LISTADO DE ENFERMEDADES ANALIZADAS

11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	CYP11B1
17-Alpha-Hydroxylase Deficiency	CYP17A1
17-Beta-Hydroxysteroid Dehydrogenase 3 Deficiency	HSD17B3
21-Hydroxylase-Deficient Classical Congenital Adrenal Hyperplasia	CYP21A2
21-Hydroxylase-Deficient Non classical Congenital Adrena lHyperplasia	CYP21A2
3-Beta-Hydroxysteroid Dehydrogenase Deficiency	HSD3B2
3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCA Related	MCCA
3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCB Related	MCCB
3-Methylglutaconic Aciduria: Type 3	OPA3
3-Phosphoglycerate Dehydrogenase Deficiency	PHGDH
5-Alpha Reductase Deficiency	SRD5A2
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	PTS
Abetalipoproteinemia	MTTP
Acrodermatitis Enteropathica	SLC39A4
Acute Infantile Liver Failure: TRMURelated	TRMU
Acyl-CoAOxidasel Deficiency	ACOX1
Adenosine Deaminase Deficiency	AMPD1
Adrenoleuko dystrophy: X-Linked	ABCD1
Alkaptonuria	HGD
Alpha-1-Antitrypsin Deficiency	SERPINA1
Alpha-Mannosidosis	MAN2B1
Alpha Thalassemia	HBA1,HBA2
Alport Syndrome: COL4A3 Related	COL4A3
Alport Syndrome: COL4A4 Related	COL4A4
Alport Syndrome: X-linked	COL4A5
Amegakaryocytic Thrombocytopenia	MPL
Andermann Syndrome	SLC12A6
Androgen Insensitivity Syndrome: Complete	AR
Antley-Bixler Syndrome	POR
Argininemia	ARG1
Argininosuccinate Lyase Deficiency	ASL
AromataseDeficiency	CYP19A1
ARSACS	SACS
Arthrogryposis,Mental Retardation, & Seizures (AMRS)	SLC35A3
Arts Syndrome	AGA
Asparagine Synthetase Deficiency	PRPS1
Aspartylglycosaminuria	ASNS
Ataxia-Telangiectasia	TTPA
Ataxia with Vitamin E Deficiency	ATM
Autosomal Recessive Polycystic Kidney Disease	PKHD1
Bardet-Biedl Syndrome: BBS11 Related	TRIM32
Bardet-Biedl Syndrome: BBS10 Related	BBS10

Bardet-Biedl Syndrome: BBS12 Related	BBS12
Bardet-Biedl Syndrome: BBS1 Related	BBS1
Bardet-Biedl Syndrome: BBS2 Related	BBS2
Bare Lymphocyte Syndrome: Type II	CIITA
Bartter Syndrome:Type 4A	BSND
Beta-Hexosaminidase Pseudodeficiency	ACAT1
Beta-Ketothiolase Deficiency	HBB
Beta Thalassemia	HEXA
Biotinidase Deficiency	BTD
Bloom Syndrome	BLM
Canavan Disease	ASPA
Carnitine-Acylcarnitine Translocase Deficiency	CPT2
Carnitine Palmitoyltransferase I A Deficiency	SLC25A20
Carnitine Palmitoyltransferase II Deficiency	CPT1A
Carpenter Syndrome	RAB23
Cartilage-Hair Hypoplasia	RMRP
Cerebrotendinous Xanthomatosis	CYP27A1
Charcot-Marie-Tooth Disease with Deafness: X-Linked: GJB1 Related	GJB1
Charcot-Marie-Tooth Disease with Deafness: X-Linked: PRPS1 Related	PRPS1
Chediak-Higashi Syndrome	VPS13A
Cholesteryl Ester Storage Disease	CHM
Choreoacanthocytosis	LYST
Choroideremia	LIPA
Chronic Granulomatous Disease: CYBA Related	CYBA
Chronic Granulomatous Disease: X-Linked	CYBB
Citrin Deficiency	SLC25A13
Citrullinemia: Type I	ASS1
Classical Galactosemia	GALT
Cockayne Syndrome:TypeA	ERCC8
Cockayne Syndrome:TypeB	ERCC6
Cohen Syndrome	VPS13B
Combined Pituitary Hormone Deficiency: PROP1 Related	PROP1
Congenital Disorder of Glycosylation: Type 1A: PMM2 Related	PMM2
Congenital Disorder of Glycosylation: Type 1B: MPI Related	MPI
Congenital Disorder of Glycosylation: Type 1C: ALG6 Related	ALG6
Congenital Insensitivity to Pain with Anhidrosis	NTRK1
Congenital Lipoid Adrenal Hyperplasia	STAR
Congenital Myasthenic Syndrome: CHRNE Related	CHRNE
Congenital Myasthenic Syndrome: DOK7 Related	DOK7
Congenital Myasthenic Syndrome: RAPSN Related	RAPSN
Congenital Ichthyosis: ABCA12 Related	ABCA12
Congenital Neutropenia: Recessive	HAX1
Copper Transport Disorders	ATP7A
Corneal Dystrophyand Perceptive Deafness	SLC4A11
Corticosterone Methyloxidase Deficiency	CYP11B2
Crigler-Najjar Syndrome	UGT1A1
Cystic Fibrosis	CFTR

