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Case Report

A Case Report of GRISCELLI syndrome type 3

Authors:
Niharika Tentu[1], Manikanta Swamy Marisetty[2],
CMR Institute of Medical Sciences Department of Dermatology Hyderabad, GSL Medical College, Department of Pathology, Rajamahendravaram.
Corresponding Author:
Manikanta Swamy Marisetty

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ABSTRACT:
Griscelli syndrome is a rare autosomal recessive disorder that results in pigmentary dilution of the skin, the silvery-grey sheen of the hair, the presence of large clumps of pigment in hair shafts, and an accumulation of melanosomes in melanocytes. Three variants of Griscelli syndrome have been identified: Griscelli syndrome types 1-3, which can be associated with primary neurological impairment (type 1), immunologic impairment (type 2), or be isolated (type 3). Griscelli syndrome type 3 is characterized by hypomelanosis with no immunological and neurological manifestation. Prognosis is very good in type 3 Griscelli syndrome and usually requires no active intervention, as opposed to type 1 and 2 where early diagnosis and treatment play a crucial role in the patient's survival. On Cutaneous examination, the patient had silvery grey hair all over the body including, eyebrows and eyelashes. Bronze tan hyperpigmentation with intervening hypopigmentation over the whole body. Sections revealed stratified squamous keratinized epithelium with an increased number of melanocytes with aggregates of melanin pigmentation in the basal layer with adjoining poorly pigmented keratinocytes.

Keywords: GRISCELLI syndrome, hypomelanosis, type 3

INTRODUCTION:

Griscelli syndrome (GS) is a rare autosomal recessive disorder that results in pigmentary dilution of the skin, silvery-grey sheen of the hair, the presence of large clumps of pigment in hair shafts, and an accumulation of melanosomes in melanocytes. It was first reported by Griscelli et al in two unrelated patients in 1978.[1] Three variants of Griscelli syndrome have been identified: Griscelli syndrome types 1-3, which can be associated with primary neurological impairment (type 1), immunologic impairment (type 2), or isolation (type 3).[2] Griscelli syndrome type 3 is characterized by hypomelanosis with no immunological and neurological manifestation.[3] Prognosis is excellent in type 3 Griscelli syndrome and usually it requires no active intervention, as opposed to type 1 and 2 where early diagnosis and treatment play a crucial role in the patient's survival.

CASE REPORT:

A 23-year-old male patient came to the dermatology, venereology, and leprosy department with the complaint of lightening of skin and hair since infancy. There is no history of fever, jaundice, abdominal pain, convulsions, or photosensitivity. He was born out of a non-consanguineous marriage, with an uneventful antenatal or perinatal period. His physical development was normal. His sibling had no similar complaints. On cutaneous examination, the patient had silvery grey hair all over the body (Fig 1). Bronze tan hyperpigmentation with intervening hypopigmentation over the whole body (Fig 2). Teeth and nails appeared normal. Systemic examination was insignificant. The Gastrointestinal System examination revealed no hepatosplenomegaly, central nervous system, respiratory, and cardiac system examination was within normal limits. The ophthalmological examination did not reveal any abnormality. Haematological parameters were normal, and serum copper levels were normal. Light microscopic examination of hair revealed- uneven clusters of aggregated melanin pigment, accumulated mainly in the medullary area of the shaft (Fig 3). Histopathological sections from the skin biopsy revealed stratified squamous keratinized epithelium with an increased number of melanocytes with aggregates of melanin pigmentation in the basal layer with adjoining poorly pigmented keratinocytes (Fig 4).

Figure 1: On cutaneous examination, the patient had silvery grey hair all over the body

