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Title: Hermansky-Pudlak syndrome type 4 with interstitial pneumonia

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Body: Hermansky–Pudlak syndrome (HPS) is an autosomal recessive disorder characterized by oculocutaneous albinism, bleeding tendency, and lysosomal accumulation of ceroid-like material, with occasional development of interstitial pneumonia (IP). Nine genetically distinct subtypes of HPS are known in humans; IP develops primarily in types 1 and 4. Most reported cases of HPS with IP are type 1, and there are no published reports of type 4 in Japanese individuals. A 58-year-old man with congenital oculocutaneous albinism and progressive dyspnea for 1 month was admitted to our hospital. We administered high-dose corticosteroids on the basis of a diagnosis of acute exacerbation of interstitial pneumonia. Respiratory symptoms and the findings of high-resolution computed tomography (CT) showed improvement. He was diagnosed with HPS type 4 with interstitial pneumonia on the basis of gene analysis. He has been receiving pirfenidone for 1 year and his condition is stable. This is the first report on the use of pirfenidone for HPS with IP caused by a novel mutation in the HPS4 gene. We conclude that HPS should be suspected in patients with albinism and interstitial pneumonia. High-dose corticosteroid treatment may be useful in cases of acute exacerbation of interstitial pneumonia due to HPS-4, and pirfenidone may be useful and well tolerated in patients with HPS-4.