**Immunology and Genetics Journal**

Case Report Article

Received: 15 April 2020
Accepted: 25 May 2020
Published online: 05 August 2020
Doi: 10.22034/igj.2020.235072.1041

# A Griscelli Syndrome, with Retropharyngeal Abscess, as the First Clinical Manifestation

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## Abstract

Griscelli syndrome (GS) is a rare autosomal recessive disorder, which is characterized by albinism with immunodeficiency and usually causes death in early childhood. Accordingly, this syndrome is a primary immune defects presented with a dilution of pigmentations of the skin and hair, recurrent pulmonary and skin infections, neurologic disorders, hypogammaglobulinemia, and variable cellular immunodeficiency. Moreover, in different phenotypes of the syndrome, three mutations have been mentioned. In most of them, GS leads to death in the first decade of life. Herein, we reported a one-year-old male child with an upper respiratory infection and retropharyngeal abscess as the first clinical manifestation.

**Keywords:** Grisel's Syndrome, albinism, immunodeficiency, hypogammaglobulinemia

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## Introduction

Griscelli and Siccardi described GS or partial albinism with other clinical manifestations, and mainly with immunology defects for the first time in Paris in 1978 (1). This syndrome is considered as a rare phenomenon, and up to now, lower than 100 cases have been reported worldwide. GS as a multisystem disorder has 3 subtypes (GS I, GS II, and GS III), which are based on genetic loci (melanophilin, Ras-related protein Rab-27A and Myosin VA, respectively) (2, 3). GS1 showed no involvement of the immunological system with primary impairment of the neurological system while GS2 showed an immunologically dysfunction and involvement of multisystem. Also, GS3 has the only hypomelanosis with some controversial reports regarding immunity in the latest. GS II is the most common one among three types

(4, 5). In this study, we attempted to discuss the difficulties in the diagnosis of these cases in developing countries like Iran, so we have evaluated GS cases and discussed it in the following.

## Case presentation

The patient was a one-year-old male child who was admitted to the emergency room due to drooling and dry cough. About 2 weeks ago, the patient had the symptoms of upper respiratory tract infections, and a week ago, he had the symptoms of dry cough, drooling, and loss of appetite that were added to the previous symptoms.

The patient had a hospitalization history about 5 months ago due to pneumonia. Also, his vaccination history was complete and had normal state of growth and development. The patients were 4/4 of family, and his family had no history of a medical problem. In addition, his parents had no family relationship. In general appearance, he was a boy with blue eyes and light hair (**Figure 1**). During the physical examination, in nasopharynx evaluation, the prominent bulge of the left lateral pharyngeal wall were observed with the displacement of the left tonsil and shift of the Uvula in the opposite side. However, the child had no respiratory distress. On neurologic examination, his deep tendon reflexes and cranial nerve function were normal. Also, an abdominal examination was normal, i.e. without organomegaly.

A spiral neck CT scan has been performed with contrast, and the image of a retropharyngeal abscess, measuring 33*24*12 mm in the left retropharyngeal space in the vicinity of the mandibular angle, which was extended to the hyoid bone has been observed along with the compression effect on the airway. Also, there was a smaller collection measured to be 14*14*8 mm, in the right retropharyngeal space. Therefore, due to the pressure effect on the airway, the surgery was quickly tolerated, and the patient was then intubated. The initial laboratory investigations showed that, leukocytosis is preceded with neutrophilia (white blood cells: 16100/ul; polymorphonuclear: 87%) and thrombocytosis (Platelet: 682000/ul) with an increase in acute phase reactants (erythrocyte sedimentation rate (ESR):82 mm/hr, C-reactive protein (CRP): +3). In peripheral blood smear, few giant granules have been observed in myeloid cells and giant platelets (**Figure 4**). Moreover, peripheral blood flow cytometry has normal morphologic cells. Notably, in this study, immunoglobulin levels were normal.

Microscopic evaluation of hair shaft observed an accumulation of large and irregular pigments granule, which mostly was in the medulla (**Figure 2, 3**). At the end of the genetic evaluation, the *RAB27A* gene was negative in this case. So, the patients were diagnosed as type III of GS with respect to these clinical manifestations and paraclinical evaluations.