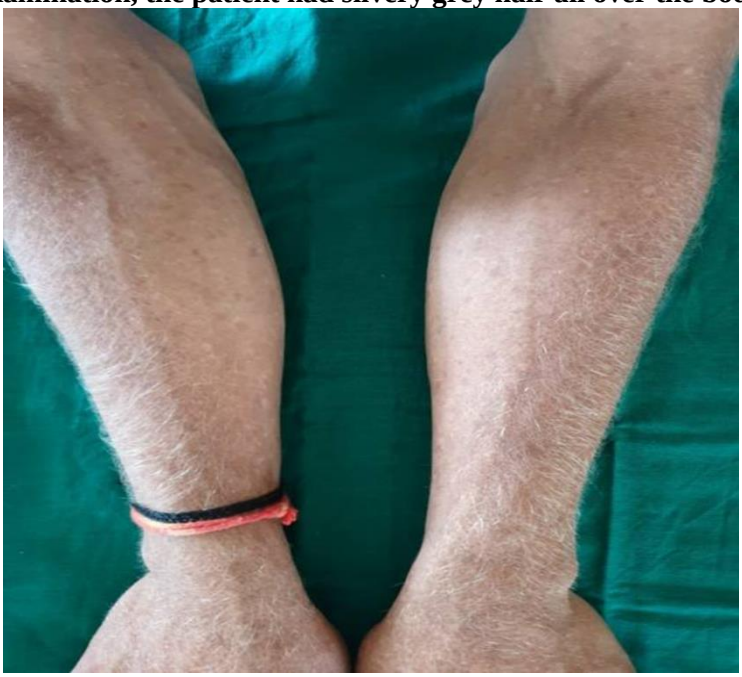


Figure 2: Bronze tan hyperpigmentation with intervening hypopigmentation over the whole body

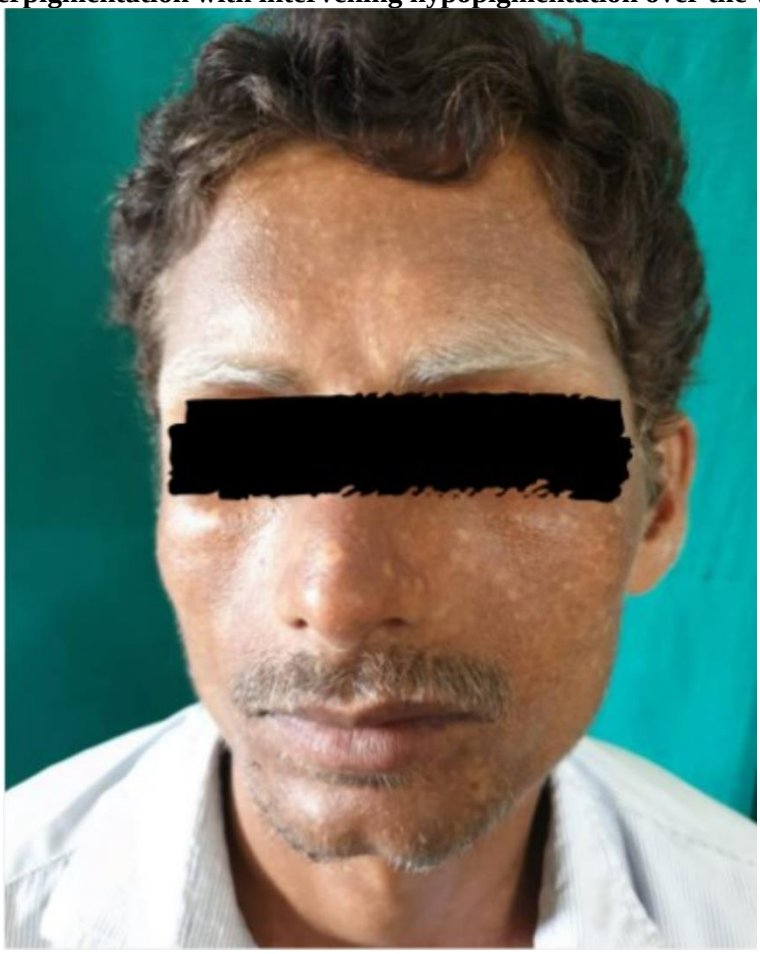
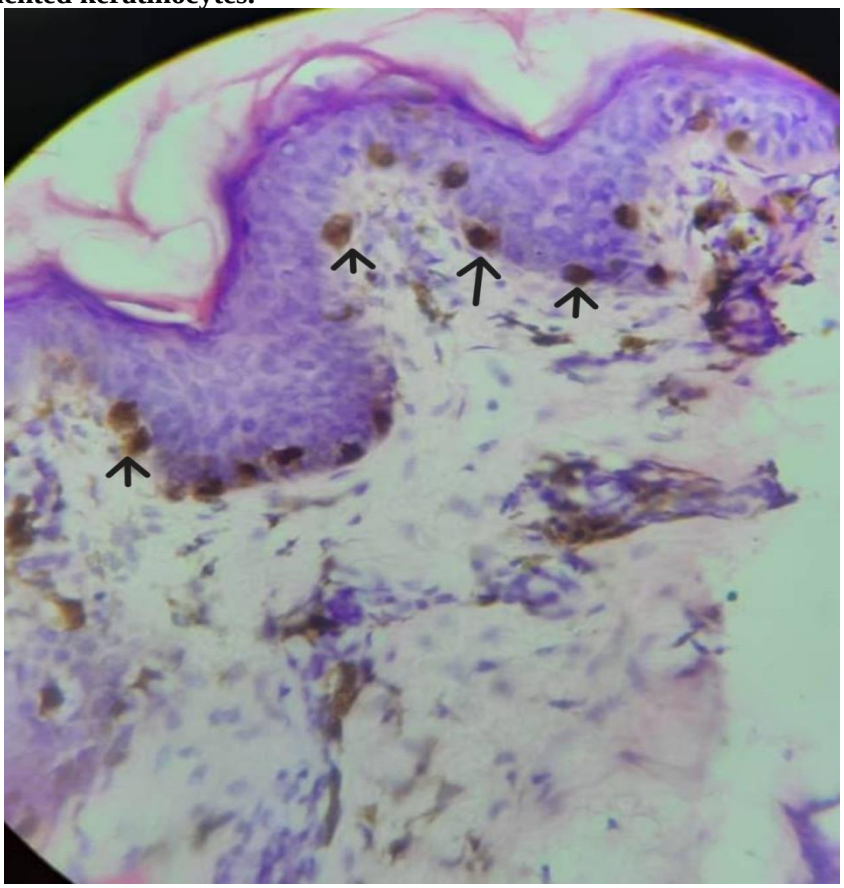


