Virtus Diagnostics Preconception Screen

What diseases does it screen your carrier status for?

Disease	Symbol
17-@BETA HYDROXYSTEROID DEHYDROGENASE III DEFICIENCY	HSD17B3
3-@METHYLGLUTACONIC ACIDURIA, TYPE III	OPA3
3-BETA-HYDROXYSTEROID DEHYDROGENASE DEFICIENCY TYPE II	HSD3B2
3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY	HMGCL
3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY	HADH
3-METHYLGLUTACONIC ACIDURIA, TYPE I	AUH
3-METHYLGLUTACONIC ACIDURIA, TYPE V	DNAJC19
ABCD SYNDROME	EDNRB
ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME	AAAS
ACHONDROGENESIS, TYPE IB ACG1B	SLC26A2
ACYL-CoA DEHYDROGENASE, LONG-CHAIN, DEFICIENCY OF	ACADL
ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN,	ACADM
ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN,	ACADVL
ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY	CYP21A2
ADRENOLEUKODYSTROPHY	ABCD1
ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM	PEX1
ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM |	PEX10
ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM |	PEX13
ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM |	PEX26
ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM |	PEX5
AGAMMAGLOBULINEMIA, X-LINKED	BTK
AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY ACCPN	SLC12A6
AICARDI-GOUTIERES SYNDROME 1	TREX1
AICARDI-GOUTIERES SYNDROME 2	RNASEH2B
AICARDI-GOUTIERES SYNDROME 3	RNASEH2C
AICARDI-GOUTIERES SYNDROME 4	RNASEH2A
AICARDI-GOUTIERES SYNDROME 5	SAMDH1
ALLAN-HERNDON-DUDLEY SYNDROME AHDS	SLC16A2
ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS	POLG
ALPHA THALASSEMIA	HBA1
ALPHA-METHYLACETOACETIC ACIDURIA	ACAT1
ALPORT SYNDROME, AUTOSOMAL RECESSIVE	COL4A3
ALPORT SYNDROME, AUTOSOMAL RECESSIVE	COL4A4
ALPORT SYNDROME, X-LINKED	Col4A5
ALSTROM SYNDROME	ALMS1
AMEGAKARYOCYTIC THROMBOCYTOPENIA, CONGENITAL	MPL
AMISH INFANTILE EPILEPSY SYNDROME	ST3GAL5
ANAUXETIC DYSPLASIA	RMRP
ANHYDROTIC ECTODERMAL DYSPLASIA, IMMUNODEFICIENCY, OSTEOPETROSIS, LYMPHEDEMA OLEDAID	IKBKG

ANTIBODY DEFICIENCY DUE TO ICOS DEFECT	ICOS
ANTLEY-BIXLER SYNDROME	POR
ARGININOSUCCINIC ACIDURIA	ASL
AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY	DDC
ARTERIAL CALCIFICATION, GENERALIZED, OF INFANCY	ENPP1
ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS	VPS33B
ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS 2	VIPAR
ARTS SYNDROME	PRPS1
ASPHYXIATING THORACIC DYSTROPHY 2	IFT80
ASPHYXIATING THORACIC DYSTROPHY 3	DYNC2H1
ATAXIA, EARLY-ONSET, WITH oculomotor apraxia AND HYPOALBUMINEMIA	APTX
ATAXIA-TELANGIECTASIA	ATM
ATELOSTEOGENESIS, TYPE II	SLC26A2
a-THALASSEMIA/MENTAL RETARDATION SYNDROME,NONDELETION TYPE, X-LINKED	ATRX
ATYPICAL MYCOBACTERIOSIS, FAMILIAL	IFNGR1
ATYPICAL MYCOBACTERIOSIS, FAMILIAL	IFNGR2
ATYPICAL MYCOBACTERIOSIS, FAMILIAL	IKBKG
ATYPICAL MYCOBACTERIOSIS, FAMILIAL	IL12B
ATYPICAL MYCOBACTERIOSIS, FAMILIAL	IL12RB1
ATYPICAL MYCOBACTERIOSIS, FAMILIAL	STAT1
ATYPICAL MYCOBACTERIOSIS, FAMILIAL	TYK2
AUTOIMMUNE POLYENDOCRINE SYNDROME TYPE I	AIRE
Autosomal mental retardation	NSUN2
Autosomal mental retardation	ST3GAL3
Autosomal mental retardation CDG 1Q	SRD5A3
BARTH SYNDROME	TAZ
BARTTER SYNDROME, ANTENATAL, TYPE 1	SLC12A1
BARTTER SYNDROME, ANTENATAL, TYPE 2	KCNJ1
BETA-HYDROXYISOBUTYRYL CoA DEACYLASE, DEFICIENCY OF	HIBCH
BILE ACID SYNTHESIS DEFECT, CONGENITAL, 4	AMACR
BIOTINIDASE DEFICIENCY	BTD
BLOOM SYNDROME	BLM
BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, BETA POLYPEPTIDE	BCKDHB
BRITTLE CORNEA SYNDROME (Ehlers-Danlos syndrome type VIB)	ZNF469
CANAVAN DISEASE	ASPA
CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY, HYPERAMMONEMIA DUE TO	CPS1
CARDIOENCEPHALOMYOPATHY, FATAL INFANTILE, DUE TO CYTOCHROME c OXIDASE	SCO2
CARDIOMYOPATHY, DILATED, 3A	TAZ
CARNITINE DEFICIENCY, SYSTEMIC PRIMARY	SLC22A5
CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY	CPT1A
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, INFANTILE	CPT2
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LATE-ONSET	CPT2

CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LETHAL NEONATAL	CPT2
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	SLC25A20
CARPENTER SYNDROME	RAB23
CARTILAGE-HAIR HYPOPLASIA	RMRP
CEREBELLAR HYPOPLASIA AND MENTAL RETARDATION WITH OR WITHOUT QUADRUPEDAL	VLDLR
CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA	SNAP29
CEREBROOCULOFACIOSKELETAL SYNDROME 1	ERCC6
CEREBROTENDINOUS XANTHOMATOSIS	CYP27A1
CEROID LIPOFUSCINOSIS, NEURONAL, 10	CTSD
CEROID LIPOFUSCINOSIS, NEURONAL, 6	CLN6
CEROID LIPOFUSCINOSIS, NEURONAL, 7	MFSD8
CEROID LIPOFUSCINOSIS, NEURONAL, 8	CLN8
CEROID LIPOFUSCINOSIS, NEURONAL, 8, NORTHERN EPILEPSY VARIANT	CLN8
CHARCOT-MARIE-TOOTH DISEASE TYPE 4A	GDAP1
CHARCOT-MARIE-TOOTH DISEASE, TYPE 4H	FGD4
CHEDIAK HIGASHI SYNDROME	LYST
CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC	ABCB4
CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 1	ATP8B1
CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 2	ABCB11
CHONDRODYSPLASIA, BLOMSTRAND TYPE	PTH1R
CITRULLINEMIA, CLASSIC	ASS1
COCKAYNE SYNDROME TYPE B	ERCC6
COCKAYNE SYNDROME, TYPE A	ERCC8
COENZYME Q10 DEFICIENCY	PDSS2
COENZYME Q10 DEFICIENCY	APTX
COENZYME Q10 DEFICIENCY	CABC1
COENZYME Q10 DEFICIENCY	COQ2
COENZYME Q10 DEFICIENCY	COQ9
COENZYME Q10 DEFICIENCY	PDSS1
COFFIN-LOWRY SYNDROME CLS	RPS6KA3
COHEN SYNDROME	VPS13B
COMBINED IMMUNODEFICIENCY, X-LINKED	IL2RG
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1	GFM1
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 2	MRPS16
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3	TSFM
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 4	TUFM
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 5	MRPS22
COMBINED SAPOSIN DEFICIENCY	PSAP
Complex I Deficiency	NDUFA7
Complex I Deficiency	NDUFA1

Complex I Deficiency	NDUFAF2
Complex I Deficiency	NDUFAF4
Complex I Deficiency	NDUFS3
Complex I Deficiency	NDUFS4
Complex I Deficiency	NDUFS5
Complex I Deficiency	NDUFS6
Complex I Deficiency	NDUFS7
Complex I Deficiency	NDUFS8
Complex I Deficiency	NDUFV1
Complex IV deficiency	COX10
Complex IV deficiency	COX15
Complex IV deficiency	COX6B1
Complex IV deficiency	FASTKD2
Complex IV deficiency	sco1
Complex IV deficiency	sco2
CONGENITAL ADRENAL HYPERPLASIA	CYP11B1
CONGENITAL ADRENAL HYPERPLASIA	CYP17A1
CONGENITAL ADRENAL HYPOPLASIA	NR0B1
CONGENITAL DEAFNESS WITH KERATOPACHYDERMIA & CONSTRICTIONS OF FINGERS & TOES	GJB2
CONGENITAL DISORDER OF GLYCOSYLATIO, TYPE IIa	MGAT2
CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ia	PMM2
CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ic	ALG6
CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ig	ALG12
CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ii	ALG2
CONGENITAL DISORDER OF GLYCOSYLATION TYPE IIb	MOGS
CONGENITAL DISORDER OF GLYCOSYLATION TYPE IIc	SLC35C1
CONGENITAL DISORDER OF GLYCOSYLATION TYPE IId	B4GALT1
CONGENITAL DISORDER OF GLYCOSYLATION TYPE Iig	COG1
CONGENITAL DISORDER OF GLYCOSYLATION TYPE Iih	COG8
CONGENITAL DISORDER OF GLYCOSYLATION TYPE Im	DOLK
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ib	MPI
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Id;	ALG3
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ie	DPM1
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE If	MPDU1
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ih	ALG8
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Iie	COG7
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Iif	SLC35A1
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ij	DPAGT1
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ik	ALG1
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Il	ALG9
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE In	RFT1

CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS	SLC4A11
CORPUS CALLOSUM, AGENESIS OF, WITH ABNORMAL GENITALIA	ARX
CORPUS CALLOSUM, PARTIAL AGENESIS OF, X-LINKED	L1CAM
CORTISOL 11-BETA-KETOREDUCTASE DEFICIENCY	HSD11B2
CRANIOFRONTONASAL SYNDROME	EFNB1
CREATINE DEFICIENCY SYNDROME, X-LINKED	SLC6A8
CRISPONI SYNDROME	CRLF1
CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE I	EFEMP2
CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE I	FBLN5
CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE II	ATP6V0A2
CYSTIC FIBROSIS	CFTR
CYSTINOSIS, ADULT NONNEPHROPATHIC	CTNS
CYSTINOSIS, LATE-ONSET JUVENILE OR ADOLESCENT NEPHROPATHIC TYPE	CTNS
CYSTINOSIS, NEPHROPATHIC	CTNS
D-BIFUNCTIONAL PROTEIN DEFICIENCY	HSD17B4
DE SANCTIS-CACCHIONE SYNDROME	ERCC6
DE SANCTIS-CACCHIONE SYNDROME	XPA
DEFICIENCY OF ACYL-CoA DEHYDROGENASE FAMILY MEMBER 9	ACAD9
DEFICIENCY OF INTERLEUKIN 1 RECEPTOR ANTAGONIST	IL1RN
DENT DISEASE 1	CLCN5
DESMOSTEROLOSIS	DHCR24
DIARRHEA 4, MALABSORPTIVE, CONGENITAL	NEUROG3
DIASTROPHIC DYSPLASIA	SLC26A2
DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY	DLD
DIHYDROPYRIMIDINE DEHYDROGENASE	DPYD
DISORDERED STEROIDOGENESIS	POR
DONNAI-BARROW SYNDROME	LRP2
DONOHUE SYNDROME	INSR
DYSSEGMENTAL DYSPLASIA, SILVERMAN-HANDMAKER TYPE	HSPG2
ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH IMMUNE DEFICIENCY	IKBKG
ECTODERMAL DYSPLASIA, HYPOHIDROTIC, X-LINKED	EDA
ELEJALDE DISEASE	MYO5A
ELLIS-VAN CREVELD SYNDROME	EVC2
ELLIS-VAN CREVELD SYNDROME; EVC	EVC
ENCEPHALOPATHY, ETHYLMALONIC	ETHE1
EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL RECESSIVE	COL7A1
EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA	ITGA6
EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA	ITGB4
EPIDERMOLYSIS BULLOSA SIMPLEX WITH MUSCULAR DYSTROPHY	PLEC1
EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE	LAMA3
EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE	LAMB3
EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE	LAMC2

EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE	COL17A1
EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE	ITGB4
EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE	LAMA3
EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE	LAMB3
EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE	LAMC2
EPIDERMOLYSIS BULLOSA, LETHAL ACANTHOLYTIC	DSP
EPILEPSY, FEMALE-RESTRICTED, WITH MENTAL RETARDATION	PCDH19
EPILEPSY, PYRIDOXINE-DEPENDENT;	ALDH7A1
EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 1	ARX
EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 3	SLC25A22
EPIPHYSEAL DYSPLASIA, MULTIPLE, WITH EARLY-ONSET DIABETES MELLITUS	EIF2AK3
FABRY DISEASE	GLA
FACIOGENITAL DYSPLASIA	FGD1
FAMILIAL MEDITERRANEAN FEVER	MEFV
Fanconi anemia type C	FANCC
FETAL AKINESIA DEFORMATION SEQUENCE	RAPSN
FIBROMATOSIS, JUVENILE HYALINE	ANTXR2
FIBULAR APLASIA OR HYPOPLASIA, FEMORAL BOWING AND POLY-, SYN-, AND	WNT7A
FRASER SYNDROME	FRAS1
FRASER SYNDROME	FREM2
FRUCTOSE INTOLERANCE, HEREDITARY	ALDOB
FUCOSIDOSIS	FUCA1
FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY	FKTN
FUMARASE DEFICIENCY	FH
GALACTOKINASE DEFICIENCY	GALK1
GALACTOSEMIA	GALT
GAUCHER DISEASE	GBA
GELEOPHYSIC DYSPLASIA	ADAMTSL2
GLUCOSE-6-PHOSPHATE DEHYDROGENASE	G6PD
GLUTARIC ACIDEMIA I	GCDH
GLUTATHIONE SYNTHETASE DEFICIENCY	GSS
GLYCINE ENCEPHALOPATHY	AMT
GLYCINE ENCEPHALOPATHY	GCSH
GLYCINE ENCEPHALOPATHY	GLDC
GLYCOGEN STORAGE DISEASE I VON GIERKE DISEASE	G6PC3
GLYCOGEN STORAGE DISEASE Ib	SLC37A4
GLYCOGEN STORAGE DISEASE Ic	SLC37A4
GLYCOGEN STORAGE DISEASE II (pompe)	GAA
GLYCOGEN STORAGE DISEASE III	AGL
GLYCOGEN STORAGE DISEASE IV	GBE1
GM1-GANGLIOSIDOSIS TYPE II	GLB1
GM1-GANGLIOSIDOSIS, TYPE I	GLB1