Cystinosis	CTNS
Cystinuria: Non-Type I	SLC7A9
Cystinuria: Type I	SLC3A1
D-Bifunctional Protein Deficiency	HSD17B4
Diabetes:Recessive Permanent Neonatal	ABCC8
Duchenn muscula dystrophy Becke muscula dystrophy	DMD
Du Pan Syndrome	GDF5
Dyskeratosis Congenita:RTEL1 Related	RTEL1
Dystrophic Epidermolysis Bullosa: Recessive	COL7A1
Ehlers-Danlos Syndrome: Type VIIC	ADAMTS2
Ellis-van Creveld Syndrome: EVC2 Related	EVC2
Ellis-van Creveld Syndrome: EVC Related	EVC
Emery-Dreifuss Myopathy: X-Linked	EMD
Enhanced S-Cone	NR2E3
Ethylmalonic Aciduria	ETHE1
Fabry's Disease	GLA
Factor IX Deficiency	F9
Factor VIII Deficiency	F8
Familial Chloride Diarrhea	SLC26A3
Familial Dysautonomia	IKBKAP
Familial Hyperinsulinism:Type 1: ABCC8 Related	ABCC8
Familial Hyperinsulinism:Type 2: KCNJ11 Related	KCNJ11
Familial Mediterranean Fever	MEFV
Fanconi Anemia: Type A	FANCA
Fanconi Anemia: Type C	FANCC
Fancon iAnemia: Type G	FANCG
Fanconi Anemia: Type J	BRIP1
Fragile X Syndrome	FMR1
Fumarase Deficiency	FH
Galactokinase Deficiency	GALK1
Gaucher Disease	GBA
Gitelman Syndrome	SLC12A3
Globoid Cell Leukodystrophy	GALC
Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD
Glutaric Acidemia:Type I	GCDH
Glutaric Acidemia:Type IIA	ETFA
Glutaric Acidemia:Type IIB	ETFB
Glutaric Acidemia:Type IIC	ETFDH
Glycine Encephalopathy: AMT Related	AMT
Glycine Encephalopathy: GLDC Related	GLDC
Glycogen Storage Disease: Type IA	G6PC
Glycogen Storage Disease: Type IB	SLC37A4
Glycogen Storage Disease: Type II	GAA
Glycogen Storage Disease: Type III	AGL
Glycogen Storage Disease: Type IV	GBE1
Glycogen Storage Disease: Type V	PYGM
Glycogen Storage Disease: Type VII	PFKM

GM1-Gangliosidoses	GLB1
GRACILE Syndrome	BCS1L
Guanidinoacetate Methyltransferase Deficiency	GAMT
Hemochromatosis: Type2A: HFE2 Related	HFE2
Hemochromatosis: Type3: TFR2 Related	TFR2
Hemoglobinopathy: HbC	HBB
Hemoglobinopathy: HbD	HBB
Hemoglobinopathy: HbE	HBB
Hemoglobinopathy: HbO	HBB
Hereditary Fructose Intolerance	ALDOB
Hereditary Spastic Paraplegia:TECPR2 Related	TECPR2
Herlitz Junctiona lEpidermolysis Bullosa:LAMA3 Related	LAMA3
Herlitz Junctiona lEpidermolysis Bullosa:LAMB3 Related	LAMB3
Herlitz Junctional Epidermolysis Bullosa:LAMC2 Related	LAMC2
Hermansky-Pudlak Syndrome: Type1	HPS1
Hermansky-Pudlak Syndrome: Type3	HPS3
Hermansky-Pudlak Syndrome: Type4	HPS4
HMG-CoA Lyase Deficiency	HMGCL
Holocarboxylase Synthetase Deficiency	HLCS
Homocystinuria Caused by CBS Deficiency	CBS
Hunter Syndrome	IDS
Hurler Syndrome	IDUA
Hypohidrotic Ectodermal Dysplasia: X-Linked	EDA
Hypophosphatasia	ALPL
Inclusion Body Myopathy:T ype2	GNE
Infantile Cerebral and Cerebellar Atrophy	MED17
Isolated Microphthalmia: VSX2 Related	VSX2
Isovaleric Acidemia	IVD
Joubert Syndrome	TMEM216
Juvenile Retinoschisis: X-Linked	RS1
Lamellar Ichthyosis: Type1	TGM1
Laryngoonychocutaneous Syndrome	LAMA3
Leber Congenital Amaurosis: CEP290 Related	CEP290
Leber Congenital Amaurosis: GUCY2D Related	GUCY2D
Leber Congenital Amaurosis: LCA5 Related	LCA5
Leber Congenital Amaurosis: RDH12 Related	RDH12
Leigh Syndrome: French-Canadian	LRPPRC
Leukoencephalopathy With Vanishing White Matter: EIF2B5 Related	EIF2B5
Leydig Cell Hypoplasia	LHCGR
Limb-Girdle Muscular Dystrophy: Type 2A	CAPN3
Limb-Girdle Muscular Dystrophy: Type 2B	DYSF
Limb-Girdle Muscular Dystrophy: Type 2C	SGCG
Limb-Girdle Muscular Dystrophy: Type 2D	SGCA
Limb-Girdle Muscular Dystrophy: Type 2E	SGCB
Limb-Girdle Muscular Dystrophy: Type 2F	SGCD
Limb-Girdle Muscular Dystrophy: Type 2I	FKRP
Lipoprotein Lipase Deficiency	LPL

