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Case Report
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Oral and dental findings of Griscelli syndrome type 3

Ozlem Marti Akgun, Ceyhan Altun, Gunseli Guven Polat, Ceren Yildirim

ABSTRACT

Griscelli syndrome (GS) is a rare autosomal recessive genetic disorder characterized by variable immunodeficiency, partial albinism, abnormal accumulation of melanosomes in melanocytes, pigmentary dilution of the skin, and shiny silver-gray hair. GS has three types, with the first and second types caused by mutations in two genes being located at band 15q21: RAB27A and MYO5A. The expression of the third form of GS is restricted to the characteristic hypopigmentation of GS, and results from mutation in the gene that encodes melanophilin MLPH. It has also been shown that an identical phenotype can result from the deletion of the MYO5A F-exon. The aim of this case report is the presentation of oral and dental features and SEM images of the hair of a 12-year-old girl with GS type 3.

Key words: *Griscelli syndrome, microdontia, genetic mutation*

Introduction

Griscelli syndrome (GS) was first described by Griscelli in 1978 [1]. It is defined by the characteristic hypopigmentation, with frequent pyogenic infection, hepatosplenomegaly, neutropenia, and thrombocytopenia. Very often, there is also impaired natural killer cell activity, absent delayed-type hypersensitivity and a poor cell proliferation response to antigenic challenge. This may be caused by the loss of three different genes, each of which has different additional effects, resulting in three types of syndrome. Its inheritance is autosomal recessive and can manifest with silver-gray hair, which may be accompanied by neurological abnormalities (type 1), immunodeficiency (type 2), or no other abnormality (type 3) [2].

Pathognomonic light and electron microscopic features in skin and hair biopsies in GS differentiate it from Chediak-Higashi syndrome (CHS), which has similar clinical manifestations. Granulocyte abnormalities seen in Chediak-Higashi syndrome are usually absent in GS. The prognosis in both disease entities is grave but out of 3 types of GS, type 3 has good prognosis [3].

Since 1978, when GS was first described in two patients, more than sixty patients have been described in the literature. GS is very rare in almost all populations, although most cases have been reported from Turkish and Mediterranean populations [4-6].

In this case report, we present oral and dental features and SEM images of the hair of a 12-year-old girl with GS type 3.

Author affiliations : *Department of Pediatric Dentistry, Center of Dental Sciences, Gulhane Military Medical Academy, Ankara, Turkey*
Correspondence : *Ozlem Martı Akgun, Department of Pediatric Dentistry, Center of Dental Sciences, Gulhane Military Medical Academy, Etlik, Ankara, Turkey*
e-mail: ozlemmartiakgun@gmail.com
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Case Report

A 12-year-old girl was referred to the Department of Pediatric Dentistry at Gulhane Medical Academy complaining of dental caries. From birth she was noted as having silver-gray (leaden) hair and silver-gray eyebrows and eyelashes (Figure 1). Her parents were unrelated and she was their only child. There was no family history associated with this condition. The patient's physical development was normal. She did not have any complaint relevant to other systems. There was no hepatosplenomegaly or lymphadenopathy. A comprehensive clinical and radiographic evaluation was performed. Extraoral examination showed a concave profile with good facial symmetry. Scanning electron microscopic (SEM) examination of the hair shaft showed large, unevenly distributed melanin aggregates which were diagnostic of GS (Figure 2).

