tellmeGen™

geraldo augusto de oliveira netto

18:39:22 - 10/06/2019

Inherited Conditions General Report

NAME	STATUS
Short chain acyl-CoA dehydrogenase deficiency (SCADD)	VARIANT PRESENT
ARSACS (Autosomal recessive spastic ataxia of Charlevoix-Saguenay)	VARIANT ABSENT
Acute intermittent porphyria	VARIANT ABSENT
Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)	VARIANT ABSENT
Alpha-1 Antitrypsin Deficiency	VARIANT ABSENT
Alpha-mannosidosis	VARIANT ABSENT
Autosomal recessive polycystic kidney disease	VARIANT ABSENT
Beta Thalassemia	VARIANT ABSENT
Biotinidase deficiency	VARIANT ABSENT
Birt-Hogg-Dube syndrome	VARIANT ABSENT
Canavan Disease	VARIANT ABSENT
Classical homocystinuria due to CBS deficiency	VARIANT ABSENT
Congenital disorder of glycosylation type 1a (PMM2-CDG)	VARIANT ABSENT
Congenital myasthenic syndrome	VARIANT ABSENT
Cystic Fibrosis	VARIANT ABSENT
Cystinosis	VARIANT ABSENT
D-Bifunctional Protein Deficiency	VARIANT ABSENT
Diastophic dysplasia	VARIANT ABSENT
Dihydrolipoamide Dehydrogenase Deficiency	VARIANT ABSENT
Ehlers-Danlos Syndrome (EDS)	VARIANT ABSENT

Inherited Conditions General Report

NAME	STATUS
Familial Advanced/Delayed Sleep-Phase Syndrome	VARIANT ABSENT
Familial Hypercholesterolemia Type B	VARIANT ABSENT
Familial Hypertrophic Cardiomyopathy (HCM)	VARIANT ABSENT
Familial Mediterranean fever	VARIANT ABSENT
Familial TTR-related amyloidosis	VARIANT ABSENT
Familial Transthyretin Amyloidosis	VARIANT ABSENT
Familial adenomatous polyposis	VARIANT ABSENT
Familial dysautonomia (Riley-Day syndrome)	VARIANT ABSENT
Fanconi Anemia (FANCC-related)	VARIANT ABSENT
GRACILE syndrome	VARIANT ABSENT
Gaucher disease	VARIANT ABSENT
Glucose-6-phosphate dehydrogenase deficiency(G6PD deficiency)	VARIANT ABSENT
Glycogen storage disease type 1A (Von Gierke Disease)	VARIANT ABSENT
Glycogen storage disease type 1B	VARIANT ABSENT
Glycogen storage disease type 2 or Pompe Disease 1 & 2	VARIANT ABSENT
Glycogen storage disease type 3	VARIANT ABSENT
Glycogen storage disease type 5	VARIANT ABSENT
Hereditary fructose intolerance	VARIANT ABSENT
Hereditary hemochromatosis associated with HFE	VARIANT ABSENT
Hypokalemic Periodic Paralysis	VARIANT ABSENT

Inherited Conditions General Report

NAME	STATUS
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
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
Neuronal Ceroid-Lipofuscinoses type 1 (associated to PPT1)	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

Inherited Conditions General Report

NAME	STATUS
Pendred 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
Refsum disease	VARIANT ABSENT
Rhizomelic Chondrodysplasia Punctata Type 1	VARIANT ABSENT
Salla Disease	VARIANT ABSENT
Sjögren-Larsson syndrome	VARIANT ABSENT
Spinal muscular atrophy	VARIANT ABSENT
Tay-Sachs disease	VARIANT ABSENT
Tyrosinemia type I	VARIANT ABSENT
Usher syndrome	VARIANT ABSENT
Wilson disease	VARIANT ABSENT
Zellweger syndrome	VARIANT ABSENT
Homocystinuria, responsive to pyridoxine (vitamin B6)	
Neuronal Ceroid-Lipofuscinoses type 3 (associated to CLN3)	
Peters plus syndrome	