Unilab Carrier Test

ACMG Tiers 1-3

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GENE	DISORDER	ACMG TIER
CFTR	Cystic fibrosis	Tier 1
FMR1	Fragile X syndrome	Tier 1
SMN1	Spinal muscular atrophy	Tier 1
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	Tier 2
DHCR7	Smith-Lemli-Opitz syndrome	Tier 2
HBB	Beta-hemoglobinopathies (HbS, HbC, HbD, HbE, HbO)	Tier 2
HEXA	Tay-Sachs disease	Tier 2
PAH	Phenylketonuria (Phenylalanine hydroxylase deficiency)	Tier 2
PMM2	Carbohydrate-deficient glycoprotein syndrome type Ia (Congenital disorder of glycosylation type IA)	Tier 2
ASPA	Canavan disease	Tier 2
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Tier 2
SMPD1	Niemann-Pick disease, type A	Tier 2
SMPD1	Niemann-Pick disease, type B	Tier 2
G6PC	Glycogen storage disease type IA	Tier 2
ABCC8	Diabetes mellitus, permanent neonatal 3	Tier 2
ALDOB	Hereditary fructosuria	Tier 2
ATP7B	Wilson disease	Tier 2
BCKDHB	Maple syrup urine disease	Tier 2
CEP290	Joubert syndrome 5	Tier 2

GENE	DISORDER	ACMG TIER
CEP290	Leber congenital amaurosis 10	Tier 2
CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel	Tier 2
CHRNE	Myasthenic syndrome, congenital, 4B, fast-channel	Tier 2
COL7A1	Recessive dystrophic epidermolysis bullosa	Tier 2
CPT2	Carnitine palmitoyltransferase II deficiency, infantile	Tier 2
CPT2	Carnitine palmitoyltransferase II deficiency, lethal neonatal	Tier 2
ERCC2	Cerebrooculofacioskeletal syndrome 2	Tier 2
ERCC2	Trichothiodystrophy 1, photosensitive	Tier 2
FANCC	Fanconi anemia, complementation group C	Tier 2
FKTN	Cardiomyopathy, dilated, 1X	Tier 2
FKTN	Walker-Warburg congenital muscular distrophy	Tier 2
GAA	Glycogen sotrage disease, type II	Tier 2
GBE1	Glycogen storage disease, type IV	Tier 2
GBE1	GBE-1 related disorders	Tier 2
GJB2	Nonsyndrmic hearing loss recessive 1A	Tier 2
GJB2	Nonsyndrmic hearing loss recessive 3A	Tier 2
MMUT	Mythylmalonic acidura-methylmalonly-CoA mutase deficiency	Tier 2
NPHS1	Finnish congenital nephrotic syndrome	Tier 2
OCA2	Oculocutaneous albinism brown and type II	Tier 2
SLC26A4	Deafness autosomal recessive 4	Tier 2

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GENE	DISORDER	ACMG TIER
SLC26A4	Pendred syndrome	Tier 2
TYR	Oculocutaneous albinism type 1A and 1B	Tier 2
USH2A	Usher syndrome, type 2A	Tier 2
XPC	Xeroderma pigmentosum	Tier 2
HBA1/HBA2	Alpha-thalessemia	Tier 3
ELP1	Familial dysautonomia	Tier 3
BLM	Bloom syndrome	Tier 3
DMD	Duchenne muscular dystrophy	Tier 3
GALT	Galactosemia	Tier 3
GBA	Gaucher disease	Tier 3
ABCA3	Surfactant metabolism dysfunction, pulmonary 3	Tier 3
ABCD1	Adrenoleukodystrophy (ALD)	Tier 3
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency	Tier 3
ACAT1	α-Methylacetoacetic aciduria	Tier 3
AGA	Aspartylglucosaminuria	Tier 3
AGXT	Hyperoxaluria, primary type I	Tier 3
AIRE	Autoimmune polyendocrinopathy syndrome type I	Tier 3
ALPL	Hypophosphatasia, adult	Tier 3
ALPL	Hypophosphatasia, childhood and infantile	Tier 3
ARSA	Metachromatic leukodystrophy	Tier 3

