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A Case Report on Griscelli Syndrome

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Abstract: ***Background: Griscelli syndrome (Partial albinism with variable immunodeficiency) is an uncommon disorder characterized by pigmentary dilution and variable cellular and humoral immunodeficiency. It is inherited as an autosomal recessive disorder. Features include hepatosplenomegaly, silvery grey sheen hair, large clumped melanosomes in hair shafts, pancytopenia, hepatitis and immunologic abnormalities. The commonest complication leading to mortality includes lymphohistiocytic proliferation in various organs, including the brain. Case Report: A 8 months old female child brought by parents with compliants of fever since 2 months [on & off], abdomimal distension since 1 month. H/O developmental delay present [neck holding attained at 5th month, sitting with support not yet attained] H/O similar hair pattern to paternal uncle & grandmother. On examination vitals stable, febrile. Child was pallor with greying of hair and eyebrows, liver 2 cm below RCM, massive splenomegaly [8cm below LCM] firm in consistency.***

Keywords: griscelli syndrome, hemophagocytosis, Lymphohistiocytic proliferation, Silvery gray hair, hepatosplenomegaly

1. Investigations

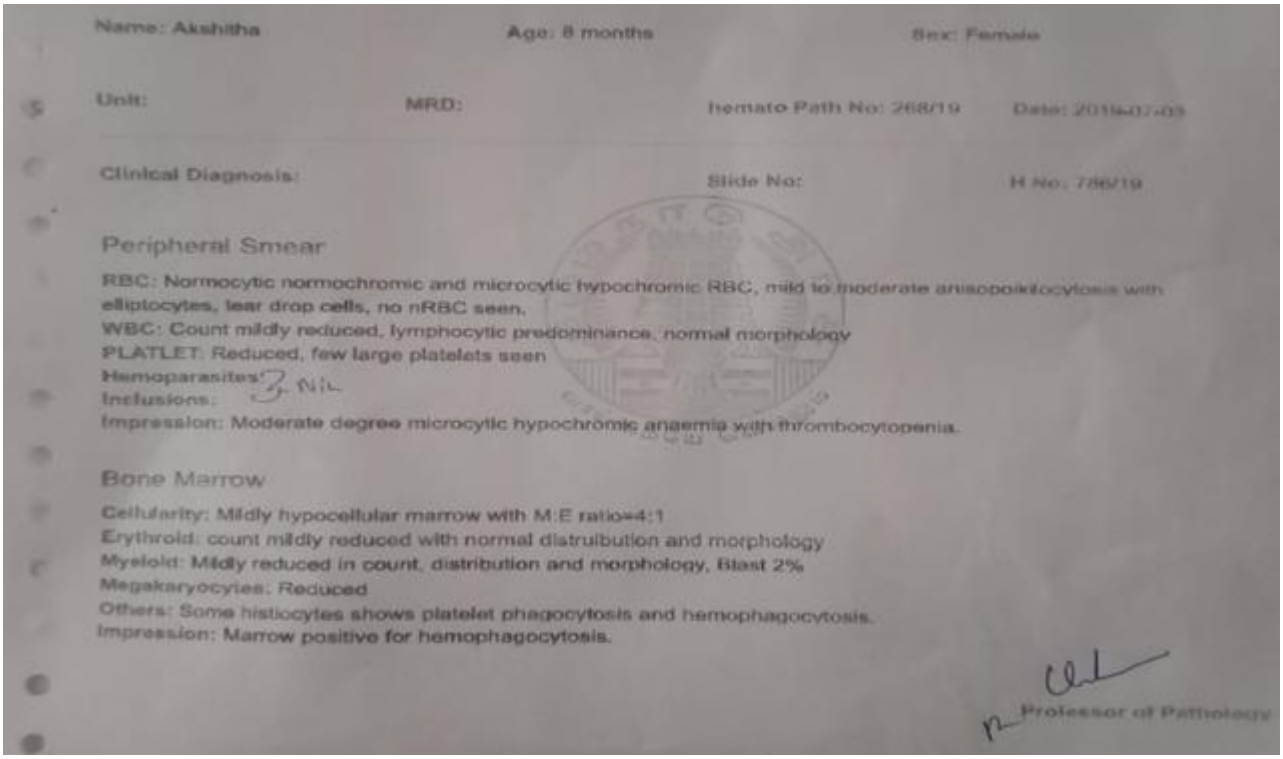
HB:-6.1g/dl, wbc count:-3300cells/cumm RBC count:-3.6M/cumm

Platelet Count: 0.54 lakhs/cumm, CRP:-23.80mg/l Routine fever investigations normal. Hypertriglyceridemia [629mg/dl], hypofibrinogemia [100mg/dl] and hypeferritinemia [1534 ng/ml].

