מחלות נדירות - מעודכן ל11/2023

49 XXXXY
AADC – Aromatic Amino Acid Decarboxylase Deficiency
ABETALIPOPROTEINEMIA
ADA2 GENE MUTATION
ADAMS OLIVER SYNDROME
AIDS
AL KAISSI SYNDROME
ALAZAMI SYNDROME
ALBRIGHT HEREDITARY OSTEODYSTROPHY
ALDRICH SYNDROME
ALEXANDER SYNDROME
Allagile
ALLGROVE (TRIPLEA)
ALSTROM SYNDROME
Alstrom Syndrome
AMBIGUOUS GENITALIA
AMELOBLASTOMA
AMINO ACID DEHYDROGENASE (AADC) DEFICIENCY
APERT SYNDROME
AROMATIC L
AROMATIC L-AMINO DECARBOXYLASE DEFICIENCY - AADC DEF
Asphyxiating Thoracic Dystrophy (see also Jeune's)
AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS
BAINBRIDGE ROPERS SYNDROME
Barth Syndrome (3-Methyl Glutaconic Aciduria type II)
Bartter
BEALS SYNDROME
Bickel Fanconi Syndrome (Completely different from Fanconi anemia)
Blackfan Diamond Anemia
BORJESON-FORSSMAN-LEHMANN SYNDROME
Byler Dis.
C syndrome- Opitz Trigonocephaly
CACNA1A MUTATION
CAMURATI ENGELMAN DISEASE
CAPILLARY MALFORMATION
CAPOS SYNDROME
Carbamoyl phosphate synthetase 1 deficiency
CARNITINE PALMITOYL TRASFERASE TYPE 2 DEF. SEVERE INFANTIL
CDH2 RELATED NEURODEVELOMENTAL DISORDER
CDK10 SYNDROME
CDON DELETION
CEREBROTENDINOUS XANTOMATOSIS
Ch. 2 deletion 2 q12.2-2q13
CHARCOT MARIE TOOTH 2N-AARS MUTATION
CHILDHOOD APRAXIA OF SPEECH
CHOPS SYNDROME
Chronic Granulomatosis
CIPA
CITRULLINEMIA TYPE 2
CNOT3 MUTATION
COATS DISEASE

Cockayne
CODAS SYNDROME
COFFIN LOWRY SYNDROME
COFFIN SIRIS SYNDROME
COL11A1 GENE MUTATION
COL2A1 MUTATION
CONGENITAL CENTRAL HYPOVENTILATION SYNDROME
CONGENITAL CHLORID DIARRHEA CH. 7 MUTATION
Congenital dyserythropoiesis –see also Crigler Najar
Congenital insensitivity to pain with anhydrosis (CIPA)
CONGENITAL MYASTHENIA SYNDROME
CONGENITAL NON PROGRESSIVE CEREBELLAR ATAXIA
CONGENITAL SUCRASE
CRANIOMETAPHYSEAL DYSPLASIA
Crigler Najar -see also congenital dyserythropoiesis
CRMO
CTNNB1 MUTATION
CTSK GENE MUTATION
Cyclic Neutropenia
CYSTIC LYMPHANGIOMA
Cystinosis
DADA2
DASS
DEND
DHDDS MUTATION
DHFR DEF.
DIFFUSE PONTINE GLIOMA
DIHIDROLIPOAMID DEHYDROGENASE DEFICIENCY
DNMT3A MUTATION
Donohue Syndrome (Leprechaunism)
Drash
DUE TO RYR1 MUTATION
DURAL SINUS MALFORMATION
Dyggve Melchior Clausen
DYSKERATOSIS CONGENITA
Dystonia Ziehen Oppenheim
ECHS1D
Ectopia cordis
Ectrodactyly ectodermal dysplasia clefting Syndrome (EEC)
EFTUD2 MUTATION SYNDROME
EHLER-DANLOS SYNDROME
Ehlers Danlos type IV
ENGELMANN 2 SYNDROME
Eosinophilic Fasciitis
EPIDERMOLYSIS BULLOSA
EPILEPTIC ENCEPHALOPATHY DUE TO ANTI FOLATE RECEPTOR AN
EPP - ERYTHROPOETIC PROTOPORPHYRIA
ERYTHEMA ANNULARE
FACTOR 10 DEFICIENCY
FASCIO-SCAPULO-HUMERAL MUSCULAR DISTROPHY
Fibrodysplasia Ossificans Progressiva
FIRES - FEBRILE INFECTION - RELATED EPILEPSY SYNDROME
Floating Harbor

