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# Case report from a resource-limited setting: Hermansky–Pudlak Syndrome in an 8-year-old Sudanese girl

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Hermansky-Pudlak Syndrome (HPS) is a rare autosomal recessive disorder with multisystem involvement, caused by mutations that lead to defective lysosomal function. The patient was an8-year-old Sudanese girl with a history of easy bruising and bleeding from minor injuries. On examination, she had various degrees of hypopigmentation with poor visual acuity and conjugated horizontal nystagmus. Based on these findings, the patient was tentatively diagnosed as having HPS although genetic confirmation was not conducted due to ongoing armed conflict in Sudan. When a patient exhibits bleeding diathesis, oculocutaneous albinism, and visual problems, HPS should be considered in the differential diagnosis.

**Keywords:** armed conflict, bleeding diathesis, hypopigmentation, Hermansky-Pudlak syndrome

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Hermansky-Pudlak Syndrome (HPS) is a rare autosomal recessive disorder with an estimated global prevalence between one in 500 000 and one in 1 000 000.[1] Two Czechoslovakian doctors who recorded patients with oculocutaneous albinism and related bleeding disorders were the first to report HPS in 1959.[2] HPS is characterised by varying degrees of oculocutaneous albinism, nystagmus, bleeding diathesis, and other phenotypes, including pulmonary fibrosis and colitis.[1-3] Lysosome-related organelle dysfunction is the cause of many systemic symptoms, including dense body and melanosome deficiency.[4] The considerable clinical variability and molecular heterogeneity among HPS subtypes can complicate accurate diagnosis and the selection of appropriate management strategies.[4,5] The diagnosis must be supported by evidence of a lack of platelet storage capacity and ocular albinism.[5] Genetic profiling is essential to identify the type of HPS and, thus, determining the prognosis.[3] In patients with HPS, albinism with variable degrees of hypopigmentation can be detected early after birth, but other symptoms take time to manifest; for instance, excessive bleeding or bruising are typically not identified until circumcision or the onset of walking.[6]

We report a case of HPS in an 8-year-old Sudanese girl, although no genetic confirmation was conducted owing to the ongoing armed conflict in Sudan.

The patient's parents provided written informed consent for publication of this case report and accompanying images.

## Case report

An 8-year-old Sudanese girl with a history of easy bruising and bleeding from minor injuries presented to our paediatrics department. She has four healthy siblings and no noteworthy family history. She is the second child of consanguineous, healthy parents with normal skin pigmentation and dark hair, consistent with the general Sudanese phenotype. Upon examination, her weight and height were normal (26 kg and 130 cm, respectively), and she was wearing glasses. Her skin appeared pale, her hair was sparse and yellowish-to-silver in colour, her irises were pale, and bruising was noted on her lower lip. (Fig. 1). An ophthalmic examination revealed poor visual acuity, conjugate horizontal nystagmus and bilateral hypopigmentation. Her haematological profile (Table 1) showed a prolonged bleeding time. Liver and renal parameters fell within normal limits. The HIV test results from the virology panel were negative. The patient's history was not suggestive of respiratory symptoms or gastrointestinal complaints such as chronic diarrhoea or abdominal pain.



*Fig. 1. Fair white skin, yellowish-to-silver sparse hair and bruising in the lower lip.*

**Table 1. Haematological profile**

| Laboratory parameter | Results | References | Units |
|---|---|---|---|
| TWBCs | 10 | 4.5 - 13.5 | $\times 10^9$/L |
| Neutrophils absolute | 1.8 | 1.5 - 8.5 | $\times 10^9$/L |
| Lymphocytes absolute | 5 | 2 - 8 | $\times 10^9$/L |
| RBCs | 4.2 | 4.1 - 5.3 | $\times 10^{12}$/L |
| HB | 13.1 | 11 - 13.5 | g/dL |
| PLT | 278 | 150 - 450 | $\times 10^9$/L |
| INR | 0.9 | <1.2 | |
| PT | 15.8 | 11 - 13.5 | Sec |
| APTT | 30.8 | 25 - 35 | Sec |

TWBC = total white blood cell count; RBC = red blood cell count; HB = haemoglobin; PLT = platelet; INR = international normalised ratio; APTT = activated partial thromboplastin clotting time.

Based on the findings of oculocutaneous albinism, bleeding diathesis, and conjugate horizontal nystagmus, the patient was diagnosed with HPS.

## Discussion

The presence of consanguinity in this case supports the autosomal recessive inheritance pattern of the disorder, although the absence of similarly affected siblings may reflect genetic variability or carrier status among family members.

There are currently 11 distinct subtypes of HPS, and the syndrome is associated with at least 11 human causative genes.[7] These genes encode the subunits of four necessary multi-subunit protein complexes: adaptor protein-3, the Biogenesis of Lysosome-related Organelles Complex [BLOC]-1, BLOC-2, and BLOC-3.[1] Lysosome-related organelles (LROs) are a diverse class of subcellular carriers that are responsible for transporting different materials throughout cells.[7] Two lysosome-related organelles (LROs) are primarily affected in HPS: melanosomes, leading to oculocutaneous albinism, and platelet dense granules, leading to impaired platelet function and bleeding tendency.[8] Patients with HPS may have white or olive skin and white to brown hair that darkens with age.[9] A study by Avila *et al.*[10] examined high-resolution computed tomography scans of 67 patients with HPS and described the appearance and defined patterns of radiological disease progression. Early signs of the disease included septal thickening, mild reticulations and ground-glass opacities. Peribronchovascular thickening, bronchiectasis, subpleural cysts and moderate-to-severe reticulation were observed in patients with more advanced disease. Although pulmonary fibrosis and colitis are recognised manifestations of HPS, their absence in this patient may be attributed to age-dependent disease expression.

Pulmonary fibrosis is a well-recognised complication of certain subtypes of HPS, particularly HPS-1 and HPS-4, and represents a major cause of morbidity and mortality. Although our patient did not demonstrate respiratory symptoms at presentation, pulmonary involvement remains clinically relevant owing to its potential for late onset.[10] Therefore, careful longitudinal follow-up is recommended to monitor for early signs of interstitial lung disease.[11]

Genetic confirmation was not performed because of significant disruption of the healthcare infrastructure during the ongoing armed conflict in Sudan, resulting in limited access to advanced diagnostic facilities, including molecular testing. Based on World Health Organization reports, the armed conflict in Sudan destroyed 70 – 80% of health facilities and affected millions of people during one of the worst humanitarian catastrophes in recent history.[12]

## Conclusion

When patients present with bleeding diathesis, oculocutaneous albinism and visual impairment, HPS should be considered in the differential diagnosis. Healthcare personnel should be aware of this association and monitor affected patients accordingly.

**Data availability statement.** All relevant data in the article are available from the corresponding author on request.

**Declaration.** None.

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**Author contributions.** THA contributed to the conceptualisation, clinical investigation, data collection, literature review, and drafting of the manuscript. OS contributed to supervision, interpretation of clinical findings, critical revision of the manuscript, and final approval of the submitted version. Both authors approved the final manuscript and agreed to be accountable for all aspects of the work.

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**Conflicts of interest.** None.

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