**Supplementary Table 1.** Clinical characteristics of patients with type I and II sialidosis.

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| **Authors** | **Number of patients (n)** | **Ethnicity** | **Genetic Alterations** | **Type of alteration** | **Age at onset** | **Age at evaluation** | **Ophthalmologic findings**  | **Neurologic findings** | **Skeletal findings** | **Other****findings** | **EEG and Brain Imaging** | **EKG** | **Cardiovascular findings including ECHO** |
| Bonten et al.1 | 11(5 type I, 6 type II) | 2 African American 2 Italian1 Greek2 German1 Dutch1 Hispanic 2 Unknown(Caucasian) | Patient 1: Arg294Ser,Leu231His (Compound Heterozygous) Patient 2: Arg296Ser, Gly218Ala (Compound Heterozygous) Patient 3:Gly227Arg (Homozygous) Patient 4: Gly227Arga (Homozygous)Patient 5 and 6 (siblings): Val54Met, Gly378stop (Compound Heterozygous) Patient 7: Gly328Ser, Dupl399HisTyr (Compound Heterozygous)Patient 8: Tyr370Cys (Homozygous)Patient 9: Phe260Tyr (Homozygous)Patient 10:Phe260Tyr,Leu363pro (Compound Heterozygous) Patient 11:Arg280stop,Pro335Gln (Compound Heterozygous) | Point mutations, deletions and insertion | Mean:7.86 yrs (20 wk gestational period-17 yrs) | Mean:7.61yrs(4 months-44 yrs (death) | Cherry-red spots: 5/11(macular 1)Visual acuity:diminution: 2/11Ocular lens opacities: 2/11Prolonged latencies on visual evoked potentials:1/11Intolerance to light: 1/11 | Seizures: 5/11(myoclonus 4)Ataxia: 4/11Slurred speech:1/11Dysmetria: 2/11Microcephaly: 1/11Hearing loss: 4/11(Sensorineural deafness 1)Moderate delay in speech: 1/11Spasticity: 1/11Hypotonia: 3/11Cerebellar signs: 1/11Dysarthria: 3/11Dysphagia: 1/11Dydiadchokinesia: 1/11Pyramidal signs: 1/11Axonal polyneuropathy: 1/11Orbital hypoplasia: 1/11Mental retardation:mild(1/11) Developmental delay: 4/11Hydrocephalus: 1/11  | N/A | N/A | EEG:Abnormal 2/11 (1with normal and 1 with borderline IQ) | Non-specific alterations of repolarization (1/11)  | Normal ECHO 4/11Cardiomegaly/Cardiomyopathy 2/11 (both type II) |
| Itoh et al2 | 4[2 type I(mild juvenile-onset) 2 type II(congenital form)] | Japanese (East Asian) | Type II: C239T (P80L), T718C (W240R) (Compound Heterozygous)Type I: C946T (p.P316S) (Homozygous) | Missensemutation | Fetal life (21 to 33 weeks’ gestation), Type I 14 yrs | Birth, 24 yrs (Type I) | Bilateral cherry-red spots | Ataxia, dysgraphia, generalized tonic clonic seizures, dysarthria, action myoclonus | N/A | Hepatosplenomegaly, ascites | N/A | N/A | N/A |
| Lukong KE et a34 | 9(2 type I,7 type II) | Patient 1: MexicanPatient 2: American (Caucasian)Patient 3: Ashkenazi Jewish Patient 4: Chinese (East Asian)Patient 5: Turkish Patient 6: Polish Patient 7 and 8 (siblings): SpanishPatient 9: Italian  | Patient 1 and 2: Gly227Arg (Homozygous)Patient 3: Ala298Val (Homozygous)Patient 4: Ser182Gly (Homozygous)Patient 5: Frameshift, Stop904 \*deletion (Homozygous)Patient 6: Gly68Val (Homozygous)Patient 7 and 8: Leu270Phe (Homozygous)Patient 9: Gly328Ser (Compound Heterozygous) | Missense mutations and a deletion | 10.84yrs(3 months-24 yrs) | N/A | Cherry-red spots: 4/9 | Seizures: 2/9Ataxia: 1/9Mental retardation: 3/9Myoclonus: 2/9 | Skeletal dysplasia 1/11 | Hydrops fetalis: 4/11Nephrosialidosis: 1/11Mild dysphagia: 1/9 | N/A | N/A | N/A |