GONADAL DYSGENESIS WITH ADRENAL FAILURE	NR5A1
GRACILE SYNDROME	BCS1L
GRISCELLI SYNDROME, TYPE 1	MYO5A
GRISCELLI SYNDROME, TYPE 2	RAB27A
GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY	GAMT
HEMOCHROMATOSIS, JUVENILE, TYPE 2A	HJV
HEMOCHROMATOSIS, JUVENILE, TYPE 2B	HAMP
HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 2	PRF1
HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 3	UNC13D
HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 4	STX11
HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 5	STXBP2
HEMOPHILIA A; HEMA	F8
HEMOPHILIA B; HEMB	F9
HEPATIC VENOOCCLUSIVE DISEASE WITH IMMUNODEFICIENCY VODI	SP110
Hermansky Pudlak Syndrome, 9	PLDN
HERMANSKY-PUDLAK SYNDROME 2	AP3B1
HERPES SIMPLEX ENCEPHALITIS, SUSCEPTIBILITY TO, 1	UNC93B1
HERPES SIMPLEX ENCEPHALITIS, SUSCEPTIBILITY TO, 2	TLR3
HETEROTAXY, VISCERAL, 1, X-LINKED	ZIC3
HOLOCARBOXYLASE SYNTHETASE DEFICIENCY	HLCS
HOMOCYSTINURIA	CBS
HONDRODYSPLASIA PUNCTATA 1, X-LINKED RECESSIVE	ARSE
HOYERAAL-HREIDARSSON SYNDROME	DKC1
HURLER SYNDROME	IDUA
HYALINOSIS, INFANTILE SYSTEMIC	ANTXR2
HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS	L1CAM
HYDROPS-ECTOPIC CALCIFICATION-MOTH-EATEN SKELETAL DYSPLASIA	LBR
HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE DEF.	HADHA
HYPEREKPLEXIA AND EPILEPSY	ARHGEF9
HYPER-IgE RECURRENT INFECTION SYNDROME, AUTOSOMAL RECESSIVE	DOCK8
HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, 1	ABCC8
HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME	SLC25A15
HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS	PMP22
HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS CMT3,	EGR2
HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS CMT3,	MPZ
HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS CMT3,	PRX
HYPOGONADOTROPIC HYPOGONADISM	GNRHR
HYPOMAGNESEMIA, RENAL, WITH OCULAR INVOLVEMENT	CLDN19
HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME	TBCE
HYPOPHOSPHATASIA, CHILDHOOD	ALPL
HYPOPHOSPHATEMIC osteopenia, AUTOSOMAL RECESSIVE	DMP1

HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 4	TSHB
ICHTHYOSIS CONGENITA, HARLEQUIN FETUS TYPE	
ICHTHYOSIS FOLLICULARIS, ATRICHIA, AND PHOTOPHOBIA SYNDROME	MBTPS2
ICHTHYOSIS, LAMELLAR, 1	TGM1
ICHTHYOSIS, LEUKOCYTE VACUOLES, ALOPECIA, AND SCLEROSING CHOLANGITIS	CLDN1
IMMUNE DYSFUNCTION WITH T-CELL INACTIVATION DUE TO CALCIUM ENTRY DEFECT 1 ORAI1	ORAI1
IMMUNE DYSFUNCTION WITH T-CELL INACTIVATION DUE TO CALCIUM ENTRY DEFECT 2 STIM1	STIM1
IMMUNODEFICIENCY DUE TO DEFECT IN CD3-EPSILON	CD3E
IMMUNODEFICIENCY DUE TO DEFECT IN CD3-GAMMA	CD3G
IMMUNODEFICIENCY DUE TO DEFECT IN CD3-ZETA	CD3Z
IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 1	CD40LG
IMMUNODEFICIENCY, COMMON VARIABLE, 3	CD19
IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME	DNMT3B
IMMUNODYSREGULATION, POLYENDOCRINOPATHY, AND ENTEROPATHY, X-LINKED	FOXP3
INFANTILE NEUROAXONAL DYSTROPHY	PLA2G6
INFANTILE SIALIC ACID STORAGE DISORDER	SLC17A5
INFANTILE-ONSET SPINOCEREBELLAR ATAXIA	C10ORF2
INFERTILE MALE SYNDROME	AR
INSENSITIVITY TO PAIN, CONGENITAL, WITH ANHIDROSIS	NTRK1
ISOVALERIC ACIDEMIA	IVD
JOHANSON-BLIZZARD SYNDROME	UBR1
JOUBERT SYNDROME 3	AHI1
JOUBERT SYNDROME 4	NPHP1
JOUBERT SYNDROME 5	CEP290
JOUBERT SYNDROME 6	TMEM67
JUVENILE AMYOTROPHIC LATERAL SCLEROSIS 2	ALS2
KRABBE DISEASE	GALC
LACTIC ACIDOSIS, FATAL INFANTILE (mtDNA depletion)	SUCLG1
LARYNGOONYCHOCUTANEOUS SYNDROME	LAMA3
LATHOSTEROLOSIS	SC5DL
LEIGH SYNDROME	SURF1
LEIGH SYNDROME, FRENCH-CANADIAN TYPE	LRPPRC
LEIGH SYNDROME, X-LINKED	PDHA1
LESCH-NYHAN SYNDROME	HPRT1
LETHAL CONGENITAL CONTRACTURE SYNDROME 1	GLE1
LETHAL CONGENITAL CONTRACTURE SYNDROME 2	ERBB3
LEUKOCYTE ADHESION DEFICIENCY TYPE III	FERMT3
LEUKODYSTROPHY, HYPOMYELINATING, 2	GJC2
LEUKODYSTROPHY, HYPOMYELINATING, 5	FAM126A
LIPOID CONGENITAL ADRENAL HYPERPLASIA	CYP11A1

