**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

1. Zech M, Boesch S, Jochim A, et al. Clinical exome sequencing in early-onset generalized dystonia and large-scale resequencing follow-up. Mov Disord. 2017; 32(4): 549–559, doi: [10.1002/mds.26808](http://dx.doi.org/10.1002/mds.26808), indexed in Pubmed: [27666935](https://www.ncbi.nlm.nih.gov/pubmed/27666935).
2. Meijer IA, Miravite J, Kopell BH, et al. Deep brain stimulation in an additional patient with adcy5-related movement disorder. J Child Neurol. 2017; 32(4): 438–439, doi: [10.1177/0883073816681353](http://dx.doi.org/10.1177/0883073816681353), indexed in Pubmed: [27920267](https://www.ncbi.nlm.nih.gov/pubmed/27920267).
3. Westenberger A, Max C, Brüggemann N, et al. Alternating hemiplegia of childhood as a new presentation of adenylate cyclase 5-mutation-associated disease: a report of two cases. J Pediatr. 2017; 181: 306–308.e1, doi: [10.1016/j.jpeds.2016.10.079](http://dx.doi.org/10.1016/j.jpeds.2016.10.079), indexed in Pubmed: [27931826](https://www.ncbi.nlm.nih.gov/pubmed/27931826).
4. Douglas AGL, Andreoletti G, Talbot K, et al. *ADCY5*-related dyskinesia presenting as familial myoclonus-dystonia. Neurogenetics. 2017; 18(2): 111–117, doi: [10.1007/s10048-017-0510-z](http://dx.doi.org/10.1007/s10048-017-0510-z), indexed in Pubmed: [28229249](https://www.ncbi.nlm.nih.gov/pubmed/28229249).
5. Kamate M, Mittal N. *ADCY5*-related dyskinesia. Neurol India. 2018; 66(Supplement): S141–S142, doi: [10.4103/0028-3886.226449](http://dx.doi.org/10.4103/0028-3886.226449), indexed in Pubmed: [29503338](https://www.ncbi.nlm.nih.gov/pubmed/29503338).
6. Holla VV, Neeraja K, Prasad S, et al. ADCY5-related dyskinesia in a child with sleep related paroxysmal dyskinesia. Indian J Pediatr. 2021; 88(3): 308–309, doi: [10.1007/s12098-020-03536-0](http://dx.doi.org/10.1007/s12098-020-03536-0), indexed in Pubmed: [33051786](https://www.ncbi.nlm.nih.gov/pubmed/33051786).
7. Eisenberg H, Malinova V, Mielke D, et al. *ADCY5*‐Induced dyskinetic storm rescued with pallidal deep brain stimulation in a 46‐year‐old man. Movement Disorders Clinical Practice. 2020; 8(1): 142–144, doi: [10.1002/mdc3.13076](http://dx.doi.org/10.1002/mdc3.13076).
8. Carecchio M, Mencacci NE, Iodice A, et al. ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. Parkinsonism Relat Disord. 2017; 41: 37–43, doi: [10.1016/j.parkreldis.2017.05.004](http://dx.doi.org/10.1016/j.parkreldis.2017.05.004), indexed in Pubmed: [28511835](https://www.ncbi.nlm.nih.gov/pubmed/28511835).
9. Miyamoto R, Kawarai T, Takeuchi T, et al. Efficacy of istradefylline for the treatment of *ADCY5*-related disease. Mov Disord Clin Pract. 2020; 7(7): 852–853, doi: [10.1002/mdc3.13067](http://dx.doi.org/10.1002/mdc3.13067), indexed in Pubmed: [33043083](https://www.ncbi.nlm.nih.gov/pubmed/33043083).
10. Bohlega SA, Abou-Al-Shaar H, AlDakheel A, et al. Autosomal recessive ADCY5-Related dystonia and myoclonus: Expanding the genetic spectrum of ADCY5-Related movement disorders. Parkinsonism Relat Disord. 2019; 64: 145–149, doi: [10.1016/j.parkreldis.2019.02.039](http://dx.doi.org/10.1016/j.parkreldis.2019.02.039), indexed in Pubmed: [30975617](https://www.ncbi.nlm.nih.gov/pubmed/30975617).
11. Shetty K, Sarma AS, Devan M, et al. Recurrent ADCY5 mutation in mosaic form with nocturnal paroxysmal dyskinesias and video electroencephalography documentation of dramatic response to caffeine treatment. J Mov Disord. 2020; 13(3): 238–240, doi: [10.14802/jmd.20014](http://dx.doi.org/10.14802/jmd.20014), indexed in Pubmed: [32713175](https://www.ncbi.nlm.nih.gov/pubmed/32713175).
12. Okamoto N, Miya F, Kitai Y, et al. Homozygous ADCY5 mutation causes early-onset movement disorder with severe intellectual disability. Neurol Sci. 2021; 42(7): 2975–2978, doi: [10.1007/s10072-021-05152-y](http://dx.doi.org/10.1007/s10072-021-05152-y), indexed in Pubmed: [33704598](https://www.ncbi.nlm.nih.gov/pubmed/33704598).
