**Supplementary Table 1.** A list of all patients included in the publication along with their symptoms and the publications from which the data were taken (n = 52)

|  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Ref.** | **Patient** | **ADCY5 mutation** | **Chorea** | **Myoclonia** | **Dystonia** | **Hypotonia** | **Speech disorders** | **Dyskinesia** | **Intelectuall disability** | **Eye-movement disoders** | **Delayed motor milestones** | **Others** |
| Our Case I.1 | Female | c.1253G>A; p.Arg418Gln | Y | Y | N | N | Y | N | N | N | N | Depression, problems with memory |
| Our Case II.2 | Male | c.1253G>A; p.Arg418Gln | N | N | N | N | N | N | N | N | N | Involuntary movements |
| [1] | male | c.1378A>T; p.Ile460Phe | N | Y | Y | N | Y | Y (facial) | N | N | Y | None |
| [2] | Female | c.1252C>T; p.Arg418Trp | Y | Y | Y | Y | Y | N | N | Y | Y | None |
| [3] | Patient 1, male | c.3045C>A;  p.Asp1015Glu | Y | N | N | Y | Y | N | N | N | Y | None |
| [3] | Patient 2, male | c.3074A>T; p.Glu1025Val | Y | N | Y | Y | Y | N | N | N | Y | None |
| [4] | Patient IV-1, female | c.3086 T>G; p. Met1029Arg | N | Y | Y | N | Y | Y | N | Y | Y | None |
| [4] | Patient III-4, female | c.3086 T>G; p. Met1029Arg | Y | N | Y | N | N | N | N | N | N | None |
| [4] | Patient II-2, female | c.3086 T>G; p. Met1029Arg | N | Y | N | N | Y | Y (facial) | N | Y | N | Titubation |
| [4] | Patient IV-2, female | c.3086 T>G; p. Met1029Arg | N | N | Y | N | Y | Y | N | N | N | None |
| [5] | Patient 1, male | c.1252 C>T; p.Arg418Trp | Y | N | N | N | N | N | N | N | Y | Nocturnal episodes, sleep problems, facial myokymia |
| [6] | female | p .1253 G>A; p. Arg418Gln | Y | N | N | N | Y | N | N | N | N | Episodes of chorea often after waking up, Deep tendon reflexes were brisk in all four limbs. |
| [7] | male | c.1252C>T; p.Arg418Trp | Y | N | Y | Y | N | Y | N | N | Y | None |
| [8] | Patient 1, female | c.1252C>T; p.Arg418Trp | Y | N | Y | Y | Y | N | N | N | Y | Language delay |
| [8] | Patient 2, male | c.1253G>A; p.Arg418Gln | Y | Y | Y | N | Y | Y | N | N | Y | None |
| [8] | Patient 3, male | c.1252C>G; p.Arg418Glu | Y | N | Y | Y | N | N | N | N | Y | Language delay |
| [8] | Patient 4, male | c.1252C>G; p.Arg418Glu | Y | N | Y | N | N | Y | N | N | Y | Language delay |
| [8] | Patient 5, female | c.1252C>T; p.Arg418Trp | Y | N | Y | Y | N | Y | Y | N | Y | Language delay |
| [8] | Patient 6, male | c.1252C>T; p.Arg418Trp | Y | N | Y | Y | N | N | N | N | Y | Language delay |
| [9] | Male | c.1252C>T; p.Arg418Trp | Y | Y | Y | Y | Y | Y (facial) | Y | N | N | DTR increased |
| [10] | Patient IV-2, female | c.1762G>A; p.Asp588Asn | N | Y | Y | Y | Y | Y (facial) | N | N | Y | Language delay |
| [10] | Patient IV-3, female | c.1762G>A; p.Asp588Asn | N | Y | Y | Y | Y | Y (facial) | N | N | Y | Language delay |
| [10] | Patient IV-5, male | c.1762G>A; p.Asp588Asn | N | N | Y | Y | Y | Y (facial) | N | N | Y | Tremor |
| [10] | Patient IV-6, male | c.1762G>A; p.Asp588Asn | N | N | Y | Y | Y | Y (facial) | N | N | N | None |
