Meetings International

6th World Congress on
DERMATOLOGY

May 29-30, 2023 | Paris, France

A case of Griscelli Syndrome Type 3

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Back ground: Griscelli syndrome (GS) is a rare autosomal recessive multisystem genetic disorder which is characterzed by partial albinism of hair and skin along with neurological and/or immunological defects. Three types of this disorder are distinguished by its genetic cause and pattern of signs and symptoms . Patients with GS type 1 have primary central nervous system dysfunction, resulting from mutations in the MYO5A gene. Type 2 patients commonly develop hemophagocytic Lymphohistiocytosis, caused by mutations in the RAB27A gene, and type 3 have only light skin and silvery hair color resulting from mutations in the MLPH.

Case Description: Sixteen Years old and Fifteen years old non twin sisters, visited ALERT hospital OPD for the compliant of skin and hair discolorations since childhood. Physical examination of both sisters revealed silvery-gray hair including the scalp hair, eyebrows and eyelashes with diffuse hypopigmentation of skin. Their investigations revealed normal blood count, differential and peripheral blood smear. Both CT and MRI of the brain are normal for both sisters. Light microscopy examination of their hair showed large clumps of pigment irregularly distributed along the hair shaft. Histopathologic result from skin biopsy showed melanocytes at basal layer are enlarged and hyper pigmented with no or markedly reduced pigmentation of adjacent keratinocytes for both sisters. Genomic DNA sequencing of the sixteen years old girl revealed a homozygous single nucleotide transition ,c.104G>A, in exon 2 of MLHP. This change leads to an arginine substitution by glutamine, p.Arg35GIn ,with in the conserved Slp homology domain of melanophilin . Her sibling has the same homozygous missense mutation.

Conclusion: Only one pathogenic mutation in MLPH has been reported previously in Griscelli syndrome type 3.That mutation was P.Arg35Trp and was observed in a French-Turkish individual as well as several affected people in Arab pedigree. Functional studies confirmed that disruption of p.Arg35GIn in these two Ethiopian sisters is also pathogenic .Griscelli syndrome type 3 is not associated with neurological or immunological abnormalities.

Biography

Kidist Yeneneh, is a dermatologist at Department of Dermatovenerology. He graduated in Addis Ababa University in Ethiopia.

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