13. Tunc S, Brüggemann N, Baaske MK, et al. Facial twitches in ADCY5-associated disease — Myokymia or myoclonus? An electromyography study. Parkinsonism Relat Disord. 2017; 40: 73–75, doi: [10.1016/j.parkreldis.2017.04.013](http://dx.doi.org/10.1016/j.parkreldis.2017.04.013), indexed in Pubmed: [28442302](https://www.ncbi.nlm.nih.gov/pubmed/28442302).
14. Padmanabha H, Ray S, Mahale R, et al. ADCY5-related dyskinesia: a genetic cause of early-onset chorea-report of two cases and a novel mutation. Ann Indian Acad Neurol. 2021; 24(5): 837–838, doi: [10.4103/aian.AIAN\_1012\_20](http://dx.doi.org/10.4103/aian.AIAN_1012_20), indexed in Pubmed: [35002175](https://www.ncbi.nlm.nih.gov/pubmed/35002175).
15. Barrett MJ, Williams ES, Chambers C, et al. Autosomal recessive inheritance of *ADCY5*-related generalized dystonia and myoclonus. Neurol Genet. 2017; 3(5): 193, doi: [10.1212/NXG.0000000000000193](http://dx.doi.org/10.1212/NXG.0000000000000193), indexed in Pubmed: [28971144](https://www.ncbi.nlm.nih.gov/pubmed/28971144).
16. Vijiaratnam N, Newby R, Kempster PA. Depression and psychosis in ADCY5-related dyskinesia-part of the phenotypic spectrum? J Clin Neurosci. 2018; 57: 167–168, doi: [10.1016/j.jocn.2018.08.049](http://dx.doi.org/10.1016/j.jocn.2018.08.049), indexed in Pubmed: [30172639](https://www.ncbi.nlm.nih.gov/pubmed/30172639).
17. Waalkens AJE, Vansenne F, van der Hout AH, et al. Expanding the phenotype toward spastic paraparesis: A mutation in the M2 domain. Neurol Genet. 2018; 4(1): e214, doi: [10.1212/NXG.0000000000000214](http://dx.doi.org/10.1212/NXG.0000000000000214), indexed in Pubmed: [29473048](https://www.ncbi.nlm.nih.gov/pubmed/29473048).
18. Nosadini M, D'Onofrio G, Pelizza M, et al. Sleep exacerbations and facial twitching: diagnostic clues for ADCY5-related dyskinesias. Neuropediatrics. 2020; 52(03): 208–211, doi: [10.1055/s-0040-1721685](http://dx.doi.org/10.1055/s-0040-1721685).
19. Méneret A, Gras D, McGovern E, et al. Caffeine and the dyskinesia related to mutations in the ADCY5 gene. Ann Intern Med. 2019; 171(6): 439, doi: [10.7326/L19-0038](http://dx.doi.org/10.7326/L19-0038), indexed in Pubmed: [31181574](https://www.ncbi.nlm.nih.gov/pubmed/31181574).
20. Zech M, Jech R, Wagner M, et al. Molecular diversity of combined and complex dystonia: insights from diagnostic exome sequencing. Neurogenetics. 2017; 18(4): 195–205, doi: [10.1007/s10048-017-0521-9](http://dx.doi.org/10.1007/s10048-017-0521-9), indexed in Pubmed: [28849312](https://www.ncbi.nlm.nih.gov/pubmed/28849312).
21. Blumkin L, Lerman-Sagie T, Westenberger A, et al. Multiple causes of pediatric early onset chorea-clinical and genetic approach. Neuropediatrics. 2018; 49(4): 246–255, doi: [10.1055/s-0038-1645884](http://dx.doi.org/10.1055/s-0038-1645884), indexed in Pubmed: [29801190](https://www.ncbi.nlm.nih.gov/pubmed/29801190).
22. Balint B, Antelmi E, Mencacci NE, et al. Oculomotor apraxia and disrupted sleep with nocturnal ballistic bouts in ADCY5-related disease. Parkinsonism Relat Disord. 2018; 54: 103–106, doi: [10.1016/j.parkreldis.2018.04.011](http://dx.doi.org/10.1016/j.parkreldis.2018.04.011), indexed in Pubmed: [29680308](https://www.ncbi.nlm.nih.gov/pubmed/29680308).
23. Dean M, Messiaen L, Cooper GM, et al. Child Neurology: Spastic paraparesis and dystonia with a novel *ADCY5* mutation. Neurology. 2019; 93(11): 510–514, doi: [10.1212/WNL.0000000000008089](http://dx.doi.org/10.1212/WNL.0000000000008089), indexed in Pubmed: [31501304](https://www.ncbi.nlm.nih.gov/pubmed/31501304).
24. Wali GM, Wali G, Kumar KR, et al. Long-term follow-up and evolution of *ADCY5*-from a ballistic to dystonic phenotype. Mov Disord Clin Pract. 2020; 7(8): 985–986, doi: [10.1002/mdc3.13069](http://dx.doi.org/10.1002/mdc3.13069), indexed in Pubmed: [34919601](https://www.ncbi.nlm.nih.gov/pubmed/34919601).
25. Agarwal PA, Ramprasad VL. Adult-onset myoclonus-dystonia syndrome preceding characteristic facial myoclonus in indian -related dyskinesia. Mov Disord Clin Pract. 2019; 6(3): 267–268, doi: [10.1002/mdc3.12733](http://dx.doi.org/10.1002/mdc3.12733), indexed in Pubmed: [33999972](https://www.ncbi.nlm.nih.gov/pubmed/33999972).