|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Clinical  Family members | **I-2** (Grandma) | **II-2（**Mother**）** | **II-3（**Uncle**）** | **II-4（**Uncle**）** | **III-1（**Proband**）** |
| Age | 72 | 33 | 45 | 39 | 12 |
| Gender | Female | Female | Male | Male | Male |
| Special facies (long face, prominent forehead, ear) | Long face | Long face | Yes | Yes | Yes |
| Joint hyperextension | Yes | Yes | No | No | No |
| Speech disorder | No | No | Yes | No | Yes |
| Intellectual disability | No | No | Yes (Severe) | Yes (Mild) | Yes (moderate) |
| Autism | No | No | Yes | No | Yes |
| Delayed or impaired motor development | No | No | Yes | No | Yes |