR AD AD AD AD AD	All P and LP
Long QT syndrome types 1 and 2	*KCNQ1* *KCNH2*	AD	All P and LP
Long QT syndrome type 3, Brugada syndrome	*SCN5A*	AD	All P and LP
Long QT syndrome types 14-16	*CALM1* *CALM2* CALM3	AD	All P and LP
Genes related to inborn errors of metabolism phenotypes			
Biotinidase deficiency	*BTD*	AR	P and LP
Cerebrotendinous xanthomatosis	*CYP27A1*	AR	P and LP
Fabry disease	*GLA*	XL	All P and LP
Ornithine transcarbamylase deficiency	*OTC*	XL	All P and LP

PHENOTYPE	GENE	INHERITANCE	VARIANTS
Pompe disease	*GAA*	AR	P and LP
X-linked adrenoleukodystrophy	*ABCD1*	XL	All hemi/hom or 2 het P or LP
Genes related to miscellaneous phenotypes			
Hereditary hemochromatosis	*HFE*	AR	p.C282Y hom only
Hereditary hemorrhagic telangiectasia	*ACVRL2* *ENG*	AD	All P and LP
Malignant hyperthermia	*RYR1* *CACNA1S*	AD	All P and LP
Maturity-onset of diabetes of the young	*HNF1A*	AD	All P and LP
RPE65-related retinopathy	*RPE65*	AR	P and LP
Wilson disease	*ATP7B*	AR	P and LP
Hereditary amyloidosis	*TTR*	AD	All P and LP

** Only P/LP LMNA variants that have any case level phenotype evidence of association with cardiac disease. Only P/LP TTN frameshift and nonsense variants, and variants known to impact the splicing.*
AD: Autosomal Dominant; AR: Autosomal Recessive; XL: X-linked; P: Pathogenic, LP: Likely Pathogenic
Hom: homozygous, Het: heterozygous, Hemi: hemizygous

In case you have questions please send an email to
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