| Naganawa et al4 | 2 (type I) | Japanese (East Asian) | G649A (V217M), G727A (G243R) (both Compound Heterozygous) | N/A | 17 & 32 yrs | 25 & 42 yrs | Lens opacities, macular cherry-red spots | Ataxic gait, speech disturbance, generalized tonic clonic seizures, dysarthria, action myoclonus, hyperactive deep tendon reflexes | N/A | N/A | Normal MRI brain | N/A | N/A |
| Oohira Tet al5 | 1 (type II) | Japanese (East Asian) | N/A | N/A | 14 days | 5 years | Macular cherry-red spots | Mental retardation, myoclonus, cerebellar ataxia, brisk tendon reflexes, positive Babinski sign,  | Coarse facies, dislocation of hip joints, contractures of the first fingers of both hands, scoliosis, oar like ribs, vertebral hypoplasia | Congenital adrenal hyperplasia, hepatosplenomegaly, gingival hyperplasia, short stature  | CT scan: Mild brain atrophy | N/A | Grade 2/6 systolic murmur |
| Pattison S et al6 | 4 (type II) | 1 Czech (Ashkenazi Jewish)1 Irish/French Canadian2 Mennonites | G674C (p.R225P), C893T (p.A298V), G3A (p.M1?), C941G (p.R341G), and G69A (p.W23X) | Missense (4) and 1 nonsense. | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A |
| Penzel R et al7  | 1 (female, type II)  | Not known | G2736C transversion (Homozygous) | Transversion (Intronic) | 27 weeks’ gestation | 27 weeks’ gestation | N/A | Myoclonus | N/A | Ascites, edema, hydrops fetalis | Fine calcifications of the thalamic striatal blood vessels on cranial USG | N/A | N/A |
| Sergi C et al8 | 2 (type II)  | Turkish | W29X (substitution of tryptophan at codon 29 by a termination codon) | Nonsense mutation | Patient 1: Delivered at 26 weeks’ gestation and died at 2 monthsPatient 2: Pregnancy terminated at 20 weeks’ gestation | N/A | N/A | N/A | N/A | Hydrops fetalis, hepatomegaly, respiratory distress syndrome, and anemia | N/A | N/A | N/A |
| Seyrantepe V et al9Ledvinova J et al10 | 4 (type I)  | 2 Czech 2 French  | 2 Czech patients C1034T (T345I) (Homozygous) French Patient 1 G407E p.G136E and T824C p.V275A (Compound Heterozygous)  French Patient 2 (Type 1): T332C p.L111P (Heterozygous). Second mutation not determined. | 3 substitutions, 1 missense | N/A | 15.5 yrs(14-20 yrs) | Ocular myoclonus, cherry-red spots  | Myoclonus, tremor, partial complex seizures, progressive ataxia, dyskinetic extrapyramidal and cerebellar syndromes, quadriparesis, tremor, hyperreflexia | Dysostosis multiplex  | N/A | N/A | N/A | N/A |
| Uhl J et al11 | 2 (type II) | Turkish | 11 kb deletion encompassing entire coding region of *NEU1* (Homozygous) | Deletion | Prenatally diagnosed | N/A | N/A | N/A | Polydactyly | Ascites, hydrops fetalis, hepatomegaly | N/A | N/A | N/A |
