

## REFERENCES

1. Zhou XY, Wu JJ, Sun YM. An atypical case of early-onset dystonia with a novel missense variant in KMT2B. *Parkinsonism Relat Disord* 2019;63:224-226.
2. Kawarai T, Miyamoto R, Nakagawa E, Koichihara R, Sakamoto T, Mure H, et al. Phenotype variability and allelic heterogeneity in KMT2B-associated disease. *Parkinsonism Relat Disord* 2018;52:55-61.
3. Dai L, Ding C, Fang F. An inherited KMT2B duplication variant in a Chinese family with dystonia and/or development delay. *Parkinsonism Relat Disord* 2019;63:227-228.
4. Miyata Y, Hamanaka K, Kumada S, Uchino S, Yokochi F, Taniguchi M, et al. An atypical case of KMT2B-related dystonia manifesting asterixis and effect of deep brain stimulation of the globus pallidus. *Neurol Clin Neurosci* 2020;8:36-38.
5. Cao Z, Yao H, Bao X, Wen Y, Liu B, Wang S, et al. DYT28 responsive to pallidal deep brain stimulation. *Mov Disord Clin Pract* 2019;7:97-99.
6. Horisawa S, Azuma K, Akagawa H, Nonaka T, Kawamata T, Taira T. Radiofrequency ablation for DYT-28 dystonia: short term follow-up of three adult cases. *Ann Clin Transl Neurol* 2020;7:2047-2051.
7. Pandey S, Bhattad S, Panda AK, Mahadevan L. Late-onset KMT2B-related dystonia in an Indian patient with normal cognition, dystonic opisthotonus and lack of oromandibular and laryngeal involvement. *Parkinsonism Relat Disord* 2020;74:33-35.
8. Mun JK, Kim AR, Ahn JH, Kim M, Cho JW, Lee JI, et al. Successful pallidal stimulation in a patient with KMT2B-related dystonia. *J Mov Disord* 2020;13:154-158.
9. Li XY, Dai LF, Wan XH, Guo Y, Dai Y, Li SL, et al. Clinical phenotypes, genotypes and treatment in Chinese dystonia patients with KMT2B variants. *Parkinsonism Relat Disord* 2020;77:76-82.
10. Aksoy A, Yaylı Köken Ö, Ceylan AC, Toptaş Dedeoğlu Ö. KMT2B-related dystonia: challenges in diagnosis and treatment. *Mol Syndromol* 2022;13:159-164.
11. Kwong AK, Tsang MH, Fung JL, Mak CC, Chan KL, Rodenburg RJT, et al. Exome sequencing in paediatric patients with movement disorders. *Orphanet J Rare Dis* 2021;16:32.
12. Rajan R, Garg K, Saini A, Kumar M, Binukumar BK, Scaria V, et al. Pallidal deep brain stimulation for KMT2B related dystonia in an Indian patient. *Ann Indian Acad Neurol* 2021;24:586-588.
13. Grosz BR, Tisch S, Tchan MC, Fung VSC, Darveniza P, Fellner A, et al. A novel synonymous KMT2B variant in a patient with dystonia causes aberrant splicing. *Mol Genet Genomic Med* 2022;10:e1923.
14. Wu MC, Chang YY, Lan MY, Chen YF, Tai CH, Lin YF, et al. A clinical and integrated genetic study of isolated and combined dystonia in Taiwan. *J Mol Diagn* 2022;24:262-273.
15. Shimazaki R, Ikezawa J, Okiyama R, Azuma K, Akagawa H, Takahashi K. Dystonic tremor in adult-onset DYT-KMT2B. *Intern Med* 2022;61:2357-2360.
16. Padmanabha H, Awati AM, Thomas K, Sarma GRK. A novel mutation in KMT2B gene causing childhood-onset generalized dystonia with expanded phenotype from India. *Neurol India* 2021;69:1400-1401.
17. Buzo EL, De la Casa-Fages B, Sánchez MG, Sánchez JRP, Carballal CF, Vidorreta JG, et al. Pallidal deep brain stimulation response in two siblings with atypical adult-onset dystonia related to a KMT2B variant. *J Neurol Sci* 2022;438:120295.
18. Feuerstein JS, Taylor M, Kwak JJ, Berman BD. Parkinsonism and positive dopamine transporter imaging in a patient with a novel KMT2B variant. *Mov Disord Clin Pract* 2021;8:279-281.
19. Damásio J, Santos M, Samões R, Araújo M, Macedo M, Sardoeira A, et al. Novel KMT2B mutation causes cerebellar ataxia: expanding the clinical phenotype. *Clin Genet* 2021;100:743-747.
20. Owczarzak LR, Hogan KE, Dineen RT, Gill CE, Li MH. A new pathologic KMT2B variant associated with childhood onset dystonia presenting as variable phenotypes among family members. *Tremor Other Hyperkin Mov (N Y)* 2022;12:7.
21. Ng A, Galosi S, Salz L, Wong T, Schwager C, Amudhavalli S, et al. Failure to thrive - an overlooked manifestation of KMT2B-related dystonia: a case presentation. *BMC Neurol* 2020;20:246.
22. Winslow N, Maldonado A, Zayas-Rodríguez L, Lamichhane D. Adult-onset KMT2B-related dystonia responsive to deep brain stimulation. *Mov Disord Clin Pract* 2020;7:992-993.
23. Ciolfi A, Foroutan A, Capuano A, Pedace L, Travaglini L, Pizzi S, et al. Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. *Clin Epigenetics* 2021;13:157.
24. Mirza-Schreiber N, Zech M, Wilson R, Brunet T, Wagner M, Jech R, et al. Blood DNA methylation provides an accurate biomarker of KMT2B-related dystonia and predicts onset. *Brain* 2022;145:644-654.
25. Cif L, Demailly D, Lin JP, Barwick KE, Sa M, Abela L, et al. KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. *Brain* 2020;143:3242-3261.