PIGMENTATION

# JAPANESE PATIENT WITH HERMANSKY-PUDLAK SYNDROME TYPE 9

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Background: Hermansky-Pudlak syndrome (HPS; OMIM 203300) is a rare autosomal recessive disorder characterized by oculocutaneous albinism (OCA), a bleeding tendency due to absent platelet granules and the accumulation of ceroid lipofuscin in lysosomes. HPS is genetically heterogeneous, and the severity of skin, hair and iris hypopigmentation and visual impairment is variable. To date, ten causative genes for HPS have been identified in humans. All those genes, except AP3B1 (HPS2) and AP3D1 (HPS10), encode components of biogenesis of lysosome-related organelles complexes (BLOCs), which are divided into three forms: BLOC-1 (HPS7/DTNBP1, HPS8/BLOC1S3 and HPS9/BLOC1S6), BLOC-2 (HPS3, HPS5 and HPS6) and BLOC-3 (HPS1 and HPS4).

Observation: We identified a patient with HPS9, using whole-exome sequencing (WES), followed by analysis of hair samples from the patient detailing their morphological and chemical characteristics. The patient was a 52 year-old female who was homozygous for a novel mutation (c.285_286dupTC, p.H96LfsX22) in BLOC1S6. Although she had no episode of recurrent infection, a blood examination revealed leucopenia and mild thrombocytopenia. She developed schizophrenia when she was in her late forties. So far, she has not developed interstitial pneumonia or granulomatous colitis. According to the electron microscopy result, the HPS patient had fewer, smaller and more immature melanosomes than the healthy control. The chemical analysis also supported the clinical manifestation. Reduced total melanin content and increased level of pheomelanin, which is strongly associated with melanoma carcinogenesis, were noted.

Key message: This is the first report showing the melanosome and melanin conditions affected by the disruption of BLOC-1 in hair samples from the patient with HPS9.