GRISCELLI SYNDROME - A RARE CASE REPORT

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ABSTRACT
Griscelli syndrome is a rare autosomal recessive disorder characterized by partial albinism with variable immunodeficiency. Silvery grey hair with large, clumped melanosomes on microscopy of hair shafts is diagnostic. A male child of 18 months presented with classic clinical features and confirmatory findings of clumped melanosomes on light microscopy of hair shaft. Skin biopsy was taken from back which on histopathological examination revealed large, clumped melanosomes in the stratum basale. Also seen were heavily pigmented hair shafts under light microscopy. The case was diagnosed as Griscelli syndrome.

Key Words: Silvery Grey Hair, Lymphohistiocytic Cells, Melanosomes, Partial Albinism and Stratumbasale

INTRODUCTION
Griscelli syndrome is a rare autosomal recessive disorder characterized by partial albinism with variable immunodeficiency. Silvery grey hair with large, clumped melanosomes on microscopy of hair shafts is diagnostic. The commonest complication leading to mortality includes lymphohistiocytic proliferation in various organs, including the brain. We present this case of ours in a child with classic clinical features and confirmatory findings of clumped melanosomes on light microscopy examination of hair shaft.

CASE REPORT
A one and half year male child presented with fever, cough, cold since 1 month. He was a product of consanguinous marriage; milestones were normal. Physical examination showed silvery grey hair, low set ears (Fig. 1). Systemic examination, revealed no hepatosplenomegaly. Complete hemogram showed Normocytic Hypochromic anaemia with raised ESR. Ultrasound abdomen examination was normal. Magnetic Resonance Imaging of brain was normal (T2 and FLAIR images show essentially normal signal intensity and morphology of the brain parenchyma; no obvious focal lesions were identified Fig. 2).

Figure 1: Child showing silvery grey hair, grey eyebrows and facial hair
Figure 2: Magnetic Resonance Imaging-Normal morphology of brain parenchyma with no focal lesion
Skin biopsy was taken from back. The skin covered biopsy measured 1.2x 0.6 cms. It was fixed in formalin and further subjected for paraffin embedding and routine processing.

**Light microscopy**

On histopathological examination revealed surface skin epithelium lined by stratified squamous epithelium with mild hyperkeratosis, thinned out at foci (Fig. 3). Stratum basaleshowed large, clumped melanophages (Fig. 4). Also seen were heavily pigmented hair shafts under light microscopy (Fig. 5).

**Figure 4:** H and E stained sections of the skin biopsy shows increased melanocyte pigment collections in stratum basale

**Figure 5:** H and E stained section (High power) - Arrow pointing towards clumped accumulated melanin pigment within the hair shaft
Fine, well-distributed pigment in normal controls (Fig. 6). Light micrographs of the hair, showing the large clumps of pigment irregularly distributed in the hair shafts of the patient (Fig. 7).

**DISCUSSION**

Griscelli syndrome is a rare autosomal recessive disorder characterized by partial albinism with variable immunodeficiency (Mancini et al., 1998). It is a fatal and rare disorder. The commonest complication leading to mortality includes lymphohistiocytic proliferation in various organs, including the brain. It is an uncommon disorder characterized by pigmented dilution and variable cellular and humoral immunodeficiency (1).: Griscelli et al., (1978) described two patients with partial albinism in 1978. As of January 2003, 60 cases have been reported by Scheinfeld & Johnson (2003). Most patients are diagnosed between 4 months and 7 years of age.

The genetic defects include mutations in either MYO 5A or RAB 27A (Sanal et al., 2002); both located on chromosome 15q21. Features include hepatosplenomegaly, silvery gray sheen to the hair, large clumped melanosomes in hair shafts, pancytopenia, hepatitis and immunologic abnormalities. It is characterized by partial albinism, variable cellular and humoral immunodeficiency and the occurrence of “accelerated phases” consisting of hemophagocytosis, pancytopenia, elevation of serum triglyceride levels, hypofibrinogenemia and hypoproteinemia (Aksu et al., 2003).

Dermatologic findings may be limited to hair, with skin and retinal pigmentation being occasionally affected. Microscopic examination of hair reveals uneven clusters of aggregated melanin pigment, accumulated mainly in the medullary area of the shaft.

Neurologic involvement with raised intracranial pressure, cerebellar signs, encephalopathy, hemiparesis, peripheral facial palsy, spasticity, hypotonia, seizures, psychomotor retardation and progressive neurologic deterioration is known (Hurvtz et al., 1993).

Immunologic abnormalities include natural killer (NK) cell function defect with absent delayed type hypersensitivity (Klein et al., 1994). Secondary hypogammaglobulinemia can occur in accelerated phases, which are characterized by lymphohistiocytic infiltration of various organs mimicking hemophagocytosis. These are thought to be associated with EBV infection, although various other viral and bacterial pathogens have also been incriminated (Klein et al., 1994). They result from abnormal cytotoxicity function of T and NK cells resulting from inability to secrete cytotoxic granules when the RAB 27A is not functional.

Differential diagnosis includes Chediak-Higashi syndrome (CHS) and Elejalde syndrome. CHS differs from GS by presence of abnormal giant cytoplasmic granules in leukocytes, more frequent cutaneous involvement, smaller, more evenly distributed pigment clumps in hair shafts and more consistent defective granulocyte activity (Göğüş et al., 1995).

The prognosis of patients with Griscelli syndrome is grave. Curative hope is offered only by bone marrow or stem cell transplantation, which is more effective when performed early in the course of the disease (Klein et al., 1994). Palliative management includes treatment of associated infections, and immunomodulatory therapy during accelerated phases (high dose systemic methylprednisolone, etoposide, intrathecal methotrexate, cytosine arabinoside and prednisolone, or ATG, cyclosporine and steroids (Gursey et al., 1994).

**CONCLUSION**

Herein, we report this case of Griscelli syndrome occurring in a child which is very rare. Differential diagnosis of immunodeficiency syndromes should be considered. Good clinical history, dermatologic and complete neurologic examination and awareness of this entity will aid in the final diagnosis.

**REFERENCES**


Case Report


