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A case of griscelli syndrome type 3

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Griscelli syndrome (GS) is a rare autosomal recessive melanocyte maturation disorder which is characterized 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.

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