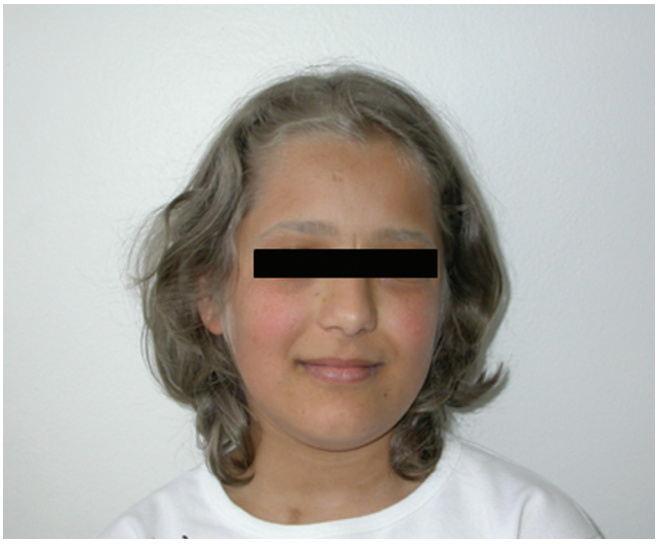

Figure 1. Extraoral appearance of the patient.

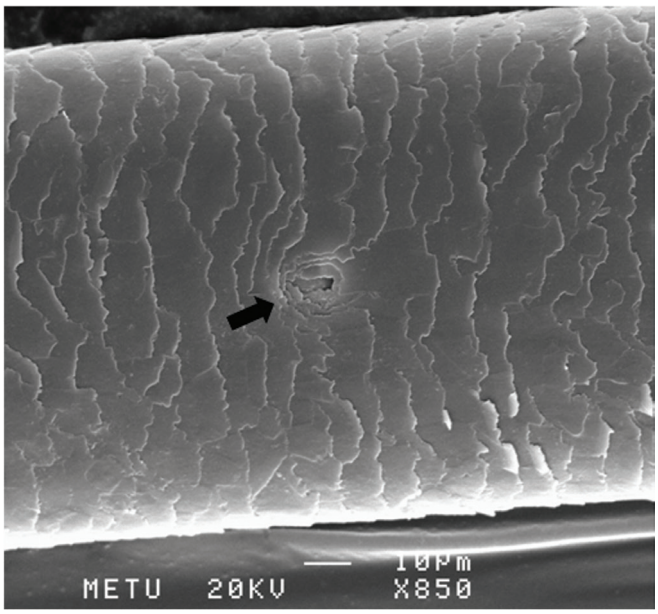

Figure 2. Scanning electron microscopic (SEM) examination of the hair of the patient.

In the oral cavity, we found normal mucosa, a deeply arched palate, deficient hygiene, and gingivitis. The panoramic radiograph revealed the absence of a tooth bud for 47, microdontia of 37 and 45, and secondary caries of 46 (Figure 3). All teeth had hypoplastic enamel. After the permission was received from the parents, the 46 was restored with amalgam. Detailed instructions on maintenance and of oral hygiene were given to the patient and her parents.

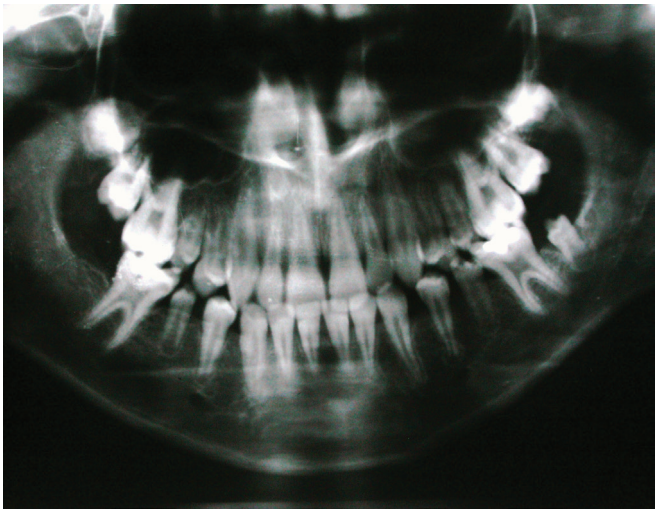

Figure 3. The panoramic radiograph revealed the absence of a tooth bud for 47, microdontia of 37 and 45, and secondary caries of 46.

