Name: Mutze User
Date: 08-06-2024

Name	Results
Alpha-1 Antitrypsin Deficiency	Variant present
Familial adenomatous polyposis	Variant present
Hereditary hemochromatosis associated with HFE	Variant present
Acute intermittent porphyria	Variant absent
Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)	Variant absent
Alpha-mannosidosis	Variant absent
ARSACS (Autosomal recessive spastic ataxia of Charlevoix-Saguenay)	Variant absent
Autosomal recessive polycystic kidney disease	Variant absent
Beta Thalassemia	Variant absent
Biotinidase deficiency	Variant absent
Birt-Hogg-Dube syndrome	Variant absent
Bloom syndrome	Variant absent
Brugada Syndrome	Variant absent
Canavan Disease	Variant absent
cblA Type Methylmalonic aciduria	Variant absent
cblB Type Methylmalonic aciduria	Variant absent
Classical homocystinuria due to CBS deficiency	Variant absent
Complete achromatopsia (type 2) and Incomplete achromatopsia	Variant absent
Congenital disorder of glycosylation type 1a (PMM2-CDG)	Variant absent
Congenital muscular alpha-dystroglycanopathy and Walker-Warburg syndrome	Variant absent
Congenital myasthenic syndrome	Variant absent
Congenital stationary night blindness 1C	Variant absent
Cowden syndrome	Variant absent
Cystic fibrosis	Variant absent
Cystinosis	Variant absent
D-Bifunctional Protein Deficiency	Variant absent
Diastrophic dysplasia	Variant absent
Dihydrolipoamide Dehydrogenase Deficiency	Variant absent
Dilated Cardiomyopathy 1A	Variant absent
Dubin-Johnson syndrome	Variant absent
Ehlers-Danlos Syndrome (EDS)	Variant absent
Familial advanced sleep phase syndrome (FASPS)	Variant absent
Familial breast cancer	Variant absent
Familial dysautonomia (Riley-Day syndrome)	Variant absent
Familial Hypercholesterolemia	Variant absent
Familial Hypertrophic Cardiomyopathy (HCM)	Variant absent
Familial Mediterranean fever	Variant absent

Familial Transthyretin Amyloidosis	Variant absent
Familiar hyperinsulinism (ABCC8-related)	Variant absent
Fanconi Anemia (FANCC-related)	Variant absent
Gaucher disease	Variant absent
Glucose-6-phosphate dehydrogenase deficiency (G6PD deficiency)	Variant absent
Glutaric Acidemia type 1	Variant absent
Glutaric Acidemia type 2	Variant absent
Glycogen storage disease type 1A (Von Gierke Disease)	Variant absent
Glycogen storage disease type 1B	Variant absent
Glycogen storage disease type 3	Variant absent
Glycogen storage disease type 5	Variant absent
Glycogenosis type 2 or Pompe disease	Variant absent
GRACILE syndrome	Variant absent
Hemophilia A	Variant absent
Hereditary fructose intolerance	Variant absent
Homocystinuria due to MTHFR deficiency	Variant absent
Hypokalemic Periodic Paralysis	Variant absent
Hypophosphatasia	Variant absent
Junctional Epidermolysis Bullosa	Variant absent
Leigh Syndrome, French-Canadian type (LSFC)	Variant absent
Leukoencephalopathy with vanishing white matter	Variant absent
Li-Fraumeni Syndrome	Variant absent
Limb-girdle muscular dystrophy	Variant absent
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	Variant absent
Lynch syndrome	Variant absent
Malignant Hyperthermia	Variant absent
Maple syrup urine disease type 1B	Variant absent
Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)	Variant absent
Metachromatic leukodystrophy	Variant absent
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	Variant absent
Mucolipidosis IV	Variant absent
Mucolipidosis type II	Variant absent
Multiple endocrine neoplasia 2B	Variant absent
Neurofibromatosis type I	Variant absent
Neuronal Ceroid-Lipofuscinoses type 1 (associated to PPT1)	Variant absent
Neuronal Ceroid-Lipofuscinoses type 3 (associated to CLN3)	Variant absent
Neuronal Ceroid-Lipofuscinoses type 5 (associated to CLN5)	Variant absent
Neuronal Ceroid-Lipofuscinoses type 6 (associated to CLN6)	Variant absent
Neuronal Ceroid-Lipofuscinoses type 7 (associated to MFSD8)	Variant absent
Niemann-Pick disease type A	Variant absent
Non-syndromic mitochondrial hearing loss	Variant absent
Nonsyndromic Hearing Loss and Deafness, DFNB1	Variant absent

Pendred syndrome	Variant absent
Peters plus syndrome	Variant absent
Phenylketonuria	Variant absent
Pontocerebellar hypoplasia	Variant absent
Primary hyperoxaluria type 1 (PH1)	Variant absent
Primary hyperoxaluria type 2 (PH2)	Variant absent
Pyridoxine-dependent epilepsy	Variant absent
Pyruvate kinase deficiency	Variant absent
Refsum disease	Variant absent
Retinitis pigmentosa	Variant absent
Rhizomelic Chondrodysplasia Punctata Type 1	Variant absent
Salla Disease	Variant absent
Short chain acyl-CoA dehydrogenase deficiency (SCADD)	Variant absent
Sjögren-Larsson syndrome	Variant absent
Tay-Sachs disease	Variant absent
Type 1 Oculocutaneous albinism (tyrosinase negative)	Variant absent
Type 2 oculocutaneous albinism (tyrosinase positive)	Variant absent
Tyrosinemia type I	Variant absent
Usher syndrome	Variant absent
Very long chain acyl-CoA dehydrogenase deficiency (VLCADD)	Variant absent
von Willebrand disease	Variant absent
Wilson disease	Variant absent
Zellweger syndrome	Variant absent

Alpha-1 Antitrypsin Deficiency

It is an autosomal recessive disease characterized by liver dysfunction and chronic obstructive pulmonary disease. Its severity depends on which pathogenic variants in the SERPINA1 gene are involved and how much alpha-1 antitrypsin activity is reduced.

Your result is

Variant present

SNP	GEN OR REGION	GENOTYPE	INTERPRETATION
rs17580	SERPINA1	TA	You have one copy of the c.863A>T variant (PI*S allele) in the SERPINA1 gene associated with alpha-1 antitrypsin deficiency. A person with a single copy of this variant would not have the disease, but can transmit the variant to their offspring.

Familial adenomatous polyposis

An inherited disease characterized by the development of colorectal polyps and an increased predisposition to develop colorectal cancer. It may be caused by the presence of pathogenic variants in the APC and MUTYH genes.

Your result is

Variant present

SNP	GEN OR REGION	GENOTYPE	INTERPRETATION
rs36053993	MUTYH	CT	You have one copy of the c.1187G>A mutation in the MUTYH gene associated with familial adenomatous polyposis. A person with one copy of this mutation would not have the disease, but can transmit the mutation to their offspring.

Acute intermittent porphyria

Acute intermittent porphyria is the most common form of porphyria, a group of metabolic disorders affecting the synthesis of the heme group that forms part of hemoglobin. It is characterized by abdominal pain, gastrointestinal dysfunction and neurological disturbances.

Your result is

Variant absent