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RESEARCH ARTICLE

TRICHOSCOPIC CLUES TO GRISCELLI SYNDROME

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Abstract

Griscelli syndrome is a rare autosomal recessive disorder characterized by pigmentary dilution of the skin and hair, associated with various systemic manifestations resulting from underlying genetic mutations. We report the case of a 5-year-old girl with no significant medical history who was admitted to the pediatric department for evaluation of hemophagocytic lymphohistiocytosis. She was noted to have gray hair, eyelashes,and eyebrows since birth. Trichoscopic examination revealed hypopigmented hair shafts lacking pigment clumps but displaying regul arly spaced pigment clusters arranged in a discontinuous linear pattern, findings consistent with Griscelli syndrome.

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Introduction:-

A 5-year-old girl, with second-degree consanguinity and no significant past medical history, was admitted to the pediatric department for an acute severe multiorgan disease with fever, hepatosplenomegaly, pancytopenia, and lymphadenopathy suggestive of hemophagocytic lymphohistiocytosis. A dermatological consultation was requested following the observation of gray hair in the child. Since birth, she had silvery-gray hair over her scalp, eyebrows, and eyelashes. There was no history of silvery-gray hair in any of the family members. Clinical examination revealed dense gray scalp hair, along with gray eyebrows and eyelashes associated with a well-defined hypopigmented macule with irregular borders on the back (Figures 1A-B). Nails, teeth, and mucosae were normal. Trichoscopy showed hypopigmented hair shafts lacking pigment aggregates, with regularly spaced pigment clusters arranged in a discontinuous linear patternandleukotrichia (Figures 2,3). In view of the suspicion of hemophagocytic lymphohistiocytosis, the congenital hypopigmentation of the hair and skin, and the presence of parental consanguinity, Griscelli syndrome type 2 was strongly suspected. Unfortunately, genetic testing to investigate a mutation in the RAB27A gene could not be performed due to financial constraints.

Griscelli syndrome is a rare autosomal recessive disorder characterized by pigmentary dilution of the skin and hair, associated with various systemic manifestations resulting from underlying genetic mutations [1]. It is classified into three types based on the specific gene involved and the corresponding clinical presentation. Griscelli syndrome type 1 is found in patients with silvery-gray hair, fair skin, severe early psychomotor retardation, and normal immune status. It is secondary to a mutation in the myosin Va (MYO5A) gene located on chromosome 15q21, which plays a role in regulating organelle transport in melanocytes and neuronal cells [2]. Griscelli syndrome type 2, initially described as partial albinism with immunodeficiency, is characterized by silvery-gray hair, recurrent severe infections, and a high risk of hemophagocytic lymphohistiocytosis. This subtype is caused by mutations in the

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RAB27A gene on chromosome 15q21, which encodes a GTPase involved in melanosome transport and cytotoxic granule exocytosis [1,3]. Griscelli syndrome type 3 is limited to cutaneous and hair abnormalities without systemic involvement.

Dermoscopy is a rapid, painless, and non-invasive tool, making it especially suitable for use in children. In fact, trichoscopy typically reveals hypopigmented hair shafts with small, regularly distributed pigment clusters arranged in a linear or discontinuous pattern along the shaft, corresponding to uneven melanosome aggregation [4]. These findings are highly suggestive and differ markedly from those seen in other causes of silvery-gray hair, such as Chediak–Higashi syndrome, which represents a major differential diagnosis. While GS is characterized by fine, regularly spaced pigment granules, Chediak–Higashi syndrome shows large, coarse, irregular melanin clumps within the hair shaft, reflecting a different defect in lysosomal trafficking [5].Trichoscopy is a valuable diagnostic tool in Griscelli syndrome, revealing characteristic hair shaft pigmentary abnormalities that allow early diagnosis, differentiation from other silvery hair disorders, and prompt multidisciplinary management, potentially improving clinical outcomes.