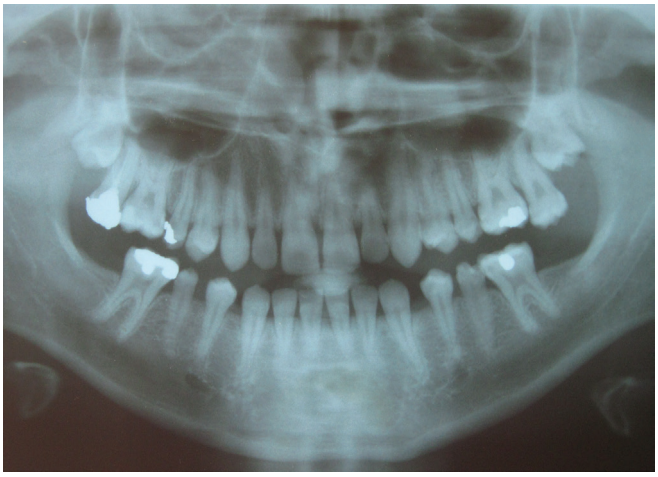

Figure 4. The panoramic radiograph of the patient was taken after 6 years.

After four years the patient referred to our clinic complaining of toothache. The periapical radiograph revealed a periodontal abscess around 37 and the tooth was extracted. The patient was followed up for 6 years for the prevention of dental caries, and was called for the fluoride application every six months (Figure 4).

Discussion

GS is characterized by the occurrence of "accelerated phases" consisting of pancytopenia, hemophagocytosis, variable cellular and humoral immunodeficiency, elevation of serum triglyceride levels, hypofibrinogenemia and hypoproteinemia, and partial albinism. In GS the genetic defects include mutations in either MYO 5A or RAB 27A, which are both located on chromosome 15q21 [7,8]. Dermatologic findings may be limited to hair, with skin and retinal pigmentation being occasionally affected. Microscopic examination of hair reveals uneven clusters of aggregated melanin pigment, accumulated mainly in the medullary area of the shaft. Neurologic involvement with encephalopathy, hypotonia, raised intra-cranial peripheral facial palsy, pressure, cerebellar signs, hemiparesis, spasticity, seizures, psychomotor retardation, and progressive neurologic deterioration is known [9]. Immunologic abnormalities include a natural killer (NK) cell function defect with absent delayed-type hypersensitivity [10]. GS type 3 represents the restricted expression of the disease characterized only by hypopigmentation in hair and skin; only two cases are reported, one of which is caused by an F-exon deletion in the MYO5a gene. Moreover, Westbroek et al. [11] diagnosed GS 3 by identifying MLPH mutations in seven affected individuals. Our case has type 3 GS, with the disease being diagnosed at the Department of Molecular Biology and Genetics. From birth she had silver-gray (leaden) hair and silver-gray eyebrows and eyelashes.

In recent literature, only two reports have focused on the issue of dental and oral findings of GS. In these reports, Kirzioglu and Altun [5] aimed to describe the orodental and physical findings of a girl with GS, atopic dermatitis (AD) and Reye's syndrome (RS), discussing the possible relationship between the three syndromes. In conclusion, they could not define any specific orodental feature of GS, but suggested that a history of RS and AD might be early warning signs of GS. In another case report, Akcakus et al. [12] reported a GS case with asymmetric crying facies (ACF) caused by congenital hypoplasia or agenesis of the depressor anguli oris muscle. In our case the patient's one tooth bud was absent, and two teeth were microdontic. These conditions may be related to genetic mutation.

In conclusion, GS is a rare condition and presents important dental findings; furthermore, several facial abnormalities may be observed. Due to the dental hypoplasia, caries susceptibility can be seen in GS patients. Patients should be given a good oral hygiene education. Also, known extra- and intraoral findings of GS may help diagnosis of the patient.

Conflict of interest statement

The authors have no conflicts of interest to declare.

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