| Bonten E et al12 | 3[2 type I(siblings), 1 type II] | Unknown | Patient 1 and 2 (siblings): G1258T transversion (Heterozygous) Patient 3: T 401G transversion (p.Leu91Arg) and a single-basedeletion at nucleotide 1337 (Compound Heterozygous)  | Point mutations and a deletion  | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A |
| Lowden JA et al13 | 37 (18 type I, 19 type II) | 7 Italian10 Japanese5 Caucasian | N/A | N/A | Type 12.4 ± 4.7 yrsType 2 Juvenile 10.4 ± 5.3 yrsInfantile: 0-10 months | N/A | Cherry-red spots (33/37), Corneal clouding/Lens opacities (17/37), loss of visual acuity (22/37) | Seizures (15/37), myoclonus (30/37), ataxia (15/37), pyramidal tract signs (7/37), mental retardation (10/37) | Coarse facies (19/37), Vertebral changes (18/37), Growth disturbance (16/37),  | Visceromegaly (6/37), Foam cells (19/37), Hearing loss (9/37) | Abnormal EEG (13/37) | N/A | N/A |
| Sergi C et al14 | 1 (type II neonatal-onset) | Syrian | N/A | N/A | Prenatal diagnosis (27 weeks’ gestation) | N/A | Corneal clouding | N/A | Coarse facies | Visceromegaly, ascites, hydrops, inguinal hernia | Normal CNS ultrasound | N/A | Normal ECHO |
| Lemyre E et al15 | 3 (type II) | 3 French Canadian | N/A | N/A | 31-32 weeks’ gestation | Birth | Clear corneae, absence of cherry-red spot | Psychomotor retardation, hypotonia, nystagmus, decreased deep tendon reflexes  | Coarse facies, widening of long bones and ribs, J-shaped sella turcica and calcaneal calcifications, dysostosis multiplex | Fetal ascite, nephrotic syndrome, heart failure, esophageal atresia type 3, gum hypertrophy, failure to thrive, hepatosplenomegaly, sensorineural deafness | N/A | N/A | Moderate LVH, cardiomegaly (patient 2 who died of heart failure) |
| Loren DJ et al16 | 1 (type II)  | Caucasian | G45A (Exon 1) and G1022A (Exon 5) (Compound Heterozygous) | Nonsense and splice site mutations | Prenatal diagnosis (18 weeks’ gestation) | Birth | N/A | N/A | N/A | Polyhydramnios, ascites, lung involvement | N/A | N/A | N/A |
| Lee BH et al17 | 1 (type II)  | Korean (East Asian) | C239T (p.Pro80Leu)(Homozygous) | N/A | Prenatal diagnosis (22 weeks’ gestation) | Birth | Lack of visual fixation and ability to track objects  | Hypotonia | Coarse facial features | Ascites, hydrops fetalis, cardiomegaly, hepatosplenomegaly, sparse facial hair, gum hypertrophy | N/A | N/A | Cardiomegaly, ventricular dilation, huge PDA which closed on day 15 |
| Tabardel Y et al18 | 1 (type II)  | French | N/A | N/A | N/A | Birth | N/A | N/A | Facial dysmorphism | Hydrops fetalis, heart failure | N/A | N/A | N/A |
| Ahn JH et al19 | 1 (type I)  | Korean (East Asian) | G928A (p.D310N), novel c.15\_16del (p.P6Qfs\*21)(Compound Heterozygous) | N/A | 12 yrs | 36 yrs | Bilateral macular cherry-red spots, thickening of peripapillary retinal fiber layer in both eyes | Ataxia, myoclonus, generalized tonic clonic seizures, dysarthria | N/A | N/A | EEG 13 years spike and polyspike wave pattern, MRI mild cerebellar atrophy | N/A | N/A |
| Gultekin M et al20 | 1 (type I) | Turkish | NM\_000434:c.625delG (p.Glu209SerfsTer94) and novel NM\_000434:c.92G8A (p.Asp310Asn) (Compound Heterozygous) | Frameshift  | 18 yrs | 24 yrs | Bilateral macular cherry-red spots | Truncal ataxia, dysarthria, intentional tremor of the upper limbs, generalized seizures | N/A | N/A | MRI: Mild brain atrophy | N/A | N/A |
