

Unraveling the Diagnostic Challenges and Complexities of Familial Gigantic Melanocytosis: A Rare Case

Ghosh Debaleena¹, Chakraborty Disha¹, Pal Shrayan¹, De Abhishek¹

¹ Department of Dermatology, Calcutta National Medical College and Hospital

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Corresponding Author: Disha Chakraborty, Senior resident, Calcutta National Medical College and Hospital, D 629 Lake gardens Kolkata 700045 West Bengal, Tel: 91-9674074135. E-mail: docdishachaks@gmail.com

Case Presentation

A boy of 11 years, born of a consanguineous marriage, presented with silvery gray hair, which he has had since birth, along with insidious onset hyperpigmentation all over the body for the past 10 years. Similar changes were found in his maternal uncle. On cutaneous examination, diffuse hyperpigmentation was seen on the face, trunk, and lower limbs along with interspersed, raindrop-like hypopigmented macules (Figure 1A). Skin biopsy showed a heavily pigmented basal layer with giant melanocytes. Fontana Masson staining showed an increased number of large melanocytes along the basal layer, with slight melanin incontinence (Figure 1, B and C). Trichogram revealed marked pigmentary dilution even in the normally pigmented hair of the patient when compared to hair from a normal individual along with irregular distribution of melanin along the hair shaft

(Figure 1D). A diagnosis of familial gigantic melanocytosis (FGM) was made.

Teaching Point

Raising awareness about this seemingly benign disorder is crucial, as it can be easily diagnosed with a simple histopathological examination. This can help prevent unnecessary investigations to differentiate it from similar-looking conditions such as Griscelli syndrome, Elejalde syndrome, and Chediak-Higashi syndrome, which are often linked to more serious neurological and hematological disorders [1,2]. In our case, although hair changes were present right at birth, the skin changes started appearing from four years of age. We also did a microscopic examination of both the normally pigmented hair and hypopigmented hair of the patient. Such unique features and findings in our case report might help to better understand the disease.

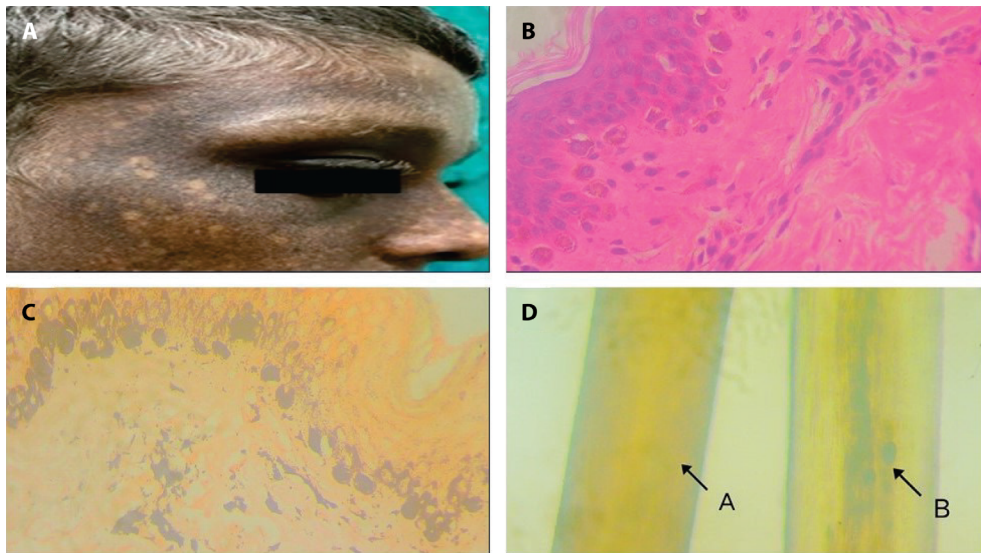


Figure 1. (A) Diffuse hyperpigmentation with hypopigmented macules all over the body. (B) Giant melanocytes in the basal layer of epidermis (H&E, $\times 40$). (C) Larger, longer, heavily pigmented melanocytes (Fontana Masson, $\times 40$). (D) A-Hair from a normal individual, B-Normally pigmented hair from a patient (unstained, $\times 40$).

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