**S1 Table. Summary of the medical history and clinical evaluation of the probands.**

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|  | **Male sibling** | **Female sibling** |
| **Age at evaluation** | 33 | 29 |
| **Psychiatric examination** | - Did not sustain eye contact or initiate interaction- Agitation- Unmotivated laughter; inappropriate social contact- Understood simple commands- Difficulty obeying some commands- No imitation- Non-verbal (guttural sounds and screams are present); absence of functional communication- Presented syndromic features and probably ID | - Presented eye contact and initiated interaction in an inappropriate form, quite puerile but with no functionality- Easy (motivated and unmotivated) laughter and collaborative- Understood simple and (some) complex commands- Used imitation- Verbal, simple sentences with substitutions (mostly "r" for "l").- Presented syndromic features and probably ID |
| **Neurological evaluation** | - Kyphoscoliosis- All deep reflexes were symmetric but slightly diminished-Discrete hypotonia-Gait was symmetric with large steps; no alteration of gait stability | - Kyphosis- Exhibited some difficulty standing on one foot- Gait was symmetric with large steps; no alteration of gait stability |