USG Abdomen: mild hepatomegaly, moderate splenomegaly

Peripheral Smear: Moderate Degree Microcytic Hypochromic Anaemia with Thrombocytopenia

Bone Marrow Report: Positive for Hemophagocytosis.



Name: Akshitha Age: 8 months Sex: Female

Unit: MRD: Hemato Path No: 268/19 Date: 2019-07-03

Clinical Diagnosis: Slide No: H No.: 786/19

Peripheral Smear

RBC: Normocytic normochromic and microcytic hypochromic RBC, mild to moderate anisopoikilocytosis with elliptocytes, tear drop cells, no nRBC seen.
WBC: Count mildly reduced, lymphocytic predominance, normal morphology
PLATLET: Reduced, few large platelets seen
Hemoparasites: Nil
Inclusions:
Impression: Moderate degree microcytic hypochromic anaemia with thrombocytopenia.

Bone Marrow

Cellularity: Mildly hypocellular marrow with M:E ratio=4:1
Erythroid: count mildly reduced with normal distribution and morphology
Myeloid: Mildly reduced in count, distribution and morphology, Blast 2%
Megakaryocytes: Reduced
Others: Some histiocytes shows platelet phagocytosis and hemophagocytosis.
Impression: Marrow positive for hemophagocytosis.

Professor of Pathology

2. Discussion

Griscelli syndrome (Partial albinism with variable immunodeficiency) is an uncommon disorder characterized by pigmentary dilution and variable cellular and humoral immunodeficiency.

The genetic defects include mutations in either MYO 5A or RAB 27A, both located on chromosome 15q21(5,6). The hemophagocytic syndrome seen in GS-1 and GS-2 is also described as the accelerated phase

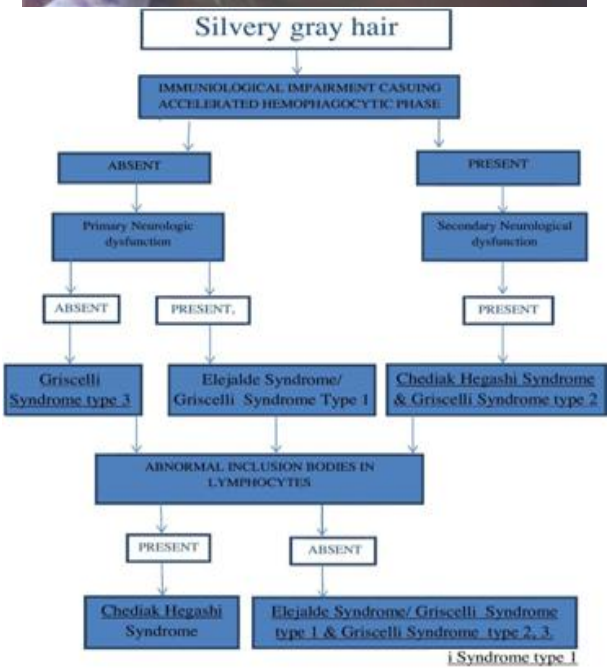
This is secondary to the uncontrollable T lymphocyte and macrophage hyperactivation. The main differentiation between GS-1 and GS-2 is the primary or secondary central nervous system (CNS) involvement.

The secondary CNS involvement in GS-2 is caused by the infiltration of lymphocytes and histiocytes as a result of hemophagocytic syndrome. Temporary remission of the

accelerated phase seen in GS-2 can be achieved with chemotherapy or immunotherapy. However, recurrent relapses of increasing severity are frequently observed as infiltration of the CNS despite maintenance therapy.

BMT is the only curative treatment of GS-2. Patients usually succumb to severe infection or CNS infiltration at an average age of 5 years without BMT. Thus prompt diagnosis is vital in the prognosis of GS-2.

Palliative management includes treatment of associated infections, and immunomodulatory therapy during accelerated phases.



i Syndrome type 1