**Figure 1.** Light hair, eyebrows, and eyelashes, and Blue eyes

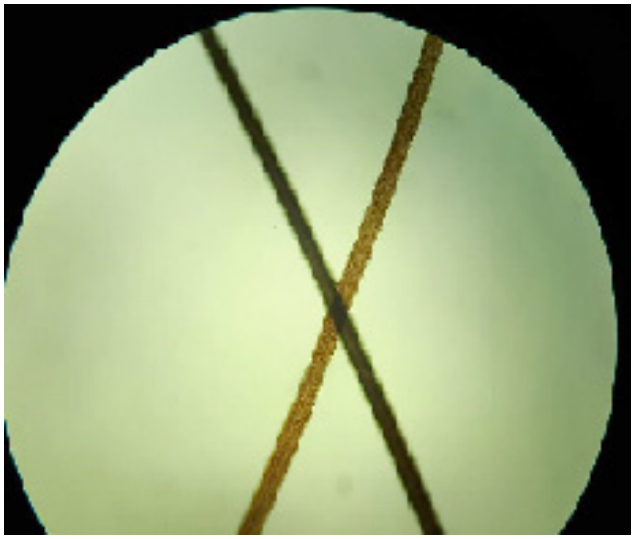

**Figure 2**. Microscopic view of normal hair shaft

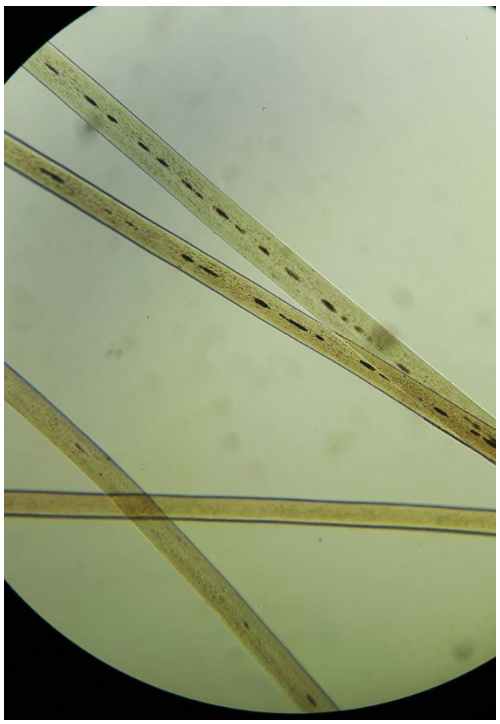

**Figure 3.** Microscopic view of the patient`s hair shaft with some irregular coarse melanin pigments

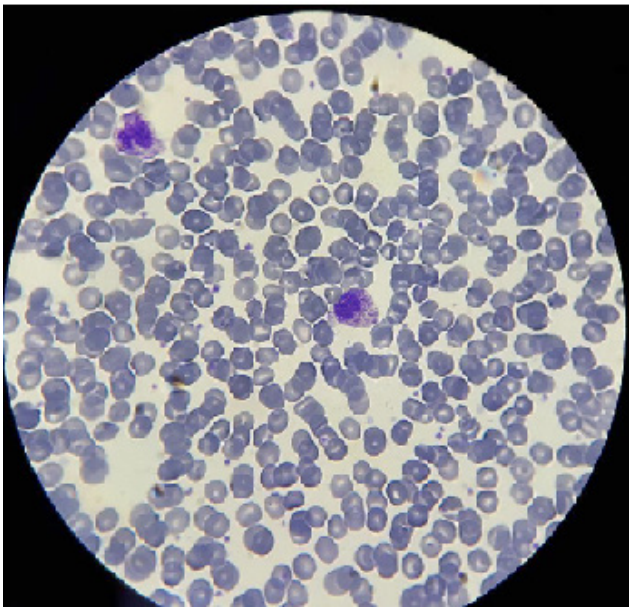

**Figure 4.** Peripheral Blood Smear of the Patient

## Discussion

In a recent study, we reported a GS case as well as his clinical manifestations; however, in following parts, another study performed on this syndrome have been discussed. GS , which is known as an autosomal recessive condition and a very rare disorder, leads to dilution of pigmentations of hair and skin with the huge clumps of pigmentations in shafts of hair resulting in silver-grey hair along with variable cellular immunodeficiency with or without respiratory infection and severe neurological defects (6).

Griscelli et al. described two patients with partial albinism in their study in 1978 (1). Moreover, studies conducted in this field have shown 3 types of GS, which are distinguished based on their patterns of symptoms, signs, and genetic causes. Also, three genes on 15q21 are responsible for GS manifestations; however, in most of the studies, it was mentioned that diagnosis has been performed based on clinical manifestations, not genetic diagnosis (7).

GS type I involves severe neurological dysfunction in brain function, as well as hair coloring and distinctive skin. The cases with this condition have typically intellectual disability, delayed development, hypotonia, and seizures. In addition, another condition called Elejalde disease has many similar signs and symptoms, and some researchers regarding this, have proposed that GS type 1 and Elejalde disease are both the same disorder (8).

Type II of GS has some abnormalities in the immunological system as well as hypopigmented hair and skin. In addition, these cases are prone to recurrent infections of the respiratory system and other organs. They also developed an immune disorder called HLH (hemophagocytic lymphohistiocytosis), in which the immune system produces too many immune cells called macrophages and T-lymphocytes (histiocytes) under this condition. Accordingly, their overactivity can damage tissues and organs throughout the body, which causes complications that are life-threatening if these conditions were untreated. Also, GS type II syndrome is related to the *RAB27A* gene(9). In most cases, colored hair and light skin are the only manifestations of type III of GS, and cases with this type of syndrome have no any other abnormality of neurological system or dysfunction of the immune system (10-12). In a case series, three adult patients with exceptional clinical presentations of haemophagocytic lymphohistiocytosis as GS were reported (13).

In the case evaluated in a recent study, we have observed blue eyes and light skin and two consolidations in the neck region, and also case genetic test as GS-2 was negative. However, the diagnosis of GS was based on clinical manifestations and microscopic diagnosis of the hair shaft, and in most cases, sever neurological defect as activated lymphohistiocytic and macrophage infiltration of the white matter of brain (14). Therefore, in our case, although we had a respiratory infection and dilution of hair pigmentation, light skin, and blue eye, the neurological defect was not corroborated. Besides, the peripheral blood smear of our patient was positive for a few giant granules in myeloid cells and giant platelets.

Long-term prognosis of these cases who are with this syndrome is poor and in the first decade of life of most cases, death happens, and survival longer than a decade are reported in few cases (10). In dangerous complications of hematological conditions, bone marrow transplantation or stem cell transplantation is recommended; however, in most studies, the success rate was poor (15).

**Conclusion:** Therefore, the case evaluated with respect to these clinical manifestations such as dilution of pigmentations of the skin and hair, recurrent pulmonary and skin infections, neurologic disorders, hypogammaglobulinemia, and variable cellular immunodeficiency was diagnosed as type III of GS.

## Conflict of Interest

The authors declare that they have no conflicts of interest.

## Acknowledgment

Written informed consents were obtained from the patients`s legal guardians for publication of this case report and its accompanying information.

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