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Abbrevations: A= alive; AD= adulthood; AGR= arthrogryposis; AO= azoospermia; AS= asymptomatic; AT= ataxia; BC= bilateral cataract; BCSH= bilateral chronic subdural hematoma; BE= brainstem edema; BL= brainstem lesions; BS= buccal swab; BSH= brain stem hypoplasia; BSL= basal ganglia lesions; C= cataract; CA= cerebellar atrophy; CAA= cerebellar ataxia; CBA= cerebral atrophy; CD= childhood; CF= cardiac failure; CH= cerebellar hypoplasia; CL= cerebellar lesions; CM= choreatic movements; CMHW= cerebellar white matter hyperintensities; CO= coma; CP= Cataplexy; CPEO= chronic progressive external ophthalmoplegia; CT= computertomography; CV= cyclic vomiting; D= died; DD= developmental delay; DF= dwarfism; DM= diabetes mellitus; DMY= delayed myelinization; DOC= disturbance of consciousness; DR= developmental regression; DY= dysphagia; DYS= dysartria; DYT= dystonia; EP= encephalopathy; EOL= edema occipital lobe, ES= epileptic stroke; F= female; FB= fibroblasts; FI= floppy infant; FPA= fronto-parietal atrophy; FTT= failure to thrive GD= gait disturbances; GDD= global developmental delay; gm= grandmother; GN= glomerulosclerosis; GS= gallstones; HE= hepatopathy; HI= hearing impairment; HL= hypothalamic lesions; HY= hypothyreosis; KY= kyphosis; L= leukocytes; LA= lactic acidosis; LD= learning disability; LLF= Leigh-like findings; LLS= Leigh-like Syndrome; LS= Leigh Syndrome; M= male; MA= multisystemic ataxia; MB= M. Basedow; MC= Morbus Crohn; MS= muscle cells; MDD= motor developmental delay; MIC= microcephaly; ML= midbrain lesions; MLS= multiple lesions; mo=mother; MR= motor regression; MRD= mental retardation; MW= muscle weakness; NARP= neurogenic weakness, ataxia, retinitis pigmentosa; n= normal; n.a..= not available; NNP= neonatal period; NP= neuropathy; NS= nephrotic syndrome; OA= optic atrophy; OI= ovarial insufficiency; ONH= optic nerve hypoplasia; PA= postnatal asphyxia; PC= pes cavus; PP= prenatal period; PS= psychosis; PWML= periventricular white matter lesions; RD= retinal detachment; RF= respiratory failure; RP= retinitis pigmentosa; S= spasticity; SA= spastic ataxia; sb= sibling; SC= scoliosis; SD= speech delay; SLE= stroke-like episode; SM= skeletal muscle; SNP= spastic neuropathy; SPD= spine deformities; SS= short stature; SU= suspected; TCC= thin corpus callosum; TL= thalamic lesions; TP= tetraparesis; TR= tremor; U= urine; VI= visual impairment; WPW= Wolff-Parkinson-White; y= years;