

## **ACANTHOCYTOSIS, RETINITIS PIGMENTOSA AND PALLIDAL DEGENERATION: A CASE REPORT WITHOUT LIPOPROTEIN ABNORMALITIES**

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**Introduction:** HARP syndrome is a rare disorder characterized by hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa and pallidal degeneration . Patients without lipoprotein abnormalities have also been reported. Mutation in the gene coding pantothenate kinase 2 (PANK2) described in Hallervorden-Spatz Syndrome is also reported in HARP Syndrome.

**Case Report:** A 13-year-old boy whose development was normal until age 7 years, admitted for evaluation of difficulty in walking and swallowing. Language skills and visual acuity were also deteriorated. Physical examination revealed sialorrhea, oromandibular and upper and lower extremity dystonia and dysarthria. Ophtalmological examination showed retinitis pigmentosa. Blood smear showed %9 acanthocytes. Brain MRI showed hypodensity in globus pallidus (tiger's eye) . Lipoprotein electrophoresis was normal. Analysis of the PANK2 gene showed the aminoacid residue valine at position 450 was substituted for leucine( Val450Leu) in exon 7.

**Discussion:** Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa and pallidal degeneration (HARP) syndrome shares not only clinical features but also PANK2 mutations with Hallervorden-Spatz syndrome. There are also rare reported cases without lipoprotein abnormalities. It is worth discussing whether patients without lipid abnormalities are variants of Hallervorden-Spatz Syndrome.