

REFERENCES

1. Hallervorden J, Spatz H. Eigenartige Erkrankung im extrapyramidalen System mit besonderer Beteiligung des Globus pallidus und der Substantia nigra. Z Gesammte Neurolog Psychiatr 1922;79:254–302.
2. Hogarth P. Neurodegeneration with brain iron accumulation: diagnosis and management. J Mov Disord. 2015;8(1):1-13.
3. Zhou B, Westaway SK, Levinson B, Johnson MA, Gitschier J, Hayflick SJ. A novel pantothenate kinase gene (PANK2) is defective in Hallervorden-Spatz syndrome. Nat Genet 2001;28:345–349.
4. Shevell M. Hallervorden and history. N Engl J Med 2003; 348:3-4.
5. Thomas M, Hayflick SJ, Jankovic J. Clinical heterogeneity of neurodegeneration with brain iron accumulation (Hallervorden-Spatz syndrome) and pantothenate kinase-associated neurodegeneration. Mov Disord; Jan 2004; 19(1):36-42.
6. Morgan, N. V., Westaway, S. K., Morton, J. E. V., Gregory et.al. PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. Nature Genet. 38: 752-754, 2006. Note: Erratum: Nature Genet 2006. 38: 957.

7. Alazami A. M., Al-Saif A., Al-Semari A., Bohlega S., Zlitni S., Alzahrani F. et al. 2008 Mutations in C2orf37, encoding a nucleolar protein, cause hypogonadism, alopecia, diabetes mellitus, mental retardation, and extrapyramidal syndrome. *Am. J. Hum. Genet.* 83, 684–691.
8. Schenck JF, Zimmerman EA. High-field magnetic resonance imaging of brain iron: birth of a biomarker? *NMR Biomed* 2004;17: 433–445.
9. Levi S, Finazzi D. Neurodegeneration with brain iron accumulation: update on pathogenic mechanisms. *Fron in Pharm* 2014;Vol 5: Article 99:1-20
10. Arber C.E, Li A, Houlden H, Wray.S. Review: Insights into molecular mechanisms of disease in neurodegeneration with brain iron accumulation: unifying theories. *Neuropathol and App Neurobiol* 2016; 42: 220–241.
11. Ramonet D, Podhajska A, Stafa K, Sonnay S, Trancikova A, Tsika E, Pletnikova O, Troncoso JC, Glauser L, Moore DJ. PARK9-associated ATP13A2 localizes to intracellular acidic vesicles and regulates cation homeostasis and neuronal integrity. *Hum Mol Genet* 2012; 21: 1725–43.
12. Hayflick SJ, Westaway SK, Ching KH, Gitschier Jet.al. Genetic, clinical, and radiographic delineation of Hallervorden-Spatz syndrome. *N Engl J Med* 2003;348:33-40.

13. Petrović, I. N., Kresojević, N. , Ganos, C. , Svetel, M. , Dragašević, N. , Bhatia, K. P. and Kostić, V. S. (2014), Characteristic “Forcible” Geste Antagoniste in Oromandibular Dystonia Resulting From Pantothenate Kinase-Associated Neurodegeneration. *Mov Disord Clin Pract*, 1: 112-114.
14. Stamelou M, Lai SC, Aggarwal A, Schneider SA, Houlden H, Yeh TH, et al. Dystonic opisthotonus: a “red flag” for neurodegeneration with brain iron accumulation syndromes? *MovDisord* 2013;28:1325-1329.
15. Kurian MA, Mcneill A, Lin, J-P, Maher E. R. Childhood disorders of neurodegeneration with brain iron accumulation (NBIA). *Developmental Medicine & Child Neurology* 2011; 53: 394–404.
16. Brugger F, Kagi G, Pandolfo M, Mencacci N, Batla A, Wiethoff S, Bhatia KP. Neurodegeneration with brain iron accumulation (NBIA) syndromes presenting with late onset craniocervical dystonia: An illustrative case series. *MovDisordClinPract* 2016; doi:10.1002
17. Aggarwal A, Schneider SA, Houlden H, Silverdale M, Paudel R, Paisan-Ruiz C, et al. Indian-subcontinent NBIA: Unusual phenotypes, novel PANK2 mutations, and undetermined genetic forms. *MovDisord* 2010;25:1424-31.
18. Lee JH, Park J, Ryu HS, Park H, Kim YE, Hong JY, et al. Clinical heterogeneity of atypical pantothenate kinase-associated neurodegeneration in Koreans. *J MovDisord* 2016;9:20–27.

