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(CASE STUDY)

A rare case of Hermansky-Pudlak Syndrome with bruise and mellitus diabetes

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Abstract

Albinism is a hereditary disorder that causes decreased pigmentation (coloring) in the body. As a consequence, albinism individuals are mostly fair-skinned with light hair known as oculocutaneous albinism. Skin, hair and color of the eye may vary, however, as some people with albinism may have dark brown hair and green or hazel / blue eyes. In both cases, poor vision and varying degrees of nystagmus (uncontrolled side-to-side eye movements) are observed. Everyone with HPS has albinism but not all people with albinism have HPS. We recorded a case with multiple comorbidities of a 37-year-old female patient with Hermansky-Pudlak syndrome (HPS) here

Keywords: Oculocutaneous albinism; Melanocytes; Torsional nystagmus; Pigmentation

1. Introduction

Oculocutaneous albinism is associated with Hermansky-Pudlak syndrome (HPS). Oculocutaneous albinism is a heterogeneous autosomal recessive condition usually characterized by congenital reduction or absence of melanin pigment, which is commonly observed in HPS. OCA includes reduced pupil, hair and skin pigment. Type 1, Type 2 and Type 3 are various forms of Oculocutaneous Albinism. Type 1 features include white hair, very pale skin and light irises. The skin is creamy white in appearance in Type 2 patients and hair can be light yellow, blonde or light brown. Type 2 usually is less serious than Type 1. Type 3 is often referred to as rufous oculocutaneous albinism that typically affects people with dark skin, and is often associated with milder vision problems than others1. It has reddish-brown skin, ginger or red hair and has hazel or brown irises. Types 1 and 2 of oculocutaneous albinism are the most common forms among the three types. Legal blindness [2], nystagmus, strabismus, transillumination of the iris, foveal hypoplasia, and albinotic retinal mid-periphery are common in HPS patients. HPS is a rare disease, doctors are frequently misdiagnosed and unfamiliar with the symptoms. Platelets are special cell parts which circulate in the bloodstream and help to clot the blood. HPS patients have high platelet levels, but they're not made properly and don't function well, because the blood doesn't coagulate properly [3]. As such, people with HPS can bruise easily, or have frequent or serious bleeds in their nose. Bleeding problems are generally mild but, in some situations, they can be very serious, particularly in circumstances such as surgery. Many women may require special medical attention during their menstrual cycle or during childbirth because they may lose too much blood.

Tiny clinical pilot study conducted at the National Eye Institute (NEI) indicates the drug Nitisinone [4] increases the development of melanin in some people with type 1B (OCA-1B) oculocutaneous albinism [5].

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2. Case Description

A 37-year-old female OCA patient was rushed to secondary care hospital with fever symptoms, cough, reduced intake of food and shortness of breath. She is a confirmed type 2 case of Mellitus Diabetes. She was found to have an unusually fair complexion, hypopigmented body, light yellowish hair and torsional nystagmus upon physical inspection. She reported heavy bleeding during menstruation and easy bruising upon medical reconciliation. The patient has been diagnosed with Hermansky-Pudlak syndrome, Oculocutaneous albinism and Mellitus type 2 diabetes.

The patient was focused about consciousness. Her BP was measured as 100/70mmHg. The pulse rate was 96 beats / min, 22 breaths / minute, 78 per cent CVS S1S2 + and SpO2. The patient was found to have an rise in WBC (12.3 x 103/mm3) and a decrease in the number of platelets (94x103/mm3), even her polymorphs were 72% and lymphocytes 22%. Electrocardiogram showed reversal of T wave and Sinus tachycardia. IgM Dengue has been done to test Dengue but it has been found negative.

She was given 2L / min nasal oxygen, ringer lactate intravenous fluids, and 1 pint @100ml / hour, C in normal saline. 20 mg omeprazole, C. 100 mg doxycycline, T. 500 mg metformin, T. aspirin 150mg, T. clopidogrel, T. atorvastatin 10 mg, Inj. paracetamol was administered 500 mg.

She was observed on the second day to have Sinus Tachycardia with inversion of T wave. 2L / min nasal oxygen, T. losartan 50 mg, T. furosemide 40 mg, T. cetirizine 10 mg was administered along with the medication given the previous day, and IV fluids were also stopped. Considering the risk benefit ratio, the dosage of Aspirin was reduced to 75 mg.

For Day three T. salbutamol 4mg, T. amlodipine 2.5 mg was administered along with other drugs and T. Losartan was stopped.

On Day four, Inj. dopamine, Inj. 20 mg furosemide STAT IV, Inj. Hydrocortisone 100 mg administered intraveneously STAT and Budecort nebulizer. The prognosis of the patient was poor, and referred for further treatment and differential diagnoses to the multispecialty hospital. Consent was obtained from patient for publishing her data, but she denied for her picture. Since of COVID-19 we did not follow up the case.

3. Discussion

Hermansky-Pudlak syndrome (HPS) is a heterogeneous group of autosomal recessive disorder with tyrosinase-positive oculocutaneous albinism (Ty-pos OCA), bleeding rates, and systemic complications associated with lysosomal dysfunctions. Photophobia is common in patients with HPS, because eyes do not have a pigment to protect against sunlight. Most people with HPS have albinism so getting HPS for all people with albinism isn't necessary. At age 30 and 40 HPS patients may experience breathing difficulties as in this case, as lung scarring worsens as their age rises. HPS patients may feel gradually tired or short of breath, and may need supplemental oxygen. HPS is a rare condition, and the causes are not known to doctors. In HPS the levels of pigmentation to non-pigmentation levels vary. The patient's condition must be clearly conveyed to the patient and caregiver, as special medical treatment is required. Many patients with HPS can lead a relatively normal life but many patients with HPS have poor vision, light skin, and face social stigma that can be due to albinism. Bleeding which is a complication of aspirin and ibuprofen in patients with HPS is contraindicated. In our case, patient was treated with supportive treatment, intravenous fluids and necessary medications.

4. Conclusion

Patients with these conditions are at increased risk of skin damage and skin cancers, hence they have to take measures to improve vision and to avoid too much sun exposure. Albinism is an autosomal recessive genetic condition causing dysfunction in one or more enzyme involved in the multi-step production of melanin inside melanocytes. A decrease or absence of melanin can reduce or obliterate pigmentation of skin, hair and eyes causing them to appear lighter in color or completely white. People with albinism seems to have problems with the development and function of the eyes, since melanin is involved in the development of optic nerves. The patient or a caregiver may be offered patient counseling. Management will concentrate exclusively on HPS. Among patients and healthcare professionals more understanding should be built so the patient does not feel one of very few others.

Compliance with ethical standards

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Disclosure of conflict of interest

The authors declare none.

Statement of informed consent

"Informed consent was obtained from all individual participants included in the study."

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