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

<table>
<thead>
<tr>
<th>Characteristics and Symptoms</th>
<th>HPO ID</th>
<th>Patient 1</th>
<th>Patient 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pedigree ID</td>
<td>NA</td>
<td>II.1</td>
<td>II.2</td>
</tr>
<tr>
<td>Origin</td>
<td>NA</td>
<td>Kurd</td>
<td>Kurd</td>
</tr>
<tr>
<td>Gender</td>
<td>NA</td>
<td>male</td>
<td>male</td>
</tr>
<tr>
<td>Age at last assessment (years)</td>
<td>NA</td>
<td>5.9</td>
<td>4.5</td>
</tr>
</tbody>
</table>

**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 - +

*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**