**Supplementary Table 1.** Final diagnosis and clinical classification of patients participated in the study

| Patient no. | Final diagnosis | Classification |
|---|---|---|
| 1 | Startgart syndrome | Ophthalmologic |
| 2 | Neurofibromatosis type 1 | Neurocutaneous |
| 3 | Prader-Willi syndrome | Neurologic |
| 4 | Neurofibromatosis type 1 | Neurocutaneous |
| 5 | Cerebrovascular diseases related to *Col4A1* | Others |
| 6 | 8q21.11 microdeletion syndrome | Neurologic |
| 7 | Prader-Willi syndrome | Neurologic |
| 8 | Congenital disorder of glycosylation 1I | Metabolic |
| 9 | Ullrich congenital muscular dystrophy | Muscular |
| 10 | Rett syndrome | Neurologic |
| 11 | Neurofibromatosis type 1 | Neurocutaneous |
| 12 | Rett syndrome | Neurologic |
| 13 | Phenylalanine hydroxylase deficiency | Metabolic |
| 14 | Angelman syndrome | Neurologic |
| 15 | Neurofibromatosis type 1 | Neurocutaneous |
| 16 | Rett syndrome | Neurologic |
| 17 | Neurofibromatosis type 1 | Neurocutaneous |
| 18 | Rett syndrome | Neurologic |
| 19 | Retinitis pigmentosa associated with systemic disease | Ophthalmologic |
| 20 | Retinitis pigmentosa associated with systemic disease | Ophthalmologic |
| 21 | Retinitis pigmentosa associated with systemic disease | Ophthalmologic |
| 22 | Retinitis pigmentosa associated with systemic disease | Ophthalmologic |
| 23 | Duchenne muscular dystrophy | Muscular |
| 24 | Neurofibromatosis type 1 | Neurocutaneous |
| 25 | Rett syndrome | Neurologic |
| 26 | Glutaric aciduria type 1 | Metabolic |
| 27 | Neurofibromatosis type 1 | Neurocutaneous |
| 28 | Neurofibromatosis type 1 | Neurocutaneous |
| 29 | Neurofibromatosis type 1 | Neurocutaneous |
| 30 | Rett syndrome | Neurologic |
| 31 | Rett syndrome | Neurologic |
| 32 | Rett syndrome | Neurologic |
| 33 | Rett syndrome | Neurologic |
| 34 | CHARGE syndrome | Neurologic |
| 35 | Kleefstra syndrome | Neurologic |
| 36 | Tetralogy of Fallot | Others |
| 37 | Stargardt disease | Others |
| 38 | CHARGE syndrome | Neurologic |
| 39 | CHARGE syndrome | Neurologic |
| 40 | CHARGE syndrome | Neurologic |
| 41 | CHARGE syndrome | Neurologic |
| 42 | CHARGE syndrome | Neurologic |
| 43 | Leber optic atrophy | Ophthalmologic |
| 44 | CHARGE syndrome | Neurologic |
| 45 | CHARGE syndrome | Neurologic |
| 46 | CHARGE syndrome | Neurologic |
| 47 | CHARGE syndrome | Neurologic |
| 48 | CHARGE syndrome | Neurologic |
| 49 | Genetic epileptic syndromes with onset in infancy | Neurologic |
| 50 | CHARGE syndrome | Neurologic |
| 51 | Genetic epileptic syndromes with onset in infancy | Neurologic |
| 52 | Congenital muscular dystrophy | Muscular |
| 53 | Congenital muscular dystrophy | Muscular |
| 54 | Phelan-McDermid syndrome | Neurologic |

(Continued to the next page)

**Supplementary Table 1.** Continued

| Patient no. | Final diagnosis | Classification |
|---|---|---|
| 55 | Smith-Magenis syndrome | Neurologic |
| 56 | *ATR-X*-related syndromes | Neurologic |
| 57 | Costello syndrome | Neurologic |
| 58 | Congenital muscular dystrophy | Muscular |
| 59 | Prader-Willi syndrome | Neurologic |
| 60 | Duchenne muscular dystrophy | Muscular |
| 61 | Genetic epileptic syndromes with onset in infancy | Neurologic |
| 62 | Angelman syndrome | Neurologic |
| 63 | Allan-Herndon-Dudley syndrome | Neurologic |
| 64 | Mitochondrial myopathies | Muscular |
| 65 | Duchenne muscular dystrophy | Muscular |
| 66 | Early-onset parkinsonism - intellectual deficit | Neurologic |
| 67 | Stickler syndrome | Others |
| 68 | Lowe syndrome | Neurologic |
| 69 | Congenital myasthenic syndromes | Muscular |
| 70 | CHARGE syndrome | Neurologic |
| 71 | Phenylalanine hydroxylase deficiency | Metabolic |
| 72 | Leber optic atrophy | Ophthalmologic |
| 73 | Phenylalanine hydroxylase deficiency | Metabolic |
| 74 | Kabuki syndrome | Neurologic |
| 75 | Congenital adrenal gland hyperplasia | Metabolic |
| 76 | Genetic epileptic syndromes with onset in infancy | Neurologic |
| 77 | Congenital muscular dystrophy | Muscular |
| 78 | Coffin-Siris syndrome | Neurologic |
| 79 | Duchenne muscular dystrophy | Muscular |
| 80 | CHARGE syndrome | Neurologic |
| 81 | X-linked hypophosphatemia | Metabolic |
| 82 | Neurofibromatosis type 1 | Neurocutaneous |
| 83 | Genetic epileptic syndromes with onset in infancy | Neurologic |
| 84 | Duchenne muscular dystrophy | Muscular |
| 85 | Retinitis pigmentosa associated with systemic disease | Ophthalmologic |
| 86 | Congenital muscular dystrophy | Muscular |
| 87 | Duchenne muscular dystrophy | Muscular |
| 88 | Congenital muscular dystrophy | Muscular |
| 89 | Congenital muscular dystrophy | Muscular |
| 90 | Prader-Willi syndrome | Neurologic |
| 91 | Congenital muscular dystrophy | Muscular |
| 92 | Panhypopituitarism | Metabolic |
| 93 | Lamb Shaffer syndrome | Neurologic |
| 94 | Congenital muscular dystrophy | Muscular |
| 95 | Opsoclonus-myoclonus-ataxia syndrome | Neurologic |
| 96 | Hereditary spastic paraplegia | Neurologic |
| 97 | Sturge-Weber syndrome | Others |
| 98 | Sotos syndrome | Others |
| 99 | Genetic epileptic syndromes with onset in infancy | Neurologic |
| 100 | Wolf-Hirschhorn syndrome | Neurologic |
| 101 | Pompe disease | Muscular |

CHARGE, coloboma, heart defects, atresia choanae, growth retardation, genital abnormalities.