LETTER TO EDITOR (VIEWERS CHOICE)

COMMON VARIABLE IMMUNODEFICIENCY ASSOCIATED WITH PARTIAL ALBINISM

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Primary immunodeficiency disorders associated with partial albinism include Chediak Higashi syndrome and Griscelli syndrome. (1) Vitiligo is described in association with common variable immunodeficiency (CVID), as a part of autoimmune phenomenon. (2) However the association of CVID with albinism is uncommon and no such association has been reported.

A 3 year old boy born to non consanguineous parents presented with failure to thrive, recurrent lower respiratory tract infections requiring hospitalizations and chronic recurrent diarrhea noticed for last 1 year. On examination his weight was 9.7 kg and height was 87 cm. His vital signs were normal. He had skin hypopigmentation suggestive of partial albinism with silvery greyish hair. Systemic examination was unremarkable. Investigations revealed hemoglobin of 11g/dL, total leukocyte count of 9300 cells/cumm (neutrophils 64%, lymphocytes 32%, eosino-phils 3%, monocyte 1%) and platelet count of 2.83 lakhs/cumm. Peripheral smear did not show any giant neutrophilic granules. Stool examination was normal and there was no steatorrhea. Urine culture grew Klebsiella pneumoniae with colony count of >105 CFU/ml. Abdominal ultrasonