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| **Supplemental Table 1.** Actionable gene list |
|  | **Phenotype** | **Gene/genes** |
| **Cardiovascular** | Ehlers-Danlos syndrome, vascular type | *COL3A1, COL5A1* |
|  | Marfan syndrome, Loeys-Dietz syndromes, familial thoracic aortic aneurysms & dissections | *FBN1, TGFBR1, TGFBR2, SMAD3, ACTA2, MYH1, MYLK* |
|  | Hypertrophic cardiomyopathy | *MYBPC3, MYH7, TNNT2, TNNI3, TPM, MYL3, ACTC1, PRKAG2,**GLA* |
|  | Dilated cardiomyopathy | *MYL2, LMNA* |
|  | Catecholaminergic polymorphic ventricular tachycardia | *RYR2* |
|  | Arrhythmogenic right ventricular cardiomyopathy | *PKP2, DSP ,DSC2,**TMEM43, DSG2* |
|  | Romano-Ward long-QT syndrome types 1, 2, and 3, Brugada syndrome | *KCNQ1, KCNH2, SCN5A,**KCNJ2, KCNE1* |
|  | Familial hypercholesterolemia | *LDLR, APOB, PCSK9* |
|  | Diabetes | *HNF1A, HNF1B* |
| **Cancer** | Hereditary breast and ovarian cancer | *BRCA1, BRCA2,* |
|  | Li-Fraumeni syndrome | *TP53* |
|  | Peutz-Jeghers syndrome | *STK11* |
|  | Lynch syndrome | *MLH1, MSH2, MSH6,**PMS2, POLD1, POLE* |
|  | Familial adenomatous polyposis | *APC* |
|  | *MYH*-associated polyposis; adenomas, multiple colorectal, *FAP* type 2; colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas | *MUTYH* |
|  | Juvenile polyposis | *BMPR1A, SMAD4* |
|  | Von Hippel–Lindau syndrome | *VHL* |
|  | Multiple endocrine neoplasia type 1 | *MEN1* |
|  | Multiple endocrine neoplasia type 2 | *RET* |
|  | Familial medullary thyroid cancerd | *RET* |
|  | *PTEN* hamartoma tumor syndrome | *PTEN* |
|  | Retinoblastoma | *RB1* |
|  | Hereditary paraganglioma-pheochromocytoma syndrome | *SDHD, SDHAF2,**SDHC, SDHB* |
| **Other** | Tuberous sclerosis complex | *TSC1, TSC2* |
|  | WT1-related Wilms tumor | *WT1* |
|  | Neurofibromatosis type 2 | *NF2* |
|  | Wilson disease | *ATP7B* |
|  | Ornithine transcarbamylase deficiency | *OTC* |
|  | Neuromuscular disorders | *RYR1, CACNA1A, CACNA1S* |
| **Supplemental Table 2.** Consensus list of actionable single nucleotide variants  |
| **rs#** | **Gene** | **Associated Disease** | **Disease Category** |
| rs77931234 | *ACADM* | Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency | Inborn error of metabolism |
| rs387906225 | *ALDOB* | Hereditary fructose intolerance | Inborn error of metabolism |
| rs386834233 | *BCKDHB* | Maple syrup urine disease | Inborn error of metabolism |
| rs79761867 | *BCKDHB* | Maple syrup urine disease | Inborn error of metabolism |
| rs80338898 | *FAH* | Tyrosinemia type I | Inborn error of metabolism |
| rs1801175 | *G6PC* | Glycogen storage disease type I | Inborn error of metabolism |
| rs397509431 | *CPT2* | Carnitine palmitoyltransferase II (CPT II) deficiency | Inborn error of metabolism |
| rs113993962 | *BLM* | Bloom Syndrome | Cancer susceptibility |
| rs193922376 | *MSH2* | Lynch syndrome# | Cancer susceptibility |
| rs6467 | *CYP21A2* | 21-hydroxylase deficiency | Endocrinology |
| rs6025 | *F5* | factor V Leiden thrombophilia\* | Thrombophilia |
| rs1800562 | *HFE* | Hereditary hemochromatosis | Iron storage |
| rs28940579 | *MEFV* | Familial Mediterranean fever | Inflammatory |
| rs61752717 | *MEFV* | Familial Mediterranean fever | Inflammatory |

Only bi-allelic (homozygous, or if applicable compound heterozygous) variants will be returned.

Mode of inheritance is autosomal recessive except #=autosomal dominant and \*=risk increased.