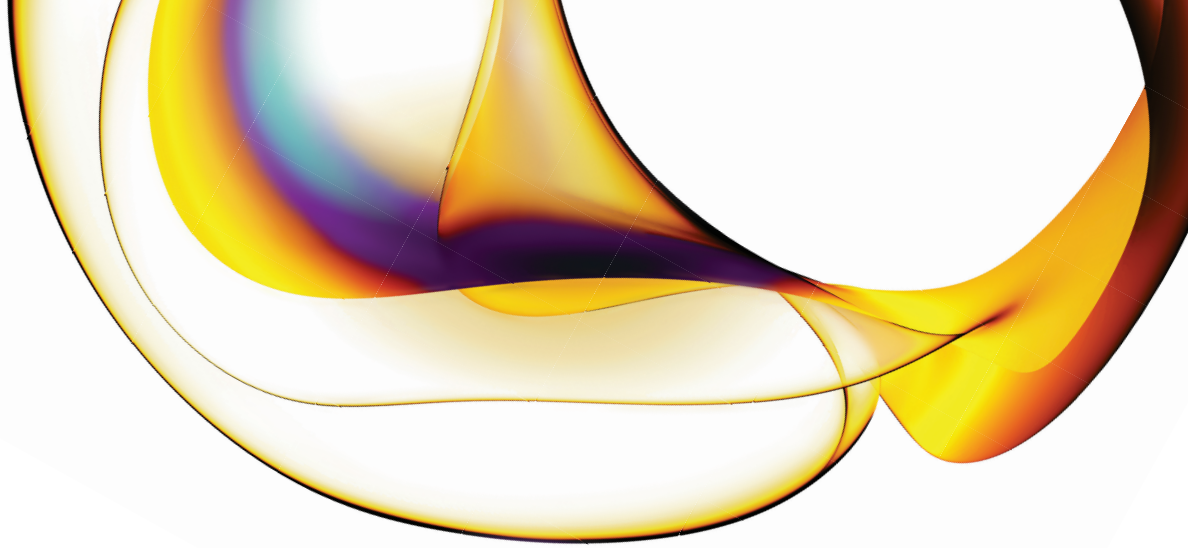

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Whole Exome and Whole Genome Sequencing

Secondary Findings | ACMG Incidental Findings

Gene	Gene MIM	Disease/Phenotype	Inheritance	Typical age of onset
ACTA2	102620	Familial thoracic aortic aneurysm; Multisystemic smooth muscle dysfunction syndrome; Moyamoya disease	AD	Child / Adult
ACTC1	102540	ACTC1 - Related Disorders	AD	Child / Adult
ACVRL1	601284	Hereditary hemorrhagic telangiectasia	AD	Child / Adult
APC	611731	APC-Associated Polyposis Conditions	AD	Child / Adult
APOB	107730	Familial hypercholesterolemia; Hypobetalipoproteinemia	AD/AR	Child / Adult
ATP7B	606882	Wilson disease	AR	Child / Adult
BMPR1A	601299	Juvenile polyposis syndrome	AD	Child / Adult
BRCA1	113705	BRCA1 - related cancer susceptibility	AD	Adult
BRCA2	600185	BRCA2 - related cancer susceptibility; Fanconi Anemia	AD/AR	Adult
BTD	609019	Biotinidase deficiency	AR	Child / Adult
CACNA1S	114208	CACNA1S - Related Disorders	AD	Child / Adult
CASQ2	114251	Catecholaminergic polymorphic ventricular tachycardia	AR	Child / Adult
COL3A1	120180	Ehlers-Danlos syndrome, vascular type	AD	Child / Adult
DSC2	125645	Arrhythmogenic right ventricular cardiomyopathy	AD	Child / Adult
DSG2	125671	Arrhythmogenic right ventricular cardiomyopathy	AD	Child / Adult

