SUDCRRC
SUDC Registry and Research Collaborative

**ACMG GENE LIST FOR SECONDARY/UNRELATED FINDINGS**

As of March 2013 (updated 2016) the ACMG specifically recommends testing for the following list of 59 specific genes for 31 diseases/disorders in which findings would have medical benefit for the patients and families of patients. These genes include some cancer or tumor syndromes, some connective tissue diseases, cardiomyopathies, and arrhythmias. Some of these conditions have onset in adulthood and an individual may not have recognizable features now. These disorders were selected because there may be changes in medical management for an individual if the individual is known to have a genetic susceptibility to one or more of these disorders. Details regarding individual genes on this list will be provided to you upon request.

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| Hereditary breast and ovarian cancer | BRCA1, BRCA2 |
| Li-Fraumeni syndrome | TP3 |
| Familial adenomatous polyposis | APC |
| Peutz-Jeghers syndrome | STK11 |
| Lynch syndrome | MLH1, MSH2, MSH6, PMS2 |
| MYH-associated polyposis | MUTYH |
| Juvenile polyposis | BMPR1A, SMAD4 |
| Von Hippel-Lindau syndrome | VHL |
| Multiple endocrine neoplasia type 1 | MEN1 |
| Multiple endocrine neoplasia type 2 | RET |
| PTEN hamartoma tumor syndrome | PTEN |
| Retinoblastoma | RB1 |
| Hereditary paraganglioma-pheochromocytoma syndrome | SDHD, SDHAF2, SDHC, SDHB |
| Tuberous sclerosis complex | TSC1, TSC2 |
| WT1-related Wilms tumor | WT1 |
| Neurofibromatosis type 2 | NF2 |
| Vascular, Ehlers Danlos syndrome | COL3A1 |
| Familial thoracic aortic aneurysm | ACTA2, MYH11 |
| Marfan syndrome | FBN1 |
| Loeys-Dietz syndrome | TGFBR1, TGFBR2, SMAD3 |
| Hypertrophic cardiomyopathy, dilated cardiomyopathy | MYBPC3, MYH7, TNNT2, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA |
| Catecholaminergic polymorphic ventricular tachycardia | RYR2 |

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| Arrhythmogenic right ventricular cardiomyopathy | PKP2, DSP, DSC2, TMEM43, DSG2 |
| Long QT syndrome | KCNQ1, KCNH2 |
| Long QT syndrome; Brugada sydnrome; dilated cardiomyopathy | SCN5A |
| Familial hypercholesteremia | LDLR, APOB, PCSK9 |
| Wilson disease | ATP7B |
| Ornithine transcarbamylase deficiency | OTC |
| Malignant hyperthermia susceptibility | RYR1, CACNA1S |

If a genetic change is identified in one of these genes, further testing for that specific gene may be recommended. The absence of a reportable finding in these genes does not mean that an individual has no disease-causing changes in these genes, so future testing if the individual has symptoms or features of one of the conditions caused by these genes should be considered. Coverage of these genes through whole exome sequencing may not be as comprehensive as in panels specifically designed to investigate them.