LIPOID CONGENITAL ADRENAL HYPERPLASIA	STAR
LISSENCEPHALY 2	RELN
LISSENCEPHALY 3	TUBA1a
LISSENCEPHALY, X-LINKED, 1	DCX
LISSENCEPHALY, X-LINKED, 2	ARX
LOWE OCULOCEREBRORENAL SYNDROME	OCRL
LUJAN-FRYNS SYNDROME	MED12
LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 1	SH2D1A
LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 2	XIAP
MANDIBULOACRAL DYSPLASIA WITH TYPE B LIPODYSTROPHY	ZMPSTE24
MANNOSIDOSIS, ALPHA B, LYSOSOMAL	MAN2B1
MAPLE SYRUP URINE DISEASE Type Ia	BCKDHA
Marinesco-Sjogren Syndrome	SIL1
MARTSOLF SYNDROME	GAP2
MASA SYNDROME	L1CAM
MECKEL SYNDROME TYPE 1	MKS1
MECKEL SYNDROME, TYPE 5	RPGRIP1L
MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS	MLC1
MENKES DISEASE	ATP7A
MENTAL RETARDATION AND MICROCEPHALY WITH PONTINE AND CEREBELLAR HYPOPLASIA CASK	CASK
MENTAL RETARDATION AUTOSOMAL RECESSIVE 13	TRAPPC9
MENTAL RETARDATION AUTOSOMAL RECESSIVE 6	GRIK2
MENTAL RETARDATION AUTOSOMAL RECESSIVE 7	TUSC3
MENTAL RETARDATION X-LINKED 19 INCLUDED	RPS6KA3
MENTAL RETARDATION X-LINKED 72	RAB39B
MENTAL RETARDATION X-LINKED 88	AGTR2
MENTAL RETARDATION X-LINKED 89	ZNF41
MENTAL RETARDATION X-LINKED 90	DLG3
MENTAL RETARDATION X-LINKED 92	ZNF674
MENTAL RETARDATION X-LINKED ASSOCIATED WITH FRAGILE SITE	AFF2
MENTAL RETARDATION X-LINKED SYNDROMIC TURNER TYPE	HUWE1
MENTAL RETARDATION X-LINKED SYNDROMIC UBE2A-RELATED	UBE2A
MENTAL RETARDATION X-LINKED SYNDROMIC ZDHHC9-RELATED	ZDHHC9
MENTAL RETARDATION X-LINKED SYP-RELATED	SYP
MENTAL RETARDATION X-LINKED WITH BRACHYDACTYLY AND MACROGLOSSIA	CUL4B
MENTAL RETARDATION X-LINKED ZNF711-RELATED	ZNF711
MENTAL RETARDATION, AUTOSOMAL RECESSIVE 1	PRSS12
MENTAL RETARDATION, X-LINKED 21	IL1RAPL1
MENTAL RETARDATION, X-LINKED 30	PAK3
MENTAL RETARDATION, X-LINKED 41, 48 MRX41,	GDI1

