Table 1. Clinical characteristics of the pedigrees.

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| --- | --- |
|  | Value |
| Age (years) | *Mean±sd (range)* |
| Symptom onset | 1.4±1.4 (0-8) |
| Group (years) | *Male (female)* |
| Neonate [0,1) | 2 (0) |
| Infant [1,3) | 5 (3) |
| Child [3,13) | 21 (7) |
| Adolescent [13,18) | 0 (2) |
| Adult [18,) | 2 (3) |
| Size of pedigree | *Number* |
| s=3 (trio) | 31 |
| s=4 (quad) | 12 |
| s=5 (quin) | 1 |
| s=6 | 1 |