

## Silver Strands and Skin Spots: Unveiling Griscelli Syndrome Type 3 in a 7-year-old Girl

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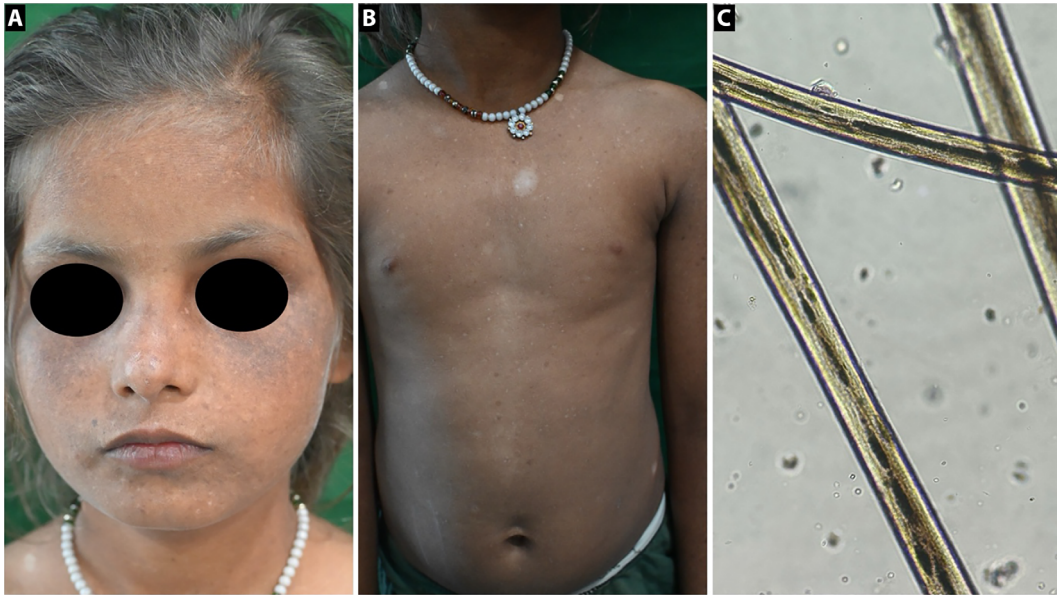
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### Case Presentation

A 7-year-old female, born to non-consanguineous parents, presented with primary complaints of light-colored scalp and body hair and white macules on her skin. Clinical examination revealed silvery gray hair predominantly localized on the frontal and temporal scalp regions as well as the eyebrows and eyelashes (Figure 1A). Numerous discrete hypopigmented and hyperpigmented macules were present in a generalized manner, with fine gray hair covering these areas (Figure 1B). Comprehensive physical and systemic examinations were unremarkable, with no similar finding in any other family member. There was no history of recurrent infections or growth failure. Hair microscopy showed coarse, unevenly distributed clumps of melanin pigment within the medulla of the hair shaft (Figure 1C). Routine hematological and neurological assessments were within normal limits without any developmental or growth delays. Genetic testing identified a pathogenic homozygous mutation in the *MLPH* gene, c.104G>T (p.Arg35Trp), confirming the diagnosis of Griscelli syndrome type 3.

### Teaching Points

Griscelli syndrome type 3 is the rarest subtype of GS, with only a few cases reported in the literature. The hallmark features include silvery gray hair and pigmentary anomalies, which are confined to the skin and hair. The *MLPH* gene mutation impairs the proper distribution of melanin in melanocytes, leading to the characteristic pigmentary dilution seen in GS3. Unlike GS1 and GS2, patients with GS3 do not exhibit neurological or immunological deficits, making it a less severe form of the disorder. The differential diagnosis for GS3 includes other syndromes with pigmentary dilution, such as Chediak-Higashi syndrome and Elejalde syndrome; however, the absence of systemic involvement helps to distinguish GS3 [1]. This case underscores the need for early diagnosis of Griscelli syndrome type 3 to distinguish it from the more severe types and prevent unnecessary interventions.



**Figure 1.** (A) Silvery gray hair predominantly localized on the frontal and temporal scalp regions, eyebrows, and eyelashes and numerous hypopigmented and hyperpigmented macules interspersed with normal skin on the face; (B) Numerous hypopigmented and hyperpigmented macules interspersed with normal skin on the chest and abdomen; (C) Coarse, unevenly distributed clumps of melanin pigment within the medulla of the hair shaft (100x).

**Ethics Committee:** The study was conducted ethically in accordance with the World Medical Association Declaration of Helsinki. The patient's father gave his written informed consent to publish the case (including the publication of images).

## Reference

1. Castaño-Jaramillo LM, Lugo-Reyes SO, Cruz Muñoz ME, et al. Diagnostic and therapeutic caveats in Griscelli syndrome. *Scand J Immunol.* 2021;93(6):e13034. DOI:10.1111/sji.13034. PMID: 33660295.