Gene	Gene MIM	Disease/Phenotype	Inheritance	Typical age of onset
DSP	125647	DSP - Related Disorders	AD/AR	Child / Adult
ENG	131195	Hereditary hemorrhagic telangiectasia	AD	Child / Adult
FBN1	134797	FBN1 - Related Disorders	AD	Child / Adult
FLNC	102565	FLNC - Related Disorders	AD	Adult
GAA	606800	Pompe disease	AR	Child / Adult
GLA	300644	Fabry disease	XL	Child / Adult
HFE	613609	Hereditary hemochromatosis (c.845G>A; p.C282Y homozygotes only)	AR	Adult
HNF1A	142410	Maturity-Onset of Diabetes of the Young	AD	Child / Adult
KCNH2	152427	Long-QT syndrome type 2	AD	Child / Adult
KCNQ1	607542	KCNQ1 - Related Disorders	AD / AR	Child / Adult
LDLR	606945	Familial hypercholesterolemia	AD	Child / Adult
LMNA	150330	LMNA - Related Disorders	AD/AR	Child / Adult
MAX	154950	Hereditary paraganglioma-pheochromocytoma syndrome	AD	Child / Adult
MEN1	613733	Multiple endocrine neoplasia type 1	AD	Child / Adult
MLH1	120436	Lynch syndrome	AD	Adult
MSH2	609309	Lynch syndrome	AD	Adult
MSH6	600678	Lynch syndrome	AD	Adult
MUTYH	604933	MUTYH -associated polyposis	AR	Adult
MYBPC3	600958	Hypertrophic cardiomyopathy; Dilated cardiomyopathy, Left ventricular noncompaction	AD	Child / Adult
MYH11	160745	Familial thoracic aortic aneurysm	AD	Child / Adult
MYH7	160760	MYH7 - Related Disorders	AD	Child / Adult
MYL2	160781	Hypertrophic cardiomyopathy	AD	Child / Adult
MYL3	160790	Hypertrophic cardiomyopathy	AD	Child / Adult
NF2	607379	Neurofibromatosis type 2	AD	Child / Adult
OTC	300461	Ornithine transcarbamylase deficiency	XL	Child
PALB2	610355	Hereditary breast cancer; Fanconi Anemia	AD/AR	Adult
PCSK9	607786	Familial hypercholesterolemia	AD	Child / Adult
PKP2	602861	Arrhythmogenic right ventricular cardiomyopathy	AD	Child / Adult
PMS2	600259	Lynch syndrome; mismatch repair cancer syndrome	AD/AR	Adult

Gene	Gene MIM	Disease/Phenotype	Inheritance	Typical age of onset
PRKAG2	602743	Hypertrophic cardiomyopathy; Glycogen storage disease of heart; Wolff-Parkinson-White syndrome	AD	Child / Adult
PTEN	601728	PTEN hamartoma tumor syndrome	AD	Child / Adult
RB1	614041	Retinoblastoma	AD	Child / Adult
RET	164761	Multiple endocrine neoplasia type 2 A/B; Familial medullary thyroid carcinoma	AD	Child / Adult
RPE65	180069	RPE65 -related retinopathy	AR	Child / Adult
RYR1	180901	RYR1 - Related Disorders	AD/AR	Child / Adult
RYR2	180902	Catecholaminergic polymorphic ventricular tachycardia; Arrhythmogenic right ventricular dysplasia	AD	Child / Adult
SCN5A	600163	SCN5A - Related Disorders	AD	Child / Adult
SDHAF2	613019	Hereditary paraganglioma-pheochromocytoma syndrome	AD	Child / Adult
SDHB	185470	Hereditary paraganglioma-pheochromocytoma syndrome	AD	Child / Adult
SDHC	602413	Hereditary paraganglioma-pheochromocytoma syndrome	AD	Child / Adult
SDHD	602690	Hereditary paraganglioma-pheochromocytoma syndrome	AD	Child / Adult
SMAD3	603109	Loeys-Dietz syndrome	AD	Child / Adult
SMAD4	600993	SMAD4 - Related Disorders	AD	Child / Adult
STK11	602216	Peutz-Jeghers syndrome	AD	Child / Adult
TGFBR1	190181	Loeys-Dietz syndrome	AD	Child / Adult
TGFBR2	190182	Loeys-Dietz syndrome	AD	Child / Adult
TMEM127	613403	Hereditary paraganglioma-pheochromocytoma syndrome	AD	Child / Adult
TMEM43	612048	Arrhythmogenic right ventricular cardiomyopathy	AD	Child / Adult
TNNI3	191044	Hypertrophic cardiomyopathy	AD	Child / Adult
TNNT2	191045	TNNT2 - Related Disorders	AD	Child / Adult
TP53	191170	Li-Fraumeni syndrome	AD	Child / Adult
TPM1	191010	Hypertrophic cardiomyopathy; Dilated cardiomyopathy, Left ventricular noncompaction	AD	Child / Adult
TRDN	603283	Long QT syndrome; catecholaminergic polymorphic ventricular tachycardia	AR	Child / Adult

Gene	Gene MIM	Disease/Phenotype	Inheritance	Typical age of onset
TSC1	605284	Tuberous sclerosis complex	AD	Child
TSC2	191092	Tuberous sclerosis complex	AD	Child
TTN	188840	TTN - Related Disorders (DCM for ACMG)	AD/AR	Child / Adult
VHL	608537	Von Hippel-Lindau syndrome	AD	Child / Adult
WT1	607102	WT1 -related Wilms tumor	AD	Child
BAG3	603883	Dilated cardiomyopathy	AD	mainly adult, ped reported
DES	125660	Dilated cardiomyopathy	AD/AR	mainly adult, ped reported
RBM20	613171	Dilated cardiomyopathy	AD	Child / Adult
TNNC1	191040	Dilated cardiomyopathy	AD	mainly adult, ped reported
TTR	176300	Hereditary TTR amyloidosis	AD	Adult

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