Long-Chain3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA
Lowe Oculocerebrorenal Syndrome	OCRL
Lysinuric Protein Intolerance	SLC7A7
Malonyl-CoA Decarboxylase Deficiency	MLYCD
Maple Syrup Urine Disease:Type1A	BCKDHA
Maple Syrup Urine Disease:Type1B	BCKDHB
Maple Syrup Urine Disease:Type2	DBT
Maple Syrup Urine Disease:Type3	DLD
Maroteaux-Lamy Syndrome	ARSB
Meckel Syndrome: Type 1	MKS1
Medium-ChainAcyl-CoA Dehydrogenase Deficiency	ACADM
Megalencephalic Leukoencephalopathy	MLC1
Metachromatic Leukodystrophy	ARSA
Methylmalonic Acidemia: MMAA Related	MMAA
Methylmalonic Acidemia: MMAB Related	MMAB
Methylmalonic Acidemia: MUT Related	MUT
Methylmalonic Aciduria and Homocystinuria: Type cblC	MMACHC
Mitochondrial ComplexI Deficiency: NDUFS6 Related	NDUFS6
Mitochondrial DNA Depletion Syndrome: MNGIE Type	TYMP
Mitochondrial Myopathyand Sideroblastic Anemia	PUS1
Mitochondrial Trifunctional Protein Deficiency: HADHB Related	HADHB
Morquio Syndrome: Type A	GALNS
Morquio Syndrome: Type B	GLB1
Mucolipidosis: Type II/III	GNPTAB
Mucolipidosis: Type IV	MCOLN1
Multiple Pterygium Syndrome	CHRNG
Multiple Sulfatase Deficiency	SUMF1
Muscle-Eye-Brain Disease	POMGNT1
Myotubular Myopathy: X-Linked	MTM1
Navajo Neurohepatopathy	MPV17
Nemaline Myopathy: NEBRelated	NEB
Nephrotic Syndrome: Type 1	NPHS1
Nephrotic Syndrome: Type 2	NPHS2
Neuronal Ceroid-Lipofuscinosis: CLN5 Related	CLN5
Neuronal Ceroid-Lipofuscinosis: CLN6 Related	CLN6
Neuronal Ceroid-Lipofuscinosis: CLN8 Related	CLN8
Neuronal Ceroid-Lipofuscinosis: MFSD8 Related	MFSD8
Neuronal Ceroid-Lipofuscinosis: PPT1 Related	PPT1
Neuronal Ceroid-Lipofuscinosis: TPP1 Related	TPP1
Niemann-PickDisease: Type A	SMPD1
Niemann-PickDisease: Type B	SMPD1
Niemann-PickDisease: Type C1	NPC1
Niemann-PickDisease: Type C2	NPC2
Nijmegen Breakage Syndrome	NBN
Nonsyndromic Hearing Lossand Deafness: GJB2 Related	GJB2
Nonsyndromic Hearing Lossand Deafness: LOXHD1 Related	LOXHD1
Nonsyndromic Hearing Lossand Deafness: MYO15A Related	MYO15A