| Ranganath P et al21 | 2 (1 type I , 1 type II) | South Asian | **Patient 1:** C880T transition in exon 5 (R294C) and a 1-base deletion at nucleotide 1191 (c.1191delG) in exon 6 (Compound Heterozygous)**Patient 2:** Homozygous for a 1-base deletion at nucleotide 73 (c.73delG)  | Missense and frameshift mutations | N/A | Patient 1: 14 yrsPatient 2: 1.5 yrs | Bilateral fundal cherry-red spots, mild corneal haziness | Gait disturbance, dysarthria, myoclonic jerks, fine tremors at rest, dysmetria, nystagmus, gait ataxia,  | Macrocephaly, coarse facies,bilateral genu valgum | Protuberant tongue, gum hypertrophy, generalized hypertrichosis, large Mongolian spots on the back, umbilical hernia, hepatomegaly | N/A | N/A | Thickening of mitral valve leaflets with mitral valve prolapse/mitral regurgitation |
| Sekijima Y et al22 | 1 (type I) | Japanese | C239T (p.P80L), G403A (p.D135N) (Compound Heterozygous) | Missense | 14 years | 16 years | Bilateral macular cherry-red spots | Subnormal intelligence, dysarthria, myoclonus, intentional tremors, limb and gait ataxia, hyperreflexia | N/A | N/A | MRI Brain atrophy | N/A | Normal ECHO |
| Sobral I et al23 | 1 (type I) | Portuguese | G700A (p.D234N) exon 4, C1021T(p.R341X) exon 5 (Compound Heterozygote) | Truncated protein | 26 yrs | 53 yrs | Reduced visual acuity, bilateral cherry-red spots, atrophy of nerve fibers, visual field defects | Myoclonus, ataxic gait, dysarthria and difficulty in writing | N/A | N/A | N/A | N/A | N/A |
| Canafoglia L et al 24 | 6 (type 1) | Italian | c.200G>T, p.S67I (Homozygous), (G679A, p.G227R; C913T, p.R305C) (Compound Heterozygous) | N/A | 22-32 yrs | 27-42 yrs | Cataract | Late onset myoclonus, dysarthria | Femur head necrosis, scoliosis | N/A | Craniospinal MRI: Cerebellar vermis atrophy, syringomyelia; Somatosensory evoked potentials: Enlarged | N/A | N/A |
| Schene IF et al25 | 2 (type 1) | Dutch | c.1195\_1200dup p.His399\_Tyr400dup; G679A, p.Glu227Arg (Compound Heterozygous) | N/A | 12, 13 yrs | 14, 15 yrs | Nummular cataract, macular cherry-red spot, and optic nerve atrophy | Tremors, ataxia, myoclonus, downbeat nystagmus | Avascular osteonecrosis of right femoral head | N/A | Normal brain MRI | N/A | N/A |
| Mutze U et al26 | 1 (type 1) | German | C699CA, p.S233R in exon 4 and A803G; p.Y268C in Exon 5 (Compound Heterozygous) | Missense | N/A | 6 yrs | Macular cherry-red spots  | Normal neurological examination | N/A | N/A | EEG: Generalized epileptic discharges; Visual evoked potentials: Prolonged bilaterallyNormal brain MRI | N/A | N/A |
| Gowda VK et al27 | 1 (type 1)  | South Asian | G742G T (p.G248C) in exon 4 (Homozygous) | Missense | 9 yrs | 9 yrs | Bilateral macular cherry-red spots | Generalized tonic clonic seizures, ataxia, myoclonus, spasticity, increased deep tendon reflexes | N/A | N/A | Normal brain MRI and EEG | N/A | N/A |