FOXP1
Freeman Sheldon (see also Whistling face Syndrome)
FULL THICKNESS CORNEAL LACERATION
GASTROPARESIS
GENE ZNF292 MUTATION - DEVEL INTELL DISORDE
GFER SYNDROME
GILLESPIE SYNDROME
GLANZMAN THROMBASTHENIA
GLANZMANN THROMBASTHENIA - LIKE
GLASS SYNDROME
GLOBAL DEVELOPMENTAL DELAY - CLTC RELATED
GLUT 1 DEFICIENCY
GLUTARIC ACIDURIA TYPE 1
Glutaric Aciduria type II
GLYCOGEN STORAGE DISEASE TYPE 9A
GNAS GENE MUTATION - PSEUDOHYPOPARATHYROIDSM
Gorham disease
GREIG CEPHALPOLYSYNDACTYLY SYNDROME
GRIN2B MUTATION
H SYNDROME
H.I.V.
HELSMOOTEL VAN DER A SYNDROME
HEMIHYPERPLASIA-LIPOMATOSIS SYNDROME
HEREDITARY FRUCTOSE INTOLERANCE -ALDOB GENE MUTATION
HEREDITARY PARAGANGLIOMA PHEOCHRMOCYTOMA SYNDROME
HEREDITARY SENSORY AUTONOMIC NEUROPATHY
HEREDITARY SPASTIC PARAPARESIS
HEREDITARY SPASTIC PARAPLEGIA
HERMANSKY PUDLAK SYNDROME 5 - HPS5
HETEROZYGOUS FOR ASXL3 MUTATION
HHH Syndrome
HIBCH SYNDROME
Histiocytosis X
Hunter
Hyper IgE (Job’s)
HYPERCHOLANEMIA
Hypercholesterolemia Familial Homozyogtic
Hyperoxaluria type I
Ichtyosis:(specific types only) Lamellar, Non-bullous Congenital, Ichtyosis e
IDIOPATHIC ACUTE TRASVERSE MYELITIS
IGF1R AND HNRNPD GENE MUTATIONS
IMAGE SYNDROME
IMMUNODEFICIENCY 14A AUTOSOMAL DOMINANT
Incontinentia Pigmenti
Infantile Polyartheritis Nodosa
INTELLECTUAL DEVELOPMENTAL DISORDER WITH SEVERESPEECH
INTRACTABLE DIARRHEA OF INFANCY SYNDROM
IPEX
ISOMALTASE DEFICIENCY
ISOVALERIC ACIDEMIA
Isovaleric Acidemia
ISOVALERIC ACIDURIA
Jacobsen Syndrome

Jeune’s (Asphyxiating Thoracic Dystrophy)
Job’s (Hyper IgE)
JOUBERT SYNDROME
KABUKI SYNDROME
KAT6A SYNDROME
KBG SYNDROME
KDM5C GENE MUTATION
Kearns Syre
Kenny-Caffey Syndrome
KINDLER SYNDROME
KINSSHIP SYNDROME
KLEEFSTRA SYNDROME
KLEEFSTRA2 SYNDROME
KLINE SYNDROME
KLIPPLE TRENAUNAY SYNDROME
KOHLER SYNDROME-COWDEN
Kostman Disease
KOSTMANN SYNDROME
Krabbe Disease
LADD Lacrimo-auricular-dento-digital Syndrome
LANDAU-KLEFFNRE SYNDROME
Langerhans Cell Dis (see also Histiocytosis X)
Larsen Syndrome
LEGIUS SYNDROME
Leucocyte Adhesion Deficiency Type 2
Leucodystrophy Van der Knapp
LEUKOCYTE ADHESION DEFICIENCY TYPE 2
LIPOAMIDE DEHYDROGENASE DEFICIENCY (E3)
LIPOPROTEIN LIPASE DEFICIENCY
LOEYS-DIETZ SYNDROME
LOEYS-DIETZ SYNDROME
LONG-CHAIN 3HYDROXY-ACYL-COA DEHYDROGENASE DEFICIENCY
LOWE SYNDROME
Lymphoma congenital
LYSINURICPROTEIN INTOLERANSE (LPI)
MACROCEPHALY CAPILLARY MALFORMATION
MAJEED SYNDROME
MALAN (SOTOS LIKE) SYNDROME
MARS MUTATION
MCCUNE ALBRIGHT SYNDROME
MCM SYNDROME - MACROCEPHALY-CAPILLARY MALFORMATION
MECOM - GENE MUTATION
MECP2 GENE MUTATION
MECR MUTATION
MEF2C HAPLOINSUFFICIENCY SYNDROME
MEN2B MUTATION
MEPAN SYNDROME
METHYL MALONIC ACIDURIA
MILROY DISEASE
MIXED GONADAL DYSGENESIS
MMP 9 SYNDROME
MOEBIUS SYNDROME
Morquio Syndrome

