Case Report :

Hypopigmented Hair, Severe Malaria and Immunedificiancy - Griscelli Syndrome

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Abstract:

Disorders of hypopigmentation and immune deficiency are rarely seen .

We report here a case of severe malaria with hypopigmented hair and immunodeficiency in four and half year old female child.her hemoglobin was 4.5 gm% ,absolute neutrophil count less than 1000with plasmodium vivax on peripheral smear. S he also had recurrent admission and treatment for pneumonia.Her immunoglobulins showed low IgM and IgG.Hair microscopy showed heterogenous clumps of chromatinmore in cortex.There was no evidence of HLH.

Key words: *case reports, immunedefiancy, hypopigmented hair, chromatin clumps.*

Introduction:

Disorders of pigmentation1 with immune deficiency of various types is rarely seen in clinical practice. Altered hair colour due to clumping of chromatin with other systemic features can be accepted as a marker of various syndromes. We report a case of similar nature with recurrent pulmonary infection, severe malaria, immunodeficiency with pigmentary dilution of hair and skin labelled as Griscelli syndrome2.

Case:

Four and half year old female child presented to our hospital with alternate day fever for 15days, progressive pallor and hepatosplenomegaly. Her blood count showed Hb was 4.5gm%, total count 5,500(N16, L83,E1), absolute neutrophil count less than 1000, Platelet adequate and peripheral smear showed trophozoites of Plasmodium vivax.

The child was admitted twice in different hospital with recurrent pneumonia in last two years and had history of multiple upper airway infection. Her nutrition parameter belongs to GDII malnution as per IAP guidelines.

Her blood album was 4.5gm%, globulin 1.6gm% and immunoglobulin profile showed high IgE, low IgM and IgG. Hair microscopy showed heterogenous clumps of chromatin more incortex less in medulla. No evidence of hemophagocytic syndrome3 with mildly elevated ferritin, triglyceride or abnormal coagulation. Peripheral blood granulocytes does not show any giant granules.

Discussion:

In tropical countries like India phenotypes like altered pigmentation of skin and its appendages commonly seen in Kala-azar, Thalasemia, tropical splenomegaly syndrome and Protein energy malnutrition. But in this case, disorder of pigmentation is by birth with immunedificiancy promted us for hair microscopy which revealed presence of large clumps of pigment in hair cortex signifying failure of homogenous and uniform distribution of chromatin. The gene responsible for this defect are MYO5A and RAB27A lies in chromosome number

15q21. Any mutation results in clustering of melanin pigment in hair shafts, accounting for pigmentary dilution, although melanin production is normal4.Three different variations of this syndrome is clinically seen, type 1 manifests with primary dysfunction of central nervous system, type 2 Griscelli syndrome commonly develops hemophagocytic lymphohistiocytosis (HLH) , type 3 manifests with merely partial albinism. Type 2 is more serious with uncontrolled activation of T-lymphocyte and macrophage activation syndromeusually results in death unless the child receives a bone marrow transplant. Presence of unusually persistant vivax malaria for 15 days also raised a valid question of altered immunity.

Absence of giant granules in peripheral blood granulocytes exclude this from Chediak Higassi syndrome whereas normal number and function of platelet ruled out Hermanesky Pudluck syndrome. Our case has immunodefiancy without HLH and no neurological finding suggests it can be a subtype of type 2. Genetic test can confirm type 2 by analysis of RAB27A gene is not done due to its high cost.

Contribution:

Diagnosis of case:Dr.Joydeep Das &Dr.Jyoti Kiran., Reference search: Dr.Suman Mandal. Histology of Hair: Dr.Swapan Sinha.Written the Manuscript: Dr.Balram Gupta.

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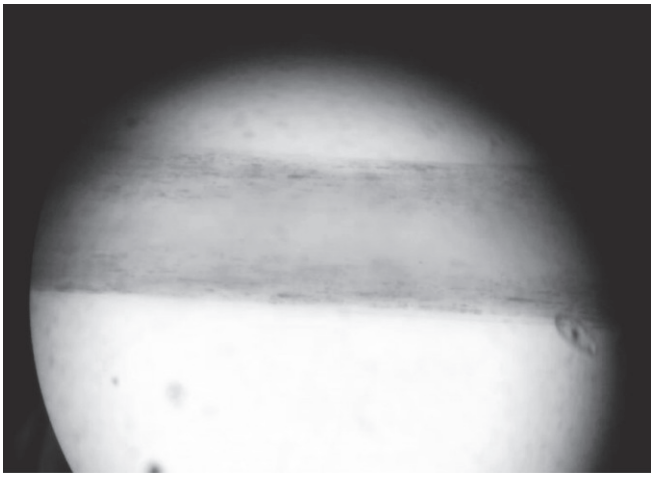

Fig. 1 Hair Microscopy

Fig. 2 Clinical Picture