Figures and legends:

Figure 1: (A) Silvery-gray hair, eyelashes, and eyebrows. (B) Hypopigmented macule on the back (blue arrow).

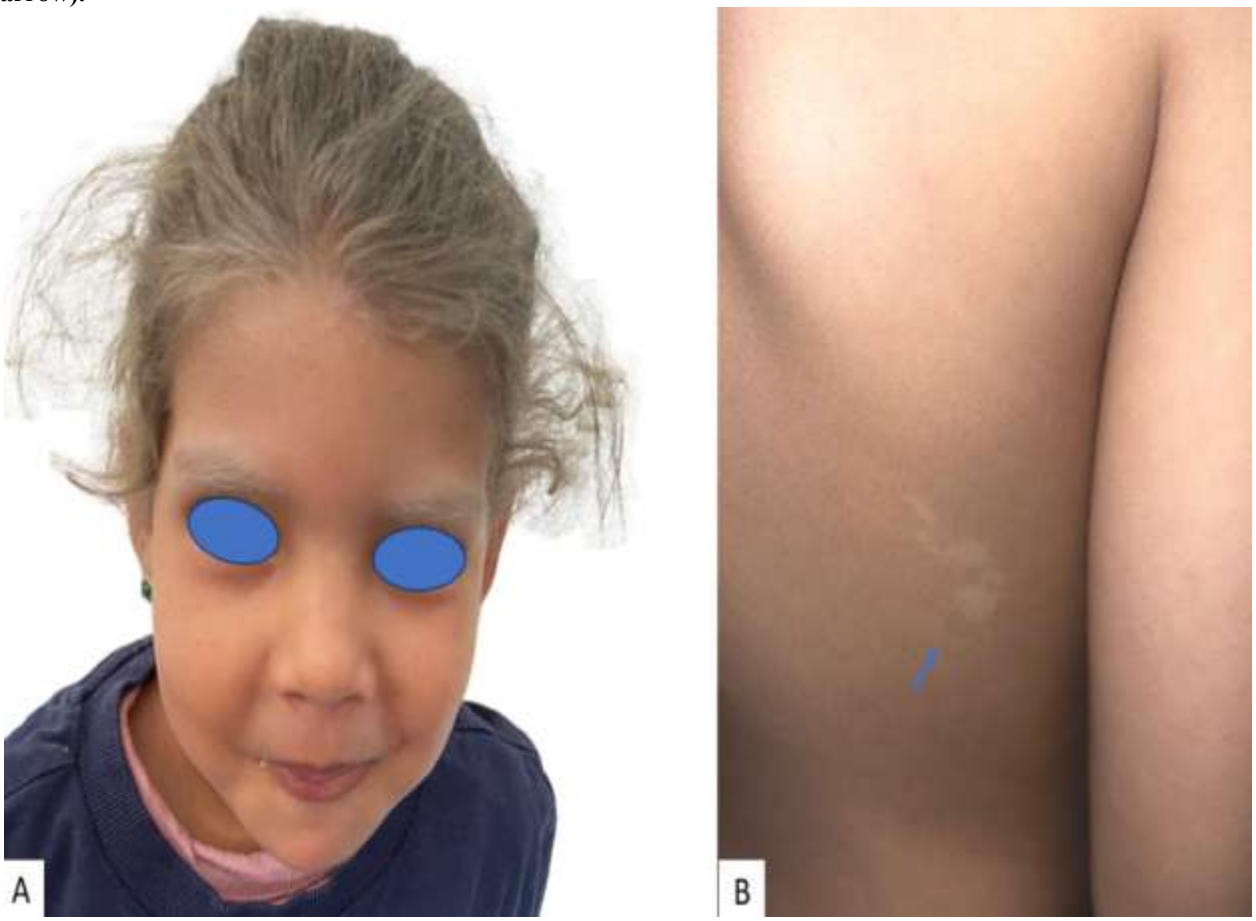

Figure 2: Dermoscopy of hypopigmented hair shaft (blue arrow) with regularly spaced pigment clusters arranged in a discontinuous linear pattern (orange arrow).

