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Genetic Characterisation of Pantothenate Kinase Associated Neurodegeneration (PKAN) in a Consanguineous Family from Jammu and Kashmir, India

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ABSTRACT Pantothenate kinase-associated neurodegeneration (PKAN), a rare neurological disorder occurs by variation(s) in the PANK2 (Pantothenate kinase 2) gene and is linked to iron accumulation in the basal ganglia. The researchers have carried out targeted gene sequencing of all exons of *PANK2* in a patient with suspected phenotype of PKAN. A missense variant in exon 6 of PANK2 gene (NM_153638.3:c.1583C>T,NP_705902.2:p.Thr528Met) has been identified in the patient. Further, sequencing of the exon in extended consanguineous family showed autosomal recessive mode of inheritance in the family. It is emphasised that inclusion of molecular diagnostics in clinical evaluation procedures of potential genetic or uncharacterised abnormalities is critical especially if the family has known history of high consanguinity. It is anticipated to provide effectively, such families with access to a variety of genetic counselling programmes, thus reducing illness burden in the affected family.