| Aravindhan A et al28 | 1 (type 1)  | Ecuadorian | C629T (p.Pro210Leu) (Homozygous) | Missense | 16 yrs | 39 yrs | Bilateral cherry-red spots | Generalized tonic clonic seizures, myoclonic seizures, broad based ataxic gait, truncal and appendicular ataxia  | N/A | N/A | EEG: Revealed multiple brief myoclonic seizures and generalized slow spike/polyspike wave complexes | N/A | N/A |
| Hu SC et a29 | 1 (type 1) | East Asian | A544 G (Ser182Gly), C619T (termination codon) | Missens, nonsense | 12 yrs | 15 yrs | Bilateral cherry -red spots | Myoclonus, gradually progressive ataxia, tremors, and psychomotor and speech regression | N/A | N/A | EEG: Focal spikes arising from bilateral centrotemporal regions, followed by ictal myoclonic seizures with generalized muscle contraction activities lasting 15–20 s; MRI: Mild brain atrophy | N/A | N/A |
| Mohammed AN et al30 | 1 (type 1) | East Asian | A554G (p.S182G) and G679A (p.G227R) (Compound Heterozygous) | Missense | 12 yrs | 20 yrs | Normal | Ataxia, generalized tonic clonic seizures, myoclonus, saccadic hypometria, increased tone, brisk deep tendon reflexes | N/A | N/A | Normal EEG, Normal brain MRI | N/A | N/A |
| Cameron PD et al12 | 1 (type II)  | Caucasian | N/A | N/A | Birth | Birth | Poor visual tracking | Absent auditory response, hypotonia | Facial dysmorphism, short stubby fingers and square set thumb | Hepatosplenomegaly, ascites | N/A | N/A | Cardiomegaly at birth, pulmonary edema |
| Copper A et al32 | 2 (type II) | Caucasian | N/A | N/A | Birth  | 3 months | N/A | Delayed psychomotor development | Coarse facial features,  | Hepatosplenomegaly | Normal CT brain | Left ventricular hypertrophy | Cardiomegaly on ECHO |
| Hale LP et al33 | 1 (type II) | African American | N/A | N/A | In utero | Birth | N/A | N/A | Short first metatarsals, epiphyseal stippling | Non-immune hydrops, ascites, gingival hypertrophy | N/A | N/A | Left ventricular dysfunction, right ventricular enlargement, vacuolation of myocytes on autopsy |
| Paschke et al34 | 1 (type II) | Austrian (Caucasian) | N/A | N/A | 10 weeks of age | 10 months | Normal fundus and corenae | Severe psychomotor retardation, seizures, hypertonia | Pectus excavatum | Hepatosplenomegaly | N/A | Right ventricular hypertrophy | Cardiomegaly, hypertrophic cardiomyopathy |
| Pueschel SM et al35 | 1 (type II) | Caucasian | N/A | N/A | In utero | Birth | N/A | Seizures | Coarse facies, anteverted nostrils, slight gibbus | Hepatosplenomegaly | N/A | N/A | Mild left ventricular dysfunction, diffuse LVH on ECHO; biventricular hypertrophy on autopsy; multiple clear vacuoles in myocardial cells |
| Schleutker J et al36 | 16 (type II) | Caucasian | N/A | N/A | Fetal life to late infancy | Birth to within the first few years of life | N/A | Delayed development, ataxia, seizures, hypotonia | Dysmorphism | Ascites, hepatosplenomegaly | N/A | N/A | Heart failure (2/16) |
| Kang E et al37 | 1 (type II) | East Asian | N/A | N/A | In utero (22 weeks’ gestation) | Birth | N/A | Hypotonia | Coarse facial features, dysostosis multiplex | Ascites, hepatomegaly, congenital diaphragmatic hernia | N/A | N/A | Persistent pulmonary hypertension |
