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| **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.