**S1 Table. Phenotype of patients with homozygous *ARHGEF2* mutation.**

| Characteristics and Symptoms | HPO ID[a] | Patient | Patient |
|---|---|---|---|
| Pedigree ID | NA | II.1 | II.2 |
| Origin | NA | Kurd | Kurd |
| Gender | NA | male | male |
| Age at last assessment (years) | NA | 5.9 | 4.5 |
| **Head** | | | |
| (Infantile) congenital microcephaly (OFC < P3) | 0011451 | + | (+) |
| High palate | 0000218 | + | - |
| Downslanted palpebral fissures | 0000494 | + | - |
| Long eyelashes | 0000527 | + | + |
| Bilateral ptosis | 0001488 | + | - |
| Congenital strabismus | 0000487 | + | + |
| Astigmatism | 0000483 | + | + |
| Horizontal pendular nystagmus | 0007811 | - | + |
| Amblyopia | 0000646 | - | + |
| Optic disc pallor | 0000543 | - | + |
| Abnormality of the retinal pigmentation | 0008051 | - | + |
| **Chest** | | | |
| Wide intermamillary distance | 0006610 | + | - |
| **Skeletal** | | | |
| Broad finger | 0001500 | + | - |
| **Skin, Hair** | | | |
| Skin rash | 0000988 | + | - |
| Low posterior hairline | 0002162 | + | - |
| **Neurologic** | | | |
| Intellectual disability, mild (IQ equivalent; years at assessment) | 0001256 | + (68; 1.5) | - |
| Intellectual disability, moderate (IQ equivalent; years at assessment) | 0002342 | + (<50; 2.2) | + (46; 0.6) |
| Delayed speech and language development | 0000750 | + | + |
| Muscular hypotonia | 0001252 | + | - |
| Hypoplasia of the pons | 0012110 | + | + |
| Cerebellar vermis hypoplasia | 0001320 | - | + |
| Abnormal auditory evoked potentials | 0006958 | + | - |
| Abnormality of vision evoked potentials | 0000649 | - | + |

[a]All symptoms are listed according to the nomenclature and the systematics of the OMIM "Clinical Synopsis" and the Human Phenotype Ontology (HPO; http://www.human-phenotype-ontology.org/) according to Robinson et al. 2008.(1) Abbreviations: AR, autosomal recessive; HPO, human phenotype ontology; IQ, intelligence quotient; OFC, occipito-frontal head circumference; NA, not applicable; OMIM, (Online Mendelian Inheritance in Man); P, percentile.

## References

1. Robinson PN, Kohler S, Bauer S, Seelow D, Horn D, Mundlos S. The Human Phenotype Ontology: a tool for annotating and analyzing human hereditary disease. Am J Hum Genet. 2008;83(5):610-5.