“Road-Dividing Line”-Like Pigmentation of Hair as a Diagnostic Clue for Griscelli Syndrome

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Established Facts

- Griscelli syndrome is a rare autosomal recessive disorder, characterized by unusual hypopigmentation of skin and hair.

Novel Insights

- In our case, the possible diagnosis of Griscelli syndrome has been set after the child’s microscopic hair examination, which revealed pigmentation of the hair shaft resembling a “road-dividing line”.

Keywords

Griscelli syndrome · Pigmentation · Hair shaft · Microscopic hair examination · Road-dividing line

teristic for Griscelli syndrome, and in this framework, dermatologic examination may be of high importance for the diagnosis.

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Abstract

We report a case of a 5-year-old girl with physical and psychomotor retardation, acquired microcephaly, and history of recurrent infections. Dermoscopic and microscopic hair examination revealed a “road-dividing line”-like pigmentation of hair shafts. The combination of history, clinical findings, and hair examination led to the diagnosis of Griscelli syndrome type II. The picture of “road-dividing line” on hair microscopic and dermoscopic examination is highly charac-

Introduction

Griscelli syndrome (GS) is a rare autosomal recessive disorder, characterized by unusual hypopigmentation of skin and hair. Its exact prevalence is unknown. The age at onset ranges between 4 months to 7 years, but most often the disorder starts in infancy. No sex predilection has been described [1, 2].

Fig. 1. Physical deformities of the patient's face (a) and lower extremities (b).

Fig. 2. Dermoscopy of hair shafts reveals longitudinal, short, pigmented lines in a linear distribution.

In this brief report, we present a possible case of GS with skin, ophthalmological, immunological, and neurological involvement, in which hair examination led to the diagnosis. The parents have given their consent for the presentation of this case.

Case Report

A 5-year-old girl presented to the Pediatrics Outpatient Clinic with severe physical retardation and deformities. There was a history of intrauterine fetal growth retardation. She also suffered from recurrent infections of various severities due to congenital leukopenia/neutropenia. There was no family history of similar condition. On clinical examination, physical retardation and acquired microcephaly were evident. Neurologic examination revealed severe psychomotor retardation of neurodegenerative type, as well as signs of demyelinating polyneuropathy. In addition, there was a mild, transient hepatosplenomegaly. Skin and hair examination

showed no signs of albinism, but compared to her parents, their color appeared lighter. Funduscopic examination showed findings consistent with albino fundus.

Magnetic resonance imaging showed a nonspecific leukodystrophy. Extensive metabolic workup was within normal limits, apart from an increase in mucopolysaccharides. Complementary laboratory investigations, however, ruled out the diagnosis of mucopolysaccharidosis.

The child was referred for further evaluation to the 2nd Department of Dermatology and Venereology, "Attikon" University General Hospital. On clinical examination, the scalp hair appeared thin and pale (Fig. 1). Ex vivo dermoscopic examination showed hypopigmented hair. At sites, there were longitudinal, short, pigmented lines in a linear distribution (Fig. 2). Examination of hair shafts under the light microscope showed unevenly distributed melanin, with a "road-dividing line"-like appearance (Fig. 3). Based on medical history, clinical examination, and laboratory investigations, the diagnosis of GS type II was made.

Discussion

GS is classified into 3 types. Type I is due to MYO5A gene mutations and it is manifested by hypomelanosis associated with a primary neurological deficit (typically delayed development, intellectual disability, seizures, and hypotonia). Type II presents with hypopigmentation combined with variable cellular immunodeficiency, and it is caused by mutation in the RAB27A gene. Affected individuals are prone to recurrent infections. They also develop hemophagocytic lymphohistiocytosis, in which overproduction and infiltration by activated histiocytes (T lymphocytes and macrophages) may damage various organs and tissues, occasionally with a fatal outcome. Type III is also characterized by hypomelanosis, but without neurological or immunological manifestations. This

Fig. 3. Light microscopy of hair shafts [magnification, $\times 40$ (a) and $\times 100$ (b)] shows unevenly distributed melanin, with a “road-dividing line”-like appearance (c), a very characteristic finding in GS.

type may result from a mutation in melanophilin (MLPH) or the MYO5A gene [3–5].

In our case, the diagnosis of GS type II was based on the combination of skin and hair hypopigmentation with abnormalities of the immune system. Microscopic examination of the hair shaft revealed the typical pattern of large pigment clumps, instead of small homogenous pigment granules seen in normal hair [1]. This distinctive picture that resembles a “road-dividing line” is highly characteristic for GS. In our brief report, we emphasized that this finding is also obvious on dermoscopy. In this context, dermatologic evaluation as well as dermoscopic

and microscopic hair examination can be of great importance for the diagnosis of GS.

Statement of Ethics

The parents have given their informed consent for the presentation of the case.

Disclosure Statement

The authors have no conflicts of interest to disclose.

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