| [10] | Patient IV-7, female | c.1762G>A; p.Asp588Asn | N | Y | Y | Y | Y | Y (facial) | N | N | N | None |
| [10] | Patient IV-8, male | c.1762G>A; p.Asp588Asn | N | N | Y | Y | Y | Y (facial) | N | N | Y | Language delay |
| [11] | male | c.1252C>T; p.Arg418Trp | Y | N | Y | N | N | Y | N | N | N | Nocturnal episodes |
| [12] | Patient 1, female | c.3712C>T; p.Arg1238Trp | N | N | Y | Y | Y | N | Y | N | Y | Deep tendon reflexes exaggerated |
| [12] | Patient 2, male | c.3712C>T; p.Arg1238Trp | N | N | Y | Y | Y | N | Y | N | Y | Hypoplasia of brainsteam |
| [13] | Patient 1, female | c.2176G>A; p.Ala726Thr | N | Y | Y | N | N | N | N | N | N | None |
| [13] | Patient 2 , female | c.2176G>A; p.Ala726Thr | Y | Y | N | N | N | Y (facial) | N | N | N | None |
| [13] | Patient 3, male | c.1252C>T; p.Arg418Thr | Y | Y | Y | N | N | Y (facial) | Y | N | N | None |
| [13] | Patient 4, male | c.1253G>A; p.Arg418Gln | Y | Y | Y | N | N | Y (facial) | Y | N | N | None |
| [13] | Patient 5, female | c.1252C>T; p.Arg418Thr | Y | Y | Y | N | N | Y (facial) | Y | N | N | None |
| [14] | Patient 1, male | c.1253G>A; p.Arg418Glu | Y | Y | N | Y | N | Y (facial) | Y | N | N | None |
| [14] | Patient 2, male | c. 1948-11\_ 1948-2del | Y | N | N | Y | N | N | N | N | Y | Language delay |
| [15] | Patient 1, Female | c.3037C>T; p.Arg1013Cys | N | Y | Y | N | N | N | N | N | N | Delayed speech development |
| [15] | Patient 2, male | c.3037C>T; p.Arg1013Cys | N | Y | Y | N | N | N | N | N | N | Delayed speech development |
| [16] | Male | c.1253G>A; p.Arg418Gln | Y | N | Y | N | N | Y (facial) | N | N | Y | Mental health problems |
| [17] | Patient 1, female | c.2722G>A; p.Glu908Lys | N | N | Y | Y | N | N | N | N | Y | Hyperreflexia |
| [17] | Patient 2, female | c.1252C>T; p.Arg418Trp | Y | N | N | Y | N | Y | N | N | Y | Hyperreflexia |
| [17] | Patient 3, female | c.1253G>A; p.Arg418Gln | Y | Y | Y | N | Y | Y (facial) | N | N | N | Anxiety and phobia |
| [18] | Male | c.1252C > T; p.Arg418Trp | Y | N | Y | Y | N | Y (facial) | Y | N | Y | Brisk tendon reflexes, drooling and language delay |
| [19] | male | c.2088G>A; p.Asp956Asn | Y | N | Y | N | N | Y | N | N | N | Diurnal and nocturnal episodes |
| [20] | Patient 4, female | c.1252C>T; p.Arg418Trp | Y | N | Y | Y | Y | N | N | N | Y | Sleep disturbances |
| [21] | Patient 2, female | c.1253G>A; p.Arg418Gln | Y | Y | Y | N | Y | Y (facial) | N | N | N | Nocturnal hyperkinetic attacks |
| [22] | Patient 1, male | c.1252C>T; p.Arg418Trp | Y | N | Y | N | Y | Y | N | Y | N | Sleep problems |
| [22] | Patient 2, male | c.1252C>T; p.Arg418Trp | Y | N | Y | N | N | Y | N | Y | N | Sleep problems |
| [22] | Patient 3, male | c.1252C>T; p.Arg418Trp | Y | N | Y | N | Y | Y | N | Y | N | Sleep problems |
| [23] | Female | c.697T>C; p.Tyr233His | N | N | Y | N | Y | N | N | N | N | Tremors, lower extremity spasticity |
| [24] | male | c.3086T>A; p.Met1029Lys | Y | N | Y | Y | Y | N | Y | N | Y | None |
| [25] | Male | c.1253G>A; p.Arg418Gln | N | Y | Y | N | N | N | N | N | N | None |

Y — symptom present N — symptom absent

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