Figure 3: Light microscopic examination of hair revealed- uneven clusters of aggregated melanin pigment, accumulated mainly in the medullary area of the shaft.

Figure 4: Histopathological sections from the skin biopsy revealed stratified squamous keratinized epithelium with an increased number of melanocytes with aggregates of melanin pigmentation in the basal layer with adjoining poorly pigmented keratinocytes.



DISCUSSION:

Griscelli-Prunieras syndrome is a rare autosomal recessive disorder that associates hypopigmentation, characterized by a silver-grey sheen of the hair and the presence of large clusters of pigment in the hair shaft, and the occurrence of either a primary neurological impairment or a severe immune disorder. All genetic alterations associated with Griscelli-Prunieras syndrome result in defective transport of melanosomes and consequently abnormal accumulation of melanosomes in melanocytes. The transport of melanosomes from the cell center to the cell periphery involves bidirectional transport. This transport takes place along microtubule tracks with the help of the motor proteins, dyneins, and kinesins. Myosin Va (Myo5a), which is a processive motor protein, attaches to melanosomes through interaction with Mlph and Rab27a.[4] The tripartite complex, which is composed of Rab27a, Mlph, and Myo5a, has roles in vesicle transport and membrane trafficking processes.[5] GS3 is due to mutations in MLPH, whose protein product, melanophilin, serves to link myosin Va to Rab27a114. Because MLPH is expressed only in melanocytes, the associated phenotype is limited to diffuse pigmentary dilution and silvery hair without any systemic involvement. Melanophilin gene defect responsible for type 3 GS presents with hypopigmentation of skin and hair without any systemic involvement [6]. The prognosis for patients with GS3 is good, and these patients do not require any treatment. The prognosis for patients with GS3 is good, and these patients do not require any treatment.

CONCLUSION:

Based on characteristic clinical features and absence of neurological and immunological abnormality with characteristic microscopic findings of hair shaft and skin, the patient was diagnosed with Griscelli Syndrome III. Early diagnosis and treatment play a crucial role in a patient's survival. The characteristic phenotypic appearance, especially the pigment dilution of the patient's hair, is emphasized here.

REFERENCES:

1. Griscelli C, Durandy A, Guy-Grand D, Daguillard F, Herzog C, Prunieras M. : A syndrome associating partial albinism and immunodeficiency . 1978, 65:691-702. 10.1016/0002-9343(78)90858-6.
2. Cağdaş D, Ozgür TT, Asal GT "et al": Griscelli syndrome types 1 and 3: analysis of four new cases and long-term evaluation of previously diagnosed patients. Eur J Pediatr. 2012, 171:1527-31. 10.1172/JCI18264.
3. Sahana M, Sacchidanand S, Hiremagalore R, Asha G: Silvery grey hair: clue to diagnose immunodeficiency. Int J Trichology. 2012, 4:83-5. 10.4103/0974-7753.96910.
4. Ménasché G, Pastural E, Feldmann J, "et al": haemophagocytic syndrome.Nat. Genet. 2000, 25:173-6. 10.1038/76024.
5. Fukuda M, Kuroda TS, Mikoshiba K: Slac2-a/melanophilin, the missing link between Rab27 and myosin Va: implications of a tripartite protein complex for melanosome transport. J Biol Chem. 2002, 5:12432-6. 10.1074/jbc.C200005200.
6. Ménasché G, Ho CH, Sanal O, "et al": Griscelli syndrome restricted to hypopigmentation results from a melanophilin defect (GS3) or a MYO5A F-exon deletion (GS1). J Clin Invest. 2003, 112:450-6. 10.1172/JCI18264.