MENTAL RETARDATION, X-LINKED 46	ARHGEF6
MENTAL RETARDATION, X-LINKED 59	AP1S2
MENTAL RETARDATION, X-LINKED 68	ACSL4
MENTAL RETARDATION, X-LINKED 9	FTSJ1
MENTAL RETARDATION, X-LINKED 93	BRWD3
MENTAL RETARDATION, X-LINKED ANGELMAN, SYNDROMIC, CHRISTIANSON	SLC9A6
MENTAL RETARDATION, X-LINKED, SNYDER-ROBINSON TYPE	SMS
MENTAL RETARDATION, X-LINKED, SYNDROMIC	KDM5C
MENTAL RETARDATION, X-LINKED, SYNDROMIC 10	HSD17B10
MENTAL RETARDATION, X-LINKED, SYNDROMIC 14	UPF3B
MENTAL RETARDATION, X-LINKED, WITH PANHYPOPITUITARISM	sox3
MENTAL RETARDATION, X-LINKED, WITH PANHYPOPITUITARISM	sox3
MENTAL RETARDATION, XLR, W CEREBELLAR HYPOPLASIA & DISTINCTIVE FACIAL APPEARANCE	OPHN1
METACHROMATIC LEUKODYSTROPHY	ARSA
METACHROMATIC LEUKODYSTROPHY DUE TO SAPOSIN B DEFICIENCY	PSAP
METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE	MMACHC
METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA MUTASE DEFICIENCY	MUT
METHYLMALONIC ACIDURIA, cblA TYPE	MMAA
METHYLMALONIC ACIDURIA, cblB TYPE	MMAB
MEVALONIC ACIDURIA	MVK
MICROPHTHALMIA, SYNDROMIC	BCOR
MICROPHTHALMIA, SYNDROMIC 9 (Matthew-Wood syndrome)	STRA6
MITOCHONDRIAL COMPLEX III DEFICIENCY	BCS1L
MITOCHONDRIAL COMPLEX III DEFICIENCY	UQCRB
MITOCHONDRIAL COMPLEX III DEFICIENCY	UQCRQ
MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM	C10ORF2
MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM	DGUOK
MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM	MPV17
MITOCHONDRIAL DNA DEPLETION SYNDROME, MYOPATHIC FORM	TK2
MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME	TYMP
MOLYBDENUM COFACTOR DEFICIENCY	MOCS2
MOLYBDENUM COFACTOR DEFICIENCY	MOCS1
MSUD type 2	DBT
mtDNA depletion, encephalomyopathic form	RRM2B
mtDNA depletion, encephalomyopathic form	SUCLA2
MUCOLIPIDOSIS II ALPHA/BETA	GNPTAB
MUCOLIPIDOSIS III ALPHA/BETA	GNPTAB
MUCOLIPIDOSIS IV	MCOLN1
MUCOPOLYSACCHARIDOSIS TYPE II	IDS
MUCOPOLYSACCHARIDOSIS TYPE IIIA (Sanfilippo type A)	SGSH
MUCOPOLYSACCHARIDOSIS TYPE IIIC (Sanfilippo type c)	HGSNAT

MUCOPOLYSACCHARIDOSIS TYPE VI MAROTEAUX-LAMY	ARSB
MUCOPOLYSACCHARIDOSIS TYPE VII SLY SYNDROME	GUSB
MULIBREY NANISM	TRIM37
MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	ETFA
MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	ETFB
MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	ETFDH
MULTIPLE PTERYGIUM SYNDROME, ESCOBAR VARIANT	CHRNG
MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE	CHRNA1
MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE	CHRND
MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE	CHRNG
MUSCLE-EYE-BRAIN DISEASE	FKRP
MUSCLE-EYE-BRAIN DISEASE	POMGNT1
MUSCULAR DYSTROPHY, CONGENITAL MEROSIN-DEFICIENT, 1A	LAMA2
MUSCULAR DYSTROPHY, CONGENITAL, 1C	FKRP
MUSCULAR DYSTROPHY, CONGENITAL, TYPE 1D	LARGE
MUSCULAR DYSTROPHY, DUCHENNE TYPE	DMD
MYD88 DEFICIENCY	MYD88D
MYOCLONIC EPILEPSY OF LAFORA	EPM2A
MYOCLONIC EPILEPSY OF LAFORA	NHLRC1
MYOCLONIC EPILEPSY OF UNVERRICHT AND LUNDBORG	CSTB
MYOTUBULAR MYOPATHY 1	MTM1
N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY	NAGS
NANCE-HORAN SYNDROME;	NHS
NAVAJO NEUROHEPATOPATHY	MPV17
NEMALINE MYOPATHY 2	NEB
NEPHRONOPHTHISIS 1	NPHP1
NEPHRONOPHTHISIS 2	INVS
NEPHRONOPHTHISIS 4	NPHP4
NEPHROSIS 1, CONGENITAL, FINNISH TYPE	NPHS1
nephrotic syndrome, STEROID-RESISTANT, AUTOSOMAL RECESSIVE	NPHS2
nephrotic syndrome, TYPE 3	PLCE1
NEURAMINIDASE DEFICIENCY	NEU1
NEURODEGENERATION DUE TO CEREBRAL FOLATE TRANSPORT DEFICIENCY	FOLR1
NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 1 (Hallervorden-Spatz)	PANK2
NEURONAL CEROID LIPOFUSCINOSIS 1	PPT1
NEURONAL CEROID LIPOFUSCINOSIS 2	TPP1
NEURONAL CEROID LIPOFUSCINOSIS 3	CLN3
NEURONAL CEROID LIPOFUSCINOSIS 5	CLN5
NEUROPATHY, CONGENITAL HYPOMYELINATING: CHARCOT-MARIE-TOOTH DISEASE, TYPE	EGR2
NEUROPATHY, CONGENITAL HYPOMYELINATING: CHARCOT-MARIE-TOOTH DISEASE, TYPE	MPZ

NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE III	IKBKAP
NEUTROPENIA, SEVERE CONGENITAL, AUTOSOMAL RECESSIVE 3	HAX1
NEVO SYNDROME EDS	PLOD1
NIEMANN-PICK DISEASE, TYPE	NPC2
NIEMANN-PICK DISEASE, TYPE A	SMPD1
NIEMANN-PICK DISEASE, TYPE B	SMPD1
NIEMANN-PICK DISEASE, TYPE C1	NPC1
NIJMEGEN BREAKAGE SYNDROME	NBN
NORRIE DISEASE	NDP
ODONTOONYCHODERMAL DYSPLASIA	WNT10A
OMENN SYNDROME	DCLRE1C
OMENN SYNDROME	RAG1
OMENN SYNDROME	RAG2
OPITZ GBBB SYNDROME, X-LINKED	MID1
OPTICOACOUSTIC NERVE ATROPHY WITH DEMENTIA	TIMM8A
ORNITHINE TRANSCARBAMYLASE DEFICIENCY, HYPERAMMONEMIA DUE TO	OTC
OSTEOGENESIS IMPERFECTA, TYPE IIB	CRTAP
OSTEOGENESIS IMPERFECTA, TYPE VIII	LEPRE1
OSTEOPETROSIS, AUTOSOMAL RECESSIVE 1	TCIRG1
OSTEOPETROSIS, AUTOSOMAL RECESSIVE 3	CA2
OSTEOPETROSIS, AUTOSOMAL RECESSIVE 4	CLCN7
OSTEOPETROSIS, AUTOSOMAL RECESSIVE 5	OSTM1
PAGET DISEASE, JUVENILE	TNFRSF11B
PELIZAEUS-MERZBACHER DISEASE	PLP1
PEROXISOMAL ACYL-CoA OXIDASE DEFICIENCY	ACOX1
PHENYLKETONURIA	PAH
PIERSON SYNDROME	LAMB2
PITUITARY DWARFISM III	HESX1
PITUITARY DWARFISM III	LHX3
PITUITARY DWARFISM III	POU1F1
PITUITARY DWARFISM III	PROP1
PITUITARY HORMONE DEFICIENCY, COMBINED, 3;	LHX3
PLASMINOGEN DEFICIENCY TYPE I	PLG
POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE	PKHD1
PONTOCEREBELLAR HYPOPLASIA TYPE 2A	TSEN54
PONTOCEREBELLAR HYPOPLASIA TYPE 4	TSEN54
PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS
PRECOCIOUS PUBERTY, MALE-LIMITED	LHCGR
PRIMARY LATERAL SCLEROSIS, JUVENILE	ALS2
PROPERDIN DEFICIENCY, X-LINKED	CFP
PROPIONIC ACIDEMIA	PCCB
PROPIONIC ACIDEMIA	PCCA

PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE	SCNN1A
PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE	SCNN1B
PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE	SCNN1G
PSEUDOVAGINAL PERINEOSCROTAL HYPOSPADIAS;	SRD5A2
PYCNODYSOSTOSIS	CTSK
PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE DEFICIENCY	PNPO
PYRUVATE CARBOXYLASE DEFICIENCY	PC
PYRUVATE DEHYDROGENASE E3-BINDING PROTEIN DEFICIENCY	PDHX
PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY	PDP1
PYRUVATE KINASE DEFICIENCY OF RED CELLS	PKLR
RAINE SYNDROME	FAM20C
RENAL-HEPATIC-PANCREATIC DYSPLASIA	NPHP3
RENPENNING SYNDROME 1	PQBP1
RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS	SFTPB
RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS	SFTPC
RETT SYNDROME	MECP2
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE 1	PEX7
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3	AGPS
RIGID SPINE MUSCULAR DYSTROPHY 1	SEPN1
ROBERTS SYNDROME	ESCO2
SANDHOFF DISEASE	HEXB
SCHNECKENBECKEN DYSPLASIA	SLC35D1
SCID W MICROCEPHALY, GROWTH RETARDATION, & SENS TO IONIZING RADIATION	NHEJ1
SCID, AUT REC, T CELL-NEGATIVE, B CELL+, NK CELL+	CD3D
SECKEL SYNDROME 1	ATR
SEGAWA SYNDROME, AUTOSOMAL RECESSIVE	TH
SENIOR-LOKEN SYNDROME 1	NPHP1
SENIOR-LOKEN SYNDROME 5	IQCB1
SEVERE COMBINED IMMUNODEFICIENCY WITH SENSITIVITY TO IONIZING RADIATION	LIG4
SEVERE COMBINED IMMUNODEFICIENCY,	ADA
SEVERE COMBINED IMMUNODEFICIENCY, AUT REC, T CELL$^-$, B CELL+, NK CELL$^-$	JAK3
SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE,	RAG1
SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE,	RAG2
SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED	IL2RG
SHWACHMAN-DIAMOND SYNDROME	SBDS
SIALURIA, FINNISH TYPE	SLC17A5
SICKLE CELL ANEMIA	HBB
SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 1	GPC3
SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 2	OFD1
SJOGREN-LARSSON SYNDROME	ALDH3A2
SKIN FRAGILITY-WOOLLY HAIR SYNDROME	DSP
SMITH-LEMLI-OPITZ SYNDROME	DHCR7

SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE	SACS
SPASTIC PARAPLEGIA 2, X-LINKED	PLP1
SPINAL MUSCULAR ATROPHY TYPE I	SMN1
SPINAL MUSCULAR ATROPHY TYPE II	SMN1
SPINAL MUSCULAR ATROPHY TYPE III	SMN1
SPINAL MUSCULAR ATROPHY, DISTAL, AUTOSOMAL RECESSIVE, 1	IGHMBP2
SPINAL MUSCULAR ATROPHY, DISTAL, AUTOSOMAL RECESSIVE, 4	PLEKHG5
SPINAL MUSCULAR ATROPHY, X-LINKED 2	UBA1
SPONDYLOCOSTAL DYSOSTOSIS, AUTOSOMAL RECESSIVE 1	WNT7A
STOCCO DOS SANTOS X-LINKED MENTAL RETARDATION SYNDROME	SHROOM4
STRIATONIGRAL DEGENERATION, INFANTILE	NUP62
STUVE-WIEDEMANN SYNDROME	LIFR
SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY	ALDH5A1
SUDDEN INFANT DEATH WITH DYSGENESIS OF THE TESTES SYNDROME	TSPYL1
SULFOCYSTEINURIA	SUOX
SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 1	SFTPB
SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 3	ABCA3
TAY-SACHS DISEASE	HEXA
T-CELL IMMUNODEFICIENCY, CONGENITAL ALOPECIA, AND NAIL DYSTROPHY	FOXN1
TETRA-AMELIA, AUTOSOMAL RECESSIVE	WNT3
THALASSEMIA MAJOR	HBB
THROMBOTIC THROMBOCYTOPENIC PURPURA,	ADAMTS13
TIGHT SKIN CONTRACTURE SYNDROME, LETHAL	ZMPSTE24
TRICHOTHIODYSTROPHY, PHOTOSENSITIVE	ERCC2
TRICHOTHIODYSTROPHY, PHOTOSENSITIVE	ERCC3
TRICHOTHIODYSTROPHY, PHOTOSENSITIVE	GTF2H5
TRIFUNCTIONAL PROTEIN DEFICIENCY	HADHA
TRIFUNCTIONAL PROTEIN DEFICIENCY	HADHB
TYROSINEMIA, TYPE I	FAH
ULNA AND FIBULA, ABSENCE OF, WITH SEVERE LIMB DEFICIENCY	WNT7A
Usher syndrome type 3A	CLRN1
USHER SYNDROME, TYPE I	MYO7A
USHER SYNDROME, TYPE IC	USH1C
USHER SYNDROME, TYPE ID	CDH23
USHER SYNDROME, TYPE IG	USH1G
USHER SYNDROME, TYPE IIA	USH2A
USHER SYNDROME, TYPE IIC	GPR98
VITAMIN D-DEPENDENT osteopenia, TYPE I	CYP27B1
VITAMIN D-DEPENDENT osteopenia, TYPE II	VDR
VITAMIN E, FAMILIAL ISOLATED DEFICIENCY OF	TTPA
WAARDENBURG-SHAH SYNDROME	EDN3
WAARDENBURG-SHAH SYNDROME	EDNRB

WALKER-WARBURG SYNDROME	POMT1
WALKER-WARBURG SYNDROME	POMT2
WARBURG MICRO SYNDROME	RAB3GAP1
WILSON DISEASE	ATP7B
WISKOTT-ALDRICH SYNDROME	WAS
XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP A	XPA
XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP B	ERCC3
XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP C	XPC
XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP D	ERCC2
XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP E	DDB2
XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP F	ERCC4
XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP G	ERCC5
X-linked Asperger syndrome-2	NLGN4
X-linked mental retardation	NXF5
X-linked mental retardation	RPL10
ZELLWEGER SYNDROME	PEX12
ZELLWEGER SYNDROME ZS	PEX1