

Table 1

|                            |                                                                             |
|----------------------------|-----------------------------------------------------------------------------|
| Variant                    | c.2011C>T, heterozygous                                                     |
|                            | p.R671W                                                                     |
|                            | <i>de novo</i>                                                              |
| Current age                | 15 years old                                                                |
| Gender                     | M                                                                           |
| Family History             | Caucasian, dutch                                                            |
|                            | 27                                                                          |
| Growth                     | (- 2SD)                                                                     |
| Microcephaly               | Borderline microcephaly                                                     |
| Neurological examination   | yes                                                                         |
| Intellectual disability    | Yes, mild ID IQ69                                                           |
| Seizures                   | No                                                                          |
| Behavioral problems        | Autism spectrum disorder, ADHD                                              |
| Ophthalmological anomalies | Congenital nystagmus                                                        |
| Craniofacial dysmorphisms  | Small ears with abnormal helices, ptosis left, epicanthus, diastasis teeth, |
| Urogenal abnormalities     | No                                                                          |
| Skeletal abnormalities     | No                                                                          |