| Huang YZ et al38 | 12 (type I) | East Asian | 10 S182G/S182G (Homozygous); 1 S182G/Q55X (Compound Heterozygous); 1 S182G/A319V (Compound Heterozygous) | N/A | 12-33 yrs | 26-50 yrs | Blurred vision (9/12) | Myoclonus, ataxia (gait, truncal), dysmetria, frequent falls |  |  | MRI: Cerebral atrophy (5/12), Cerebellar atrophy (1/12), Tiny bright dots in basal ganglia (2/12)EEG: Paroxysmal diffuse theta waves (1/12)NCV: Abnormal (6/12) | N/A | N/A |
| Young ID et al39 | 1 (type II) | South Asian | N/A | N/A | 18 months | 3 yrs | Cherry-red spots, extensive bilateral lens opacities | Delayed milestones, grand mal seizures, myoclonic seizures, ataxia, intention tremor, nystagmus, hypotonia, extensor plantar response | Coarse facial features, flattening of lumbar vertebrae, bony abnormalities  | Gingival hypertrophy, | Delayed visually evoked potentials | N/A | N/A |
| Arora V et al40 | 7 (type 2) | South Asian | G679A (p.Gly227Arg) (Exon 4) (Homozygous)  |  | 5 months to 6 yrs | 3-11 yrs | Cherry-red spot (6/7), Bull’s eye maculopathy (1/7) | Developmental delay (7/7), ataxia (2/7), seizures (2/7) | Coarse facial features (7/7), short stature (7/7), contractures (4/7), scoliosis (5/7), dysostosis (4/7) | Hearing loss, (4/7), hepatomegaly (4/7), Splenomegaly (2/7), hernia (1/7), Nephrosialidosis/single kidney (1/7)  | MRI: Cerebral atrophy (1/7) | N/A | N/A |
| Lee YJ et al41 | 1 (type II) | Korean (East Asian) | C239T (p.Pro80Leu) (exon 2) (Homozygous) | Missense | In utero (19 weeks’ gestation) | 3 months | Bilateral congenital cataract with foveal hypoplasia | Hypotonia | Coarse facies | Fetal ascites, hepatosplenomegaly | N/A | N/A | N/A |
| Neeraja K et al42 | 2 (type 1) | South Asian | p.Arg294Cys (Homozygous), p.Arg305Pro (Homozygous) | Missense | N/A | N/A | No cherry-red spot | Ataxia, progressive myoclonus | N/A | N/A | N/A | N/A | N/A |
| Caciotti A et al43 | 2 (type II)12 (type II) juvenile-onset from other studies18 (type II) neonatal-onset from other studies | Italian | G679A (p.G227R) (Homozygous); splicing transition G807 + 1A (Homozygous) | Missense, splicing translation | Birth to 1 year | 1-9 yrs | Cherry-red spot (Infantile onset), cataract | Developmental delay, myoclonic jerks | Coarse facies, dysostosis multiplex  | Hepatosplenomegaly, renal anomaliesNeonatal onset: ascites, edema, hydrops | N/A | N/A | Cardiac anomalies (1)Juvenile-onset type II (3/12) cardiac abnormalities Neonatal-onset type II (6/18) cardiac abnormalities |
| Godra A et al44 | 1 (type II) | N/A | N/A | N/A | In utero | Birth | N/A | N/A | N/A | Ascites, hepatomegaly | N/A | N/A | Enlarged ventricles, mild decrease in LV function, large PDA, cardiac dilation, vacuoles in myocytes |
| Chen CM et al45 | 2 (type 1) | East Asian | (A544G) (p.Ser182Gly) exon 3 (Homozygous) | Missense | 14-17 yrs | N/A | Optic atrophy | Ataxia, myoclonus, nystagmus | N/A | N/A | MRI: Mild cerebellar atrophy | N/A | N/A |