19. Grimes DA, Lang AE, Bergeron C. Late adult onset chorea with typical pathology of HallervordenSpatz syndrome. *Journal of neurology, neurosurgery and psychiatry* 2000;69:392-395.
20. Pellechia MT, Valente EM, Cif L, et.al. The diverse phenotype and genotype of pantothenate kinase associated neurodegeneration. *Neurology* 2005;64:1810-2.
21. Schneider, S. A., Hardy, J. and Bhatia, K. P. Syndromes of neurodegeneration with brain iron accumulation (NBIA): An update on clinical presentations, histological and genetic underpinnings, and treatment considerations. *Mov.Disord* 2012; 27: 42–53.
22. Gregory A, Polster BJ, Hayflick SJ. Clinical and genetic delineation of neurodegeneration with brain iron accumulation. *J Med Genet* 2009; 46: 73–80.
23. Ching KH, Westaway SK, Gitschier J, Higgins JJ, Hayflick SJ. HARP syndrome is allelic with pantothenate kinase associated neurodegeneration. *Neurology* 2002;58:1673-4.
24. Paisan-Ruiz C, Bhatia KP, Li A, Hernandez D, Davis M, Wood NW, et al. Characterization of PLA2G6 as a locus for dystonia-parkinsonism. *Ann Neurol*. 2009;65:19–23.
25. Schneider SA, Bhatia KP. Excess iron harms the brain: the syndromes of neurodegeneration with brain iron accumulation (NBIA). *J Neural Transm* 2013;120:695-703.

26. Kurian MA, Hayflick SJ. Pantothenate kinase-associated neurodegeneration (PKAN) and PLA2G6-associates neurodegeneration (PLAN): review of two major neurodegeneration with brain iron accumulation (NBIA) phenotypes. *Int Rev Neurobiol* 2013;110:49-71.
27. Kurian MA, Morgan NV, MacPherson L, et al. Phenotypic spectrum of neurodegeneration associated with mutations in the PLA2G6 gene (PLAN). *Neurology* 2008; 70: 1623–9.
28. Gregory A, Westaway SK, Holm IE, et al. Neurodegeneration associated with genetic defects in phospholipase A(2). *Neurology* 2008; 71: 1402–9.
29. Scheithauer BW, Forno LS, Dorfman LJ, Kane CA. Neuroaxonal dystrophy (Seitelberger's disease) with late onset, protracted course and myoclonic epilepsy. *J NeurolSci* 1978; 36:247–58.
30. Mubaidin, A., Roberts, E., Hampshire, D., Dehyyat, M., Shurbaji, A.,et.al. Karak syndrome: a novel degenerative disorder of the basal ganglia and cerebellum. *J. Med. Genet.* 40: 543-546, 2003.
31. Morgan, N. V., Westaway, S. K., Morton, J. E. V., Gregory, A., et.al. PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. *Nature Genet.* 38: 752-754, 2006.

32. Paisan-Ruiz C, Bhatia KP, Li A, Hernandez D, Davis M, Wood NW, et.al. Characterization of PLA2G6 as a locus for dystonia-parkinsonism. Ann Neurol. 2009 Jan; 65(1):19-23.
33. Erro, R., Balint, B., Kurian, M. A., Brugger, F., Picillo, M., Bhatia, K. P. et.al Early Ataxia and Subsequent Parkinsonism: PLA2G6 Mutations Cause a Continuum Rather Than Three Discrete Phenotypes. 2016 MovmntDisordsClnclPractice.doi: 10.1002/mdc3.12319
34. Shi CH, Tang BS, Wang L, Lv ZY, Wang J, Luo LZ, Shen L et.al PLA2G6 gene mutation in autosomal recessive early-onset parkinsonism in a Chinese cohort. Neurology. 2011 Jul 5; 77(1):75-81
35. Giri A, Guven G, Hanagasi H, Hauser AK et.al. PLA2G6 Mutations Related to Distinct Phenotypes: A New Case with Early-onset Parkinsonism. Tremor Other HyperkinetMov (N Y).2016; 6: 363.
36. Curtis, A. R. J., Fey, C., Morris, C. M., Bindoff, L. A., Ince, P. G., Chinnery, P. F., et.al Mutation in the gene encoding ferritin light polypeptide causes dominant adult-onset basal ganglia disease. Nature Genet 2001; 28: 350-354.
37. Chinnery, P. F., Crompton, D. E., Birchall, D., Jackson, M. J., Coulthard A., et.al Clinical features and natural history of neuroferritinopathy caused by the FTL1 460insA mutation. Brain 2007;130: 110-119.