Oculocutaneous Albinism: Type1	TYR
Oculocutaneous Albinism: Type 3	TYRP1
Oculocutaneous Albinism: Type 4	SLC45A2
Omenn Syndrome: DCLRE1C Related	DCLRE1C
Omenn Syndrome: RAG2 Related	RAG2
Ornithine Transcarbamylase Deficiency	OTC
Ornithine Translocase Deficiency	SLC25A15
Osteopetrosis: TCIRG1 Related	TCIRG1
Papillon-Lefevre Syndrome	CTSC
Pendred Syndrome	SLC26A4
Persistent Mullerian Duct Syndrome: Type I	AMH
Persistent Mullerian Duct Syndrome: Type II	AMHR2
Phenylalanine Hydroxylase Deficiency	PAH
POLG Related Disorders: Autosomal Recessive	POLG
Polyglandular Autoimmune Syndrome: Type 1	AIRE
Pontocerebellar Hypoplasia: EXOSC3 Related	EXOSC3
Pontocerebellar Hypoplasia: RARS2 Related	RARS2
Pontocerebellar Hypoplasia: SEPSECS Related	SEPSECS
Pontocerebellar Hypoplasia: TSEN54 Related	TSEN54
Pontocerebellar Hypoplasia: VPS53 Related	VPS53
Pontocerebellar Hypoplasia: VRK1 Related	VRK1
Primary Carnitine Deficiency	SLC22A5
Primary Ciliary Dyskinesia: DNAI1 Related	DNAI1
Primary Ciliary Dyskinesia: DNAI2 Related	DNAI2
Primary Congenital Glaucoma	CYP1B1
Primary Hyperoxaluria: Type1	AGXT
Primary Hyperoxaluria: Type2	GRHPR
Primary Hyperoxaluria: Type3	HOGA1
Progressive Familial Intrahepatic Cholestasis: Type 2	ABCB11
Propionic Acidemia: PCCA Related	PCCA
Propionic Acidemia: PCCB Related	PCCB
Pseudocholinesterase Deficiency	BCHE
Pycnodysostosis	CTSK
PyruvateCarboxylase Deficiency	PC
Pyruvate Dehydrogenase Deficiency	PDHB
Pyruvate Dehydrogenase Deficiency: X-Linked	PDHA1
Renal Tubular Acidosisand Deafness	ATP6V1B1
Retinal Dystrophies: RLBP1 Related	RLBP1
Retinal Dystrophies: RPE65 Related	RPE65
Retinitis Pigmentosa: CERKL Related	CERKL
Retinitis Pigmentosa: DHDDS Related	DHDDS
Retinitis Pigmentosa: FAM161A Related	FAM161A
Rhizomelic Chondrodysplasia Punctata: Type 1	PEX7
Salla Disease	SLC17A5
Sandhoff Disease	HEXB
Sanfilippo Syndrome: Type A	SGSH
Sanfilippo Syndrome: Type B	NAGLU

Sanfilippo Syndrome: Type C	HGSNAT
Sanfilippo Syndrome: Type D	GNS
SCID: X-Linked	IL2RG
Short-ChainAcyl-CoA Dehydrogenase Deficiency	ACADS
Sickle-Cell Anemia	HBB
Sjogren-Larsson Syndrome	ALDH3A2
Sly Syndrome	GUSB
Smith-Lemli-Opitz Syndrome	DHCR7
Spinal Muscular Atrophy: SMN1 Related	SMN1
Stargardt Disease	ABCA4
Stuve-Wiedemann Syndrome	LIFR
Sulfate Transporter-Related Osteochondrodysplasia	SLC26A2
Tay-Sachs Disease	HEXA
Trichohepatoenteric Syndrome:Type1	TTC37
Tyrosine Hydroxylase Deficiency	TH
Tyrosinemia:Type I	FAH
Tyrosinemia:Type II	TAT
Usher Syndrome:Type 1B	MYO7A
Usher Syndrome:Type 1C	USH1C
Usher Syndrome:Type 1D	CDH23
Usher Syndrome:Type 1F	PCDH15
Usher Syndrome:Type 2A	USH2A
Usher Syndrome:Type 3	CLRN1
VeryLong-ChainAcyl-CoA Dehydrogenase Deficiency	ACADVL
Walker-Warburg Syndrome	FKTN
Werner Syndrome	WRN
Wilson Disease	ATP7B
Wiskott-Aldrich Syndrome	WAS
Wolcott-Rallison Syndrome	EIF2AK3
Wolman Disease	LIPA
Xeroderma Pigmentosum: Group A	XPA
Xeroderma Pigmentosum: Group C	XPC
Zellweger Spectrum Disorders: PEX10 Related	PEX10
Zellweger Spectrum Disorders: PEX1 Related	PEX1
Zellweger Spectrum Disorders: PEX2 Related	PEX2
Zellweger Spectrum Disorders: PEX6 Related	PEX6