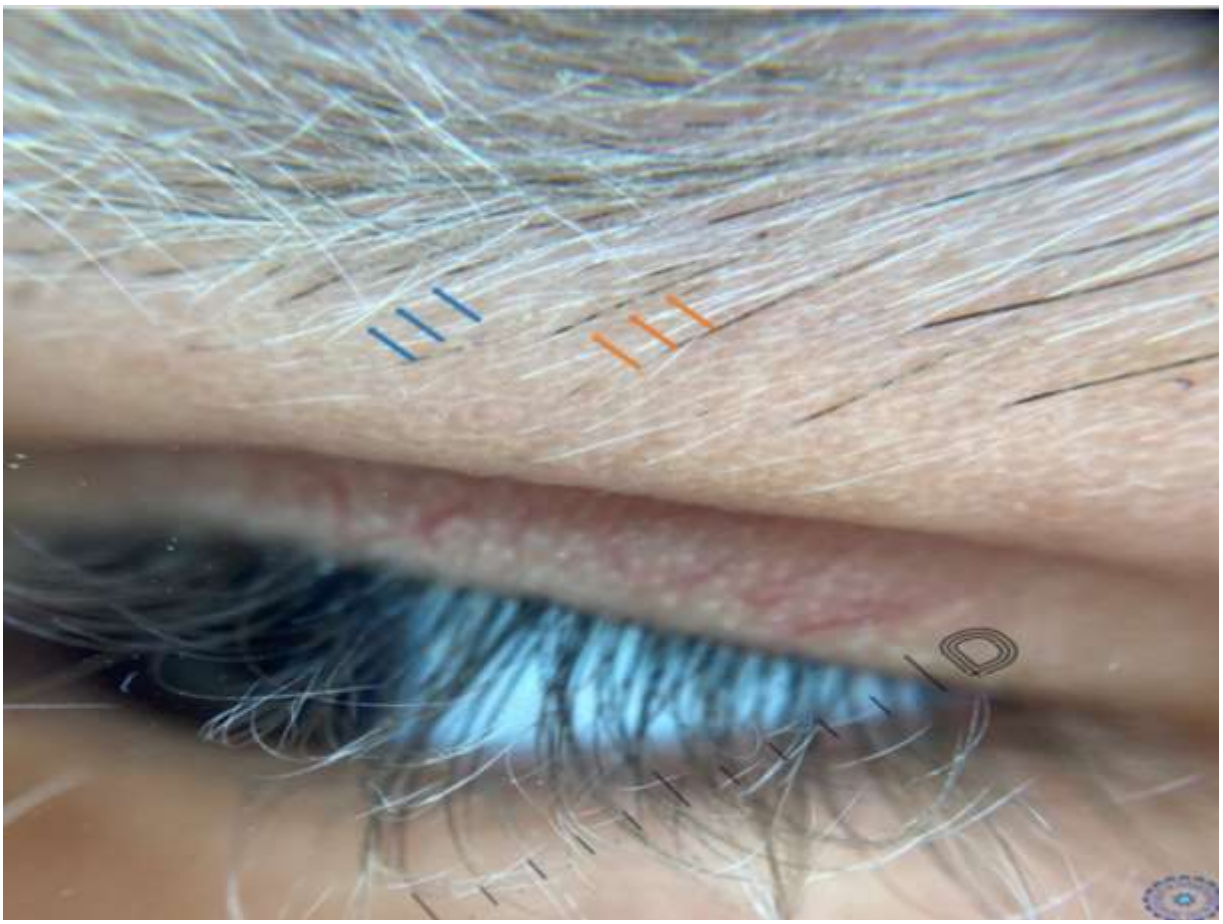

Figure 3: Dermoscopy of the hair shafts reveals short longitudinal pigmented lines arranged in a linear distribution (orange arrow), leukotrichia (blue arrow).

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