| Steinman L et al46 | 1 (type 1) | Mixed East Asian and Portuguese  | N/A | N/A | 20 yrs | N/A | Cherry-red spot | Myoclonus, dysphagia, dysarthria | Clinodactyly of terminal phalanx of the fifth finger, mild clubbing of fingers  | N/A | EEG: Abnormal; NCV: Abnormal; Visually evoked potentials: Abnormal | N/A | N/A |
| Varela MD et al47 | 7 (type 1) | N/A | N/A | N/A | 27.5+9.8 yrs |  | Lens opacities (8/8), optic atrophy (2/8), macular cherry-red spot (8/8), visual disability (2/8) | N/A | N/A | N/A | N/A | N/A | N/A |
| Palmeri S et al48 | 1 (type I) | Italian | N/A | N/A | 17 yrs | 17 yrs | Macular cherry-red spot | Ataxia, myoclonus, generalized epilepsy, nystagmus, dysarthria | N/A | N/A | MRI: Cerebellar atrophy EEG: Spikes preceding the myoclonus | N/A | N/A |
| Vieira de Rezende Pinto WB et al49 | 2 (1 type 1, 1 type 2) | Brazilian | N/A | N/A | 17, 23 yrs | 27, 30  | Bilateral macular cherry-red spots | Ataxia, slurred speech, dysarthria, dysmetria, hyperreflexia, myoclonic seizures | N/A | N/A | MRI: cerebral atrophy | N/A | N/A |
| Michalewska Z et al50 | 1 (type 1) | Polish | N/A | N/A | 15 yrs | 20 yrs | Bilateral macular cherry-red spot | Myoclonus, ataxia, generalized tonic-clonic and partial seizures, dystonia | N/A | N/A | MRI: Normal; EEG: discharges of sharp waves and diffuse theta waves | N/A | N/A |
| Heroman WJ et al51 | 1 (type 1) | Caucasian | N/A | N/A | 14 yrs | 14 yrs | Bilateral macular cherry-red spot | N/A | N/A | N/A | N/A | N/A | N/A |
| Rosenberg R et al52 | 2 (type II) | French | N/A | N/A | 3,5 yrs | N/A | Bilateral macular cherry-red spot | Hypotonia | Dysostosis | Hepatosplenomegaly, hearing loss, nystagmus | N/A | N/A | N/A |
| Ganguly S et al53 | 1 (type 1) | South Asian | N/A | N/A | 10 yrs | 12 yrs | Bilateral macular cherry-red spot | Myoclonus | N/A | N/A | N/A | N/A | N/A |
| Kersten HM et al54 | 1 (type 1) | Samoan | N/A | N/A | 22 yrs | 32 yrs | Bilateral macular cherry-red spot | Myoclonus, generalized tonic-clonic seizures | N/A | N/A | N/A | N/A | N/A |
| CoppolaA et al55 | 4 (type 1) | 3 Italian1 Turkish | G982A (p.Gly328Ser, rs534846786), deletion c.1208delG (p.Ser403ThrfsTer85, rs1301852124) (Compound Heterozygous)T272G (p.Leu91Arg, rs104893972) G982A (p.Gly328Ser, rs534846786) (Compound Heterozygote)G914A (p.Arg305His, rs774362886) deletion of a nucleotide c.625delG, which determines a frameshift mutation p.Glu209SerfsTer94 (Compound Heterozygote)c.1208delG (p.Ser403ThrfsTer85, rs1301852124) and G982A (p.Gly328Ser, rs534846786) (Compound Heterozygote) | Missense, frameshift and deletion | 12-17 yrs | 18-43 yrs | Bilateral cataract, cherry-red spots | Ataxia, nystagmus, dysarthria, dysmetria, myoclonus | N/A | N/A | MRI Brain: Cortical atrophy, cerebellar atrophy | N/A | N/A |

MRI=Magnetic resonance imaging; EEG=Electroencephalography; ECHO=Echocardiography; IQ=Intelligence quotient; LV=Left ventricle; LVH=Left ventricular hypertrophy; PDA=Patent ductus arteriosus; NCV=Nerve conduction velocity

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