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Genotype and phenotype analysis of two unrelated patients with Beta-Propeller protein-Associated Neurodegeneration (*BPAN*)

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Beta-propeller protein-associated neurodegeneration (*BPAN*) is a type of neurodegeneration with brain iron accumulation (NBIA) disorders. *BPAN* is characterized by developmental delay, intellectual disability, seizures, absent to limited expressive language, ataxia, Parkinsonism, dystonia and abnormal behaviors like autism spectrum disorder. Mutations of *WDR45* gene cause *BPAN*. This gene encodes a member of the WD-repeat protein family that plays an important role in autophagy. Two unrelated Iranian patients were diagnosed as *BPAN*. First, DNAs were extracted from peripheral blood leukocytes. Subsequently, thirteen exons and flanking intronic sequences of *WDR45* were amplified by PCR and sequenced. Variations were assessed by comparison with reference sequences available at NCBI. Patient-1 was a 24-year-old woman who presented with the progressive slowness of movements, episodes of generalized tonic-clonic seizures, developmental delay, the absence of expressive language, and severe psychomotor retardation. Neurologic examination revealed a coarse and masked face, hypokinesia and rigidity of limbs, mild dystonia of right foot and severe postural instability. Her brain MRI showed mild iron deposition in pallidum and significant in substantia-nigra with a hallo-sign on the T1-weighted sequence. Patient-2 was a 39-year-old woman who manifested mental problems and mild right hemiparesis since early childhood, progressive slowing of movements starting 6-7 years before, gait freezing, difficulty in arising from chair and depression. Neurologic examination revealed masked face, slow saccades, hypokinesia and rigidity of extremities, shuffling gait and decreased arm swings. Her brain MRI was similar to patient-1 except for normal globus pallidus and moderate frontotemporal atrophy. Results of the genetic analysis showed two different de novo heterozygous variations (a splice site and an insertion) in *WDR45* in both patients. It seems these variations affect interactions of the encoded proteins with autophagy proteins. Further analysis is needed to confirm the pathogenic effect of these variations.

Biography

Afagh Alavi has completed her MSc, and PhD studies at University of Tehran. She is an assistant professor in the University of Social Welfare and Rehabilitation Sciences currently. She has published more than 17 papers in reputed journals.

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