GENE	DISORDER	ACMG TIER
ASL	Argininosuccinate acidura	Tier 3
BBS1	Bardet-Biedl syndrome 1	Tier 3
BBS2	Bardet-Biedle syndrome 2	Tier 3
BBS2	Retinitis pigmentosa 74	Tier 3
BDT	Biotinidase deficiency	Tier 3
CBS	Homocystinuria, B6 responsive and nonresponsive	Tier 3
CC2D2A	Joubert syndrome 9	Tier 3
CC2D2A	Meckel syndrome 6	Tier 3
CCDC88C	Congenital hydrocephalus 1	Tier 3
CLCN1	Congenital myotonia, autosomal recessive form	Tier 3
CLRN1	Usher syndrome 3a	Tier 3
CNGB3	Achromatopsia 3	Tier 3
CYP11A1	Adrenal insufficiency, congenital, with 46, XY sex reversal, partial or complete	Tier 3
CYP27A1	Cerebrotendinous xanthomatosis	Tier 3
CYP27B1	Vitamin D-dependent rickets, type 1	Tier 3
DHDDS	Congenital disorder of glycosylation type 1	Tier 3
DHDDS	Retinitis pigmentosa 59	Tier 3
DLD	Dihyrdolipomide dehydroxenase deficiency	Tier 3
EVC2	Chondroectodermal dysplasia	Tier 3
F8	Hemophilia A (HEMA)	Tier 3

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GENE	DISORDER	ACMG TIER
F9	Hemophilia B (HEMB)	Tier 3
FAH	Tyrosinemia type I	Tier 3
FKRP	Muscular dystrophy-dystroglycanopathy, type A, 5	Tier 3
FKRP	Muscular dystrophy-dystroglycanopathy, type B, 5	Tier 3
GLA	Fabry disease	Tier 3
GNPTAB	Mucolipidosis type II alpha/beta	Tier 3
GNPTAB	Mucolipidosis type III alpha/beta	Tier 3
HPS1	Hermansky Pudlak S. 1	Tier 3
HPS3	Hermansky Pudlak S. 3	Tier 3
IDUA	Mucopolysaccharidosis, Ih (Hurler S)	Tier 3
IDUA	Mucopolysaccharidosis, Ih/s (Hurler-Scheie S)	Tier 3
MCC2	3-methylcrotonyl CoA carboxylase 2 deficiency	Tier 3
MCOLN1	Mucolipidosis type IV	Tier 3
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	Tier 3
MMACHC	Methylmalonic aciduria with homocytinuria cblC type	Tier 3
NEB	Nemaline myopathy 2	Tier 3
NROB1	Adrenal hypoplasia, congenital (AHC)	Tier 3
OTC	Ornithine transcarbamylase deficiency	Tier 3
PCDH15	Deafness, autosomal recessive 23	Tier 3
PCDH15	Usher syndrome, type IF	Tier 3

GENE	DISORDER	ACMG TIER
PKHD1	Autosomal recessive polycystic kidney disease	Tier 3
POLG	Mitochondrial depletion syndrome 4A	Tier 3
POLG	Mitochondrial depletion syndrome 4B	Tier 3
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	Tier 3
RARS2	Pontcocerebellar hypoplasia type 6	Tier 3
RS1	Retinoschisis 1, X-linked, juvenile RS1)	Tier 3
SLC26A2	Epiphyseal dysplasia, multiple, 4	Tier 3
SLC26A2	Achondrogenesis Ib	Tier 3
SLC37A4	Glycogen storage disease Ib	Tier 3
SLC37A4	Glycogen storage disease Ic	Tier 3
SLC6A8	Cerebral creatine deficiency syndrome 1 (CCDS1)	Tier 3
TMEM216	Joubert syndrome 2	Tier 3
TMEM216	Meckel syndrome 2	Tier 3