Introduction

Griscelli Syndrome (GS), initially described by Griscelli and Pritzker in 1972, is an autosomal recessive disorder that causes partial albinism and silver-gray hair. GS has three variants each caused by different gene mutations. GS Type II involves a nonsense mutation of the GTP-binding protein RAB27A, a gene whose product is a RAB-associated protein. Type III is considered a member of the HLF (hemophagocytic lymphohistiocytosis) family. Both disorders are associated with defective genes which cause uncontrolled activation of T-Lymphocytes and macrophage, sequestering high amounts of inflammatory cytokines that can affect multiple organs including the liver, spleen, bone marrow, and CNS. However, RAB27A in the GTPase family was also understood to be involved in intracellular melanosome transport. As a result of this impairment of vesicular trafficking, clumps of melanocytes are deposited on the hair shaft.

Other differentials to consider in the patient with silver-gray hair should also include Chediak-Higashi and Elyjalde syndromes. Whereas all three syndromes consist of silver hair, similar age of onset, and autosomal recessive inheritance, diagnosis of GS can be parsed out through imaging studies, analysis of pathology specimens, and genetic analysis.

Abstract

Griscelli syndrome (GS) is a rare autosomal recessive disorder with fewer than 10 cases reported in the US. Type II has a poor prognosis and is fatal with early identification and intervention. Few descriptions are in the US literature, and we found no articles comprehensively describing the array of diagnostic tests that may help identify the disease.

We report an 11 month old Hispanic female admitted for intermittent fever, pallor, pancytopenia and massive hepatosplenomegaly. She was noted to have distinctive silvery gray hair. GS has three variants each with different clinical presentations. Type I presents with premature stop of the protein.

Results

We report an 11 month old Hispanic female admitted for intermittent fever, pallor, pancytopenia and massive hepatosplenomegaly. She was noted to have distinctive silvery gray hair. GS has three variants each with different clinical presentations. Type I presents with premature stop of the protein.

Evaluation included hair microscopy, skin and liver biopsy, bone marrow aspiration and biopsy, brain imaging and genetic testing.

- **2.7 weight deficit** showed multi-locally active plaque-like lesions.

- **Skin biopsy** showed epidermal vacuolization and large clumps of melanocytes.

- **Liver biopsy** showed evidence of hemophagocytosis.

- Genetic testing revealed new missense and frame shift mutation in RAB27A.