

Supplementary Materials

1. Al-Futaisi AM, Al-Kindi MN, Al-Mawali AM, Koul RL, Al-Adawi S, Al-Yahyaee SA. Novel mutation of GLRA1 in Omani families with hyperekplexia and mild mental retardation. *Pediatr Neurol* 2012;46:89-93.
2. Becker K, Hohoff C, Schmitt B, Christen HJ, Neubauer BA, Sandrieser T, et al. Identification of the microdeletion breakpoint in a GLRA1null allele of Turkish hyperekplexia patients. *Hum Mutat* 2006;27:1061-1062.
3. Brune W, Weber RG, Saul B, von Knebel Doeberitz M, Grond-Ginsbach C, Kellerman K, et al. A GLRA1 null mutation in recessive hyperekplexia challenges the functional role of glycine receptors. *Am J Hum Genet* 1996; 58:989-997.
4. Chan KK, Cherk SW, Lee HH, Poon WT, Chan AY. Hyperekplexia: a Chinese adolescent with 2 novel mutations of the GLRA1 gene. *J Child Neurol* 2014;29:111-113.
5. Coto E, Armenta D, Espinosa R, Argente J, Castro MG, Alvarez V. Recessive hyperekplexia due to a new mutation (R100H) in the GLRA1 gene. *Mov Disord* 2005;20:1626-1629.
6. del Giudice EM, Coppola G, Bellini G, Cirillo G, Scuccimarra G, Pascotto A. A mutation (V260M) in the middle of the M2 pore-lining domain of the glycine receptor causes hereditary hyperekplexia. *Eur J Hum Genet* 2001;9:873-876.
7. Doria Lamba L, Giribaldi G, De Negri E, Follo R, De Grandis E, Pintaudi M, et al. A case of major form familial hyperekplexia: prenatal diagnosis and effective treatment with clonazepam. *J Child Neurol* 2007;22:769-772.
8. Forsyth RJ, Gika AD, Ginjaar I, Tijssen MA. A novel GLRA1 mutation in a recessive hyperekplexia pedigree. *Mov Disord* 2007;22:1643-1645.
9. Gregory ML, Guzauskas GF, Edgar TS, Clarkson KB, Srivastava AK, Holden KR. A novel GLRA1 mutation associated with an atypical hyperekplexia phenotype. *J Child Neurol* 2008;23:1433-1438.
10. Hayashi T, Tachibana H, Kajii T. Hyperekplexia: pedigree studies in two families. *Am J Med Genet* 1991;40:138-143.
11. Hmami F, Wood SE, Chaouki S, Oulmaati A, Hida M, Rees MI, et al. Neonatal hyperekplexia with homozygous p.R392H mutation in GLRA1. *Epileptic Disord* 2014;16:354-357.
12. Horváth E, Farkas K, Herczegfalvi A, Nagy N, Széll M. Identification of a novel missense GLRA1 gene mutation in hyperekplexia: a case report. *J Med Case Rep* 2014;8:233.
13. Humeny A, Bonk T, Becker K, Jafari-Boroujerdi M, Stephani U, Reuter K, et al. A novel recessive hyperekplexia allele GLRA1 (S231R): genotyping by MALDI-TOF mass spectrometry and functional characterisation as a determinant of cellular glycine receptor trafficking. *Eur J Hum Genet* 2002;10:188-196.
14. Jungbluth H, Rees MI, Manzur AY, Mercuri E, Sewry CA, Gobbi P, et al. An unusual case of hyperekplexia. *Eur J Paediatr Neurol* 2000;4:77-80.
15. Kang HC, You SJ, Chey MJ, Baik JS, Kim JW, Ki CS. Identification of a de novo Lys304Gln mutation in the glycine receptor alpha-1 subunit gene in a Korean infant with hyperekplexia. *Mov Disord* 2008;23: 610-613.
16. Kurczynski TW. Hyperekplexia. *Arch Neurol* 1983;40:246-248.
17. Kwok JB, Raskin S, Morgan G, Antoniuk SA, Bruk I, Schofield PR. Mutations in the glycine receptor alpha1 subunit (GLRA1) gene in hereditary hyperekplexia pedigrees: evidence for non-penetrance of mutation Y279C. *J Med Genet* 2001;38:E17.
18. Lee CG, Kwon MJ, Yu HJ, Nam SH, Lee J, Ki CS, et al. Clinical features and genetic analysis of children with hyperekplexia in Korea. *J Child Neurol* 2013;28:90-94.
19. Milani N, Dalprá L, del Prete A, Zanini R, Larizza L. A novel mutation (Gln266-->His) in the alpha 1 subunit of the inhibitory glycine-receptor gene (GLRA1) in hereditary hyperekplexia. *Am J Hum Genet* 1996;58: 420-422.
20. Mine J, Taketani T, Yoshida K, Yokochi F, Kobayashi J, Maruyama K, et al. Clinical and genetic investigation of 17 Japanese patients with hyperekplexia. *Dev Med Child Neurol* 2015;57:372-377.
21. Miraglia Del Giudice E, Coppola G, Bellini G, Ledaal P, Hertz JM, Pascootto A. A novel mutation (R218Q) at the boundary between the N-terminal and the first transmembrane domain of the glycine receptor in a case of sporadic hyperekplexia. *J Med Genet* 2003;40:e71.
22. Morley DJ, Weaver DD, Garg BP, Markand O. Hyperexplexia: an inherited disorder of the startle response. *Clin Genet* 1982;21:388-396.
23. Poon WT, Au KM, Chan YW, Chan KY, Chow CB, Tong SF, et al. Novel missense mutation (Y279S) in the GLRA1 gene causing hyperekplexia. *Clin Chim Acta* 2006;364:361-362.
24. Rees MI, Andrew M, Jawad S, Owen MJ. Evidence for recessive as well as dominant forms of startle disease (hyperekplexia) caused by mutations in the alpha 1 subunit of the inhibitory glycine receptor. *Hum Mol Genet* 1994;3:2175-2179.
25. Ryan SG, Sherman SL, Terry JC, Sparkes RS, Torres MC, Mackey RW. Startle disease, or hyperekplexia: response to clonazepam and assignment of the gene (STHE) to chromosome 5q by linkage analysis. *Ann Neurol* 1992;31:663-668.
26. Sirén A, Legros B, Chahine L, Misson JP, Pandolfo M. Hyperekplexia in Kurdish families: a possible GLRA1 founder mutation. *Neurology* 2006; 67:137-139.
27. Tijssen MA, Brown P, MacManus D, McLean MA, Davie C. Magnetic resonance spectroscopy of cerebral cortex is normal in hereditary hyperekplexia due to mutations in the GLRA1 gene. *Mov Disord* 2003;18: 1538-1541.
28. Tsai CH, Chang FC, Su YC, Tsai FI, Lu MK, Lee CC, et al. Two novel mutations of the glycine receptor gene in a Taiwanese hyperekplexia family. *Neurology* 2004;63:893-896.
29. Vergouwe MN, Tijssen MA, Peters AC, Wielanda R, Frants RR. Hyperekplexia phenotype due to compound heterozygosity for GLRA1 gene mutations. *Ann Neurol* 1999;46:634-638.
30. Zoons E, Ginjaar IB, Bouma PA, Carpay JA, Tijssen MA. A new hyperekplexia family with a recessive frameshift mutation in the GLRA1 gene. *Mov Disord* 2012;27:795-796.