

DIAGNOSED DISEASES AT “M. BASCHIROTTO” INSTITUTE FOR RARE DISEASES

- Achondroplasia (FGFR3)
- Adrenoleukodystrophy (ABCD1)
- Alpha-1 Antitrypsin Deficiency (SERPINA1)
- Alzheimer Disease Type 1 (APP), 3 (PSEN1), 4 (PSEN2), Linked To APOE
- Amyotrophic Lateral Sclerosis 1 (SOD1)
- Angelman Syndrome (UBE3A)
- Ataxia, Early-Onset, With Oculomotor Apraxia - Hypoalbuminemia (APTX)
- Autoimmune Polyendocrine Syndrome Type 1 (AIRE)
- Basal Cell Nevus Syndrome (PTCH1)
- Ceroid Lipofuscinosis, Neuronal, 1 (PPT1)
- Charcot-Marie-Tooth Disease, Demyelinating, Type 1A (PMP22), X-Linked 1 (GJB1)
- Creutzfeldt-Jakob Disease (PRNP)
- Cystic Fibrosis (CFTR)
- Darier-White Disease (ATP2A2)
- Deafness, Aminoglycoside-Induced (MTRNR1)
- Deafness, Autosomal Recessive 1A (GJB2), 1B (GJB6)
- Diabetes Insipidus, Nephrogenic, X-Linked (AVPR2)
- Epileptic Encephalopathy, Early Infantile, 2 (CDKL5)
- Farber Lipogranulomatosis (ASAH1)
- Fatal Familial Insomnia (PRNP)
- Friedreich Ataxia 1 (FXN)
- Frontotemporal Dementia (GRN)
- Gaucher Disease, Type I (GBA)
- Gout, Hprt-Related (HPRT1)
- Groenouw Type I Corneal Dystrophy (TGFB1)
- Hemochromatosis (HFE), Juvenile (HFE2, HAMP), 3 (TFR2), 4 (SLC40A1)
- Hepatic Venoocclusive Disease With Immunodeficiency (SP110)
- Homocysteinemia (CBS)
- Homocystinuria Due To Deficiency Of N(5,10)- MTHFR Activity (MTHFR)
- Hyperferritinemia-Cataract Syndrome (FTL)
- Huntington Disease (HTT), Like 2 (JPH3)
- Hyperkalemic Periodic Paralysis (SCN4A)
- Hypokalemic Periodic Paralysis (CACNA1S, KCNE3, SCN4A)
- Krabbe Disease (GALC)
- Leber Optic Atrophy (MTND1-4-5-6)
- Lesch-Nyhan Syndrome (HPRT1)
- Li-Fraumeni Syndrome 1 (TP53), 2 (CHEK2)
- Lymphedema-Distichiasis Syndrome (FOXC2)
- Malignant Hyperthermia, Susceptibility To, 1 (RYR1), 5 (CACNA1S)
- Mental Retardation, X-Linked, Arx-Related (ARX)
- Metachromatic Leukodystrophy (ARSA, PSAP)
- Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes
- Multiple Sulfatase Deficiency (SUMF1)
- Myoclonic Epilepsy Of Lafora (EPM2A); Associated With Ragged-Red Fibers
- Myotonia, Potassium-Aggravated (SCN4A)
- Netherton Syndrome (SPINK5)
- Neuropathy, Ataxia, And Retinitis Pigmentosa (MTATP6)
- Niemann-Pick Disease, Type A (SMPD1), B (SMPD1), C1 (NPC1)
- Ornithine Aminotransferase Deficiency (OAT)
- Paramyotonia Congenita Of Von Eulenburg (SCN4A)
- Parkinson Disease, Type 1 (SNCA), 2 (PARK2), 5 (UCHL1), 6 (PINK1), 7 (PARK7), 8 (LRRK2), 13 (HTRA2)
- Parkinson-Dementia Syndrome (MAPT)
- Pelizaeus-Merzbacher Disease (PLP1)
- Pendred Syndrome (SLC26A4)
- Prader-Willi Syndrome
- Pseudoxanthoma Elasticum (ABCC6)
- Renal Tubular Acidosis Distal Aut. Domin. (SLC4A1); Renal Tubular Acidosis With Hemolytic Anemia (SLC4A1)
- Rett Syndrome (MECP2)
- Silver-Russell Syndrome (Chr 7)
- Spastic Ataxia, Charlevoix-Saguenay Type (SACS)
- Spastic Paraplegia 3, Aut. Domin (ATL1), 4 (SPAST), 7 Aut. Reces. (SPG7)
- Spherocytosis, type 4 (SLC4A1)
- Spinal And Bulbar Muscular Atrophy, X-Linked 1 (AR)
- Spinal Muscular Atrophy, Type I (SMN1)
- Spinocerebellar Ataxia Type 1 (ATXN1), 2 (ATXN2), 3 (ATXN3), 5 (SPTBN2), 6 (CACNA1A), 7 (ATXN7), 8 (ATXN80S), 10 (ATXN10), 12 (PPP2R2B), 14 (PRKCG), 17 (TBP), 27 (FGF14)
- Thrombophilia (F2), Thrombophilia Due To Activated Protein C Resistance (F5)
- TP53 Associated Diseases (TP53)
- Tuberous Sclerosis (TSC1, TSC2)
- Waardenburg Syndrome, Type I (PAX3)
- Wilson Disease (ATP7B)
- Yellow Nail Syndrome (FOXC2)