Moya-Moya Disease
MRFACD SDR
MSUD
MUCKLE WELLS SYNDROME
Mucopolysaccharidosis type II, type IVa, type IVb & type VI
MULTIPLE HEREDITARY EXOSTOSIS AND OSTEOCHONDROMATOSIS
MUSCLE EYE BRAIN DISEASE
MUTATION IN CUL4B GENE
MYCOSIS FUNGOIDES
MYH7 GENE MUTATUON
MYHRE SYNDROME
Myhre Syndrome
NEONATAL PROGERIA
Nephrogenic Diabetes Insipidus (only X-linked N.D.I)
Netherton Syndrome
NEVUS UNIUS LATERIS
NFIA MUTATION - BRAIN MALFORMATION WITH OR WITHOUT URINAI
NICOLAIDES-BARAITSER SYNDROME
NIEMANN PICK C
NOONAN SYNDROME 7- CARDIOFACIOCUTANEOUS SYNDROME
NSD1 MUTATION
O'DONNEL-LURIA-RODAN SYNDROME
OCCULO FACIAL DENTAL SYNDROME
OGDEN SYNDROME
OHDO SYNDROME SBBYS VARIANT
OLLIIER DISEASE
Omenn
Pallister-Hall Syndrome
PAN - Periartheritis Nodosa
Papillorenal Syndrome (Pax2 gene mutation)
PARKES WEBER SYNDROME
Pearson marrow pancreas syndrome
PEDIATRIC MULTIPLE SCLEROSIS
Pelizaeus-Merzbacher Disease
PENTA X
PEPCK-C OMIM 26680
PERCC1
PERIODIC PARALYSIS - DUE TO RYR1 MUTATION
PFEIFFER SYNDROME
PHELAN MCDERMID SYNDROME
PHF21A GENE MUTATION
PHOSPHOGLUCOMUTASE 1 DEFICIENCY
PICNODYSOSTOSIS
PITT HOPKINS LIKE SYNDROME
PITT HOPKINS SYNDROME
PITUITARY STALK INTERUPTION SYNDROME
PLP1 MUTATION
POLAND SYNDROME
POPLITEAL PTERYGIUM SYNDROME
POTOKI-LUPSKI SYNDROME
PPB - PLEUROPULMONARYBLASTOMA
PRIMARY HYPEROXALURIA TYPE 1
Progyria

PROLIDASE DEFICIENCY
PRSS1 MUTATION
Pseudo-hypo-aldosteronism
PSEUDOHYPOALDOSTERONISM TYPE 1B
PSEUDOHYPOPARATHYROIDSM TYPE2
PTEN MUTATION
PTLD (post transplantation lymphoproliferative dis)
PUDLAK TYPE 2 SYNDROME
PULMONARY ALVEOLAR PROTEINOSIS
PURA SYNDROME
Pycnodysostosis
Pyruvate Dehydrogenase deficiency
RENPENNING SYNDROME - PQBP1 MUTATION
RETICULATE PIGMENTED DISORDER
Rogers Syndrome
RT TMJ ANKYLOSIS
SATB2 ASSOCIATED SYNDROME
SCHAAF - YANG SYNDROME
Schimke Immuno-Osseous Dysplasia
SCHINZEL GIEDION
SCHUURS JMAKERS SYNDROME
SCIMITAR SYNDROME
SCN4A MUTATION
SCYL1 MUTATION - SPINOCEREBELLAR ATAXIA TYPE 21
SEPIAPTERIN REDUCTASE DEFICIENCY
SETD5 MUTATION
SHANK 2 MUTATION
SHANK3 GENE MUTATION
SHORT SYNDROME
SIALURIA MIM 269921
SIFRIM-HITS-WEISS SYNDROME
SIFRIM-HITZ-WEISS SYNDROME
SILVER RUSELL SYNDROME
SLC9A7 GENE MUTATION
SLO -SMITH LEMLI OPITZ SYNDROME
SMARCA 2 MUTAION
SMITH KINGSMORE SYNDROME
SMOOTH MUSCLE DYSFUNCTION - ACTA2 MUTATION
SNIJDERS BLOCK CAMPEAU SYNDROME
SOX2 MUTATION
SPONDYLO METAPHYSEAL DYSPLASIA - VARIANT RPL 13
Spondyloepiphyseal Dysplasia
STAT 3 MUTATION
Stevens Johnson
SUCRASE ISOMALTASE DEFICIENCY
SYNGAP1- RELATED INTELLECTUAL DISABILITY
SYP1
SYSTEMIC MASTOCYTOSIS
Systemic Scleroderma
SZT2 GENE MUTATION
Takayasu Vasculitis
TAR SYNDROME
TATTON-BROWN-RAHMANSYNDROME - TBRS

TEC -PR2 SYNDROME - OBSTRUCTIVE SLEEP APNEA SYNDROME
TRICHOHEPATOENTERIC SYNDROME
TRICHORHINOPHALANGEAL SYNDROME
TRMT10A RELATED DISORDER
TUBER CINEREUM HAMARTOMA
TYROSINEMIA TYPE 2
TYROSINEMIA TYPE 3
Tyrosinemia type I
Tyrosinemia type II
Upshaw Schulman Syndrome
USP7 GENE MUTATION
Van der Knapp (see also Leucodystrophy)
VERHEIJ SYNDROME
VERVERI - BRADY SYNDROME
VICI SYNDROME
VON WILLEBRANDS DIS.
Wegener Granulomatosis
WEISS- KRUSZKA SYNDROME
Widemann Rautenstrauch Syndrome
WIEDEMANN-STEINER SYNDROME
Wiskott Aldrich
WISKOTTALDRICH SYNDROME
X LINKED INTELLECTUAL DISABILITY - SIDERIUS TYPE
Xeroderma Pigmentosum
XIA-GIBBS SYNDROME
Ziehen Oppenheim Dystonia
ZNF292
ZNF335 -GENE MUTATION
ZTTK
16P11.2P12.2 MICRODELETION SYNDROME
820KB DUPLICATION FROM CHROMOSOME 17P13.1
ADNP SYNDROME
AICARDI SYNDROME
AICARDI-GOUTIÈRES SYNDROME
ALLAN-HERNDON-DUDLEY SYNDROME
ALSTRÖM SYNDROME
ALTERNATING HEMIPLEGIA OF CHILDHOOD
ARBOLEDA-THAM SYNDROME
AUTOSOMAL DOMINANT HYPER-IGE SYNDROME DUE TO STAT3 DEFICIENC
AUTOSOMAL DOMINANT INTELLECTUAL DISABILITY-CRANIOFACIAL ANOMA
AUTOSOMAL RECESSIVE CEREBELLAR ATAXIA-EPILEPSY-INTELLECTUAL DI
AUTOSOMAL RECESSIVE LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 28
AYMÉ-GRIPP SYNDROME
BRD4 RELATED SYNDROME
BROMODOMAIN AND WD REPEAT DOMAIN CONTAINING 3 - BRWD3
BRUTON TYPE AGAMMAGLOBULINEMIA
CAUDAL DUPLICATION
CENTRAL CORE DISEASE
CHD3-RELATED DEVELOPMENTAL DELAY-SPEECH DELAY-INTELLECTUAL D
CHYLOMICRON RETENTION DISEASE
CLTC NON-SPECIFIC EARLY-ONSET EPILEPTIC ENCEPHALOPATHY
COFFIN-SIRIS SYNDROME
COL4A1-RELATED BRAIN SMALL VESSEL DISEASE WITH HEMORRHAGE

CONGENITAL CHRONIC DIARRHEA WITH PROTEIN-LOSING ENTEROPATHY
CONGENITAL CONTRACTURAL ARACHNODACTYLY DUE TO DELITION OF EX
CONGENITAL PSEUDOARTHROSIS OF THE TIBIA
CONGENITAL STATIONARY NIGHT BLINDNESS
CRANIOECTODERMAL DYSPLASIA (WDR35 MUTATION)
CRANIOFRONTONASAL DYSPLASIA
CTX - CEREBROTENDINOUS XANTHOMATOSIS
CURRARINO SYNDROME
CUTIS MARMORATA TELANGIECTATICA CONGENITA
DENT DISEASE TYPE 1
EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY
EARLY-ONSET EPILEPTIC ENCEPHALOPATHY AND INTELLECTUAL DISABILIT
EMANUEL SYNDROME
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS
FAMILIAL ISOLATED RESTRICTIVE CARDIOMYOPATHY
FAMILIAL SHORT QT SYNDROME
FIBRODYSPLASIA OSSIFICANS PROGRESSIVA
FOCAL DERMAL HYPOPLASIA
FORSIUS-ERIKSSON SYNDROME
GORHAM SYNDROME
GREIG CEPHALOPOLYSYNDACTYLY SYNDROME
HADDS - HYPOTONIA, ATAXIA, AND DELAYED DEVELOPMENT SYNDROME
HEREDITARY HYPOPHOSPHATEMIC RICKETS WITH HYPERCALCIURIA
HIDEA SYNDROME
ICHTHYOSIS TYPE ARC1 AUTOSOMAL RECESSIVE CONGENITAL
INAD1-INFANTILE NEUROAXONAL DYSTROPHY
INTELLECTUAL DISABILITY-EXPRESSIVE APHASIA-FACIAL DYSMORPHISM S
INTELLECTUAL DISABILITY-FACIAL DYSMORPHISM SYNDROME DUE TO SET
INTELLECTUAL DISABILITY, BIRK-BAREL TYPE
KLIPPEL-TRÉNAUNAY-WEBER SYNDROME
KOHLSCHÜTTER-TÖNZ SYNDROME
LAMELLAR ICHTHYOSIS
LEBER CONGENITAL AMAUROSIS
MAGEL2-RELATED PRADER-WILLI-LIKE SYNDROME / SCHAAF-YANG SYNDR
MCTD - MIXED CONNECTIVE TISSUE DISEASE
MED13L-RELATED INTELLECTUAL DISABILITY SYNDROME
METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, TYPE CBLC
MICROCEPHALY-CORPUS CALLOSUM HYPOPLASIA-INTELLECTUAL DISABILI
MICRODELETION 9Q22.3
MUSD- MAPLE SYRUP URINE DISEASE
MYOCLONUS EPILEPSY AND ATAXIA DUE TO POTASSIUM CHANNEL MUTATI
OKUR-CHUNG NEURODEVELOPMENTAL SYNDROME
PARANEOPLASTIC OPSOCLONUS-MYOCLONUS-ATAXIA SYNDROME
PEARSON SYNDROME
PHELAN-MCDERMID SYNDROME
PHOCOMELIA - CONGENITAL ABSENCE OF THIGH AND LOWER LEG WITH FO
PHOCOMELIA - CONGENITAL ABSENCE OF UPPER ARM AND FOREARM WIT
PIK3CA-RELATED OVERGROWTH SYNDROME
POMT1-RELATED LGMD R11
POSTSYNAPTIC CONGENITAL MYASTHENIC SYNDROMES
PPB FAMILIAL TUMOR AND DYSPLASIA SYNDROME
PRIMARY MICROCEPHALY-MILD INTELLECTUAL DISABILITY-YOUNG-ONSET
PRIMROSE SYNDROME

PROGRESSIVE MYOCLONIC EPILEPSY TYPE 1
PSEUDOTUMOR CEREBRI
RASMUSSEN SUBACUTE ENCEPHALITIS
RENAL NUTCRACKER SYNDROME
RETT SYNDROME
RUBINSTEIN-TAYBI SYNDROME
SCN2A-RELATED NEURODEVELOPMENTAL DISORDER
SPENCDI
STXBP1-RELATED ENCEPHALOPATHY
TATTON-BROWN-RAHMAN SYNDROME
TEMPLE SYNDROME
TET3-RELATED BECK-FAHRNER SYNDROME
WIEACKER-WOLFF SYNDROME
WIEDEMANN-STEINER SYNDROME
WOLFRAM SYNDROME
X-LINKED INTELLECTUAL DISABILITY-CEREBELLAR HYPOPLASIA SYNDROM
X-LINKED INTELLECTUAL DISABILITY-HYPOTONIA-MOVEMENT DISORDER S
XQ28 MICRODUPLICATION SYNDROME
ZINC FINGER HOMEOBOX 3 - ZFHX3