38. Kumar N, Rizek P, Jog M. Neuroferritinopathy: Pathophysiology, Presentation, Differential Diagnoses and Management. Walker R, ed. Tremor and Other Hyperkinetic Movements. 2016;6:355.
39. Keogh MJ, Singh B, Chinnery PF. Early neuropsychiatry features in neuroferritinopathy. MovDisord. 2013 Aug; 28(9):1310-3.
40. Hartig, M. B., Iuso, A., Haack, T., Kmiec, T., Jurkiewicz, E., Heim, K., Roeber, S., et.al Absence of an orphan mitochondrial protein, C19orf12, causes a distinct clinical subtype of neurodegeneration with brain iron accumulation. Am. J. Hum. Genet.2011; 89: 543-550.
41. Hogarth, P., Gregory, A., Kruer, M. C., Sanford, L., Wagoner, W., Natowicz et.al. New NBIA subtype: genetic, clinical, pathologic, and radiographic features of MPAN. Neurology 2013; 80: 268-275.
42. Haack TB, Hogarth P, Kruer M, Gregory A, Wieland T, Schwarzmayr T, et al. Exome sequencing reveals de novo mutations in WDR45 causing a phenotypically distinct, X-linked dominant form of NBIA. Am J Hum Genet2012; 7:11444-9.
43. Hayflick SJ, Kruer MC, Gregory A, et al. BPAN: a new X-linked dominant form of neurodegeneration with brain iron accumulation. Brain 2013;136:1708–1717.

44. Dusi, S., Valletta, L., Haack, T. B., Tsuchiya, Y., Venco, P., Pasqualato, S. et.al. Exome sequence reveals mutations in CoA synthase as a cause of neurodegeneration with brain iron accumulation. Am. J. Hum. Genet 2014; 94: 11-22.
45. Harris, Z. L., Takahashi, Y., Miyajima, H., Serizawa, M., MacGillivray, R. T. A., Gitlin, J. D. Aceruloplasminemia: molecular characterization of this disorder of iron metabolism. Proc. Nat. Acad. Sci. 1995; 92: 2539-2543.
46. Kono S, Miyajima H. Aceruloplasminemia. In: Rosenberg RN, Pascual JM, eds. Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease. 5 ed. Elsevier, Academic Press. 2015:495-506.
47. Krueger MC, Pisan-Ruiz C, Boddaert N et al. Defective FA2H leads to a novel form of neurodegeneration with brain iron accumulation (NBIA). Ann Neurol. 2010;68:610–618.
48. Pierson TM, Simeonov DR, Sincan M, Adams DA, Markello T, Golas G, et al. Exome sequencing and SNP analysis detect novel compound heterozygosity in fatty acid hydroxylase-associated neurodegeneration. Eur J Hum Genet 2012; 20:476-479.
49. Najim al-Din A, Wriegat A, Mubaidin A, Dasouki M, Hiari M. Pallido-pyramidal degeneration, supranuclear gaze paresis and dementia: Kufor-Rakeb syndrome. Acta Neurol Scand 1994;89: 347-352.

50. Prashanth L.K., Murugan S, Kamath V, Gupta R, Jadav R, Sreekantaswamy S, Ramprasad V.L. First Report of Kufor-Rakeb Syndrome (PARK 9) from India, and a Novel Nonsense Mutation in ATP13A2 Gene. *MovDisord Cl Pract* 2015; Vol2 (3) 326-327.

51. Koshy G, Danda S, Thomas N Mathews V, Viswanathan V. Three siblings with Wood house Sakati syndrome in an Indian family. *Clin Dysmorph* 2008;17:57-60

52. Ferdinandusse S, Kostopoulos P, Denis S, Rusch H, Overmars H, Dillmann U, et al. Mutations in the gene encoding peroxisomal sterol carrier protein X (SCP_X) cause leukencephalopathy with dystonia and motor neuropathy. *Am J Hum Genet* 2006;78(6):1046e52

53. Horvath R, Lewis-Smith D, Douroudis K, Duff J, Keogh M, Pyle A, et al. SCP2 mutations and neurodegeneration with brain iron accumulation. *Neurology* 2015;85(21):1909e11.

54. Jaberi E, Rohani M, Shahidi GA, Nafissi S, Arefian E, Soleimani M, et al. Identification of mutation in GTPBP2 in patients of a family with neurodegeneration accompanied by iron deposition in the brain. *Neurobiol Aging* 2016;38:216

55. Kruer MC , Boddaert N,Scheider SA, Houlden H, Bhatia kP, Gregory A et.al. Neuroimaging Features of Neurodegeneration with Brain Iron Accumulation. *AJNR* 201;33: 407-414.

56. McNeill A, Birchall D, Hayflick S.J., Gregory A, et.al. T2* and FSE MRI distinguishes four subtypes of neurodegeneration with brain iron accumulation. *Neurology* 2008; Apr 29;70(18):1614-9
57. Chang CL, Lin CM. Eye of the tiger sign is not pathognomonic of Pantothenate kinase Associated Neurodegeneration in Adult cases. *Brain Behav* 2011;1:55-66.
58. Batla A, Adams ME, Erro R, Ganos C, et.al. Cortical pencil lining in neuroferritinopathy: a diagnostic clue. *Neurology* 2015 Apr 28;84(17):1816-8.