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NAME	CONFIDENCE	STATUS
Hemochromatosis (HFE-related)	★★★★	Variant Present
Phenylketonuria	★★★★	Variant Absent
Familial Dysautonomia	★★★★	Variant Absent
Canavan Disease	★★★★	Variant Absent
Familial Hyperinsulinism (ABCC8-related)	★★★★	Variant Absent
Primary Hyperoxaluria Type 2 (PH2)	★★★★	Variant Absent
Sjögren-Larsson Syndrome <span>new</span>	★★★★	Variant Absent
Rhizomelic Chondrodysplasia Punctata Type 1 (RCDP1)	★★★★	Variant Absent
Torsion Dystonia	★★★★	Variant Absent
Autosomal Recessive Polycystic Kidney Disease	★★★★	Variant Absent
TTR-Related Cardiac Amyloidosis	★★★★	Variant Absent
Mucopolidosis IV	★★★★	Variant Absent
Limb-girdle Muscular Dystrophy	★★★★	Variant Absent
Leigh Syndrome, French Canadian Type (LSFC)	★★★★	Variant Absent
Alpha-1 Antitrypsin Deficiency	★★★★	Variant Absent
Congenital Disorder of Glycosylation Type 1a (PMM2-CDG)	★★★★	Variant Absent
DPD Deficiency	★★★★	Variant Absent
Dihydrolipoamide Dehydrogenase Deficiency	★★★★	Variant Absent
Neuronal Ceroid Lipofuscinosis (PPT1-related)	★★★★	Variant Absent
Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency	★★★★	Variant Absent
Glycogen Storage Disease Type 1a	★★★★	Variant Absent
Glycogen Storage Disease Type 1b	★★★★	Variant Absent
Gaucher Disease	★★★★	Variant Absent
ARSACS	★★★★	Variant Absent
G6PD Deficiency	★★★★	Variant Absent
Cystic Fibrosis	★★★★	Variant Absent
Factor XI Deficiency	★★★★	Variant Absent
Zellweger Syndrome Spectrum	★★★★	Variant Absent
Nijmegen Breakage Syndrome	★★★★	Variant Absent
D-Bifunctional Protein Deficiency	★★★★	Variant Absent
Usher Syndrome Type I (PCDH15-related) <span>new</span>	★★★★	Variant Absent
LAMB3-related Junctional Epidermolysis Bullosa	★★★★	Variant Absent
Familial Mediterranean Fever	★★★★	Variant Absent
Usher Syndrome Type III <span>new</span>	★★★★	Variant Absent
TTR-Related Familial Amyloid Polyneuropathy	★★★★	Variant Absent
Pendred Syndrome	★★★★	Variant Absent
Tyrosinemia Type I	★★★★	Variant Absent
Hereditary Fructose Intolerance	★★★★	Variant Absent

Inherited Syndrome	Rating	Variant Status
Tyrosinemia Type I	★★★★	Variant Absent
Hereditary Fructose Intolerance	★★★★	Variant Absent
Familial Hypercholesterolemia Type B	★★★★	Variant Absent
Hypertrophic Cardiomyopathy (MYBPC3 25bp-deletion)	★★★★	Variant Absent
BRCA Cancer Mutations (Selected)	★★★★	Variant Absent
Connexin 26-Related Sensorineural Hearing Loss	★★★★	Variant Absent
Beta Thalassemia	★★★★	Variant Absent
Sickle Cell Anemia & Malaria Resistance	★★★★	Variant Absent
Niemann-Pick Disease Type A	★★★★	Variant Absent
Fanconi Anemia (FANCC-related)	★★★★	Variant Absent
Bloom's Syndrome	★★★★	Variant Absent
Salla Disease	★★★★	Variant Absent
GRACILE Syndrome	★★★★	Variant Absent
Maple Syrup Urine Disease Type 1B	★★★★	Variant Absent
Tay-Sachs Disease	★★★★	Variant Absent
Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)	★★★★	Variant Absent
Neuronal Ceroid Lipofuscinosis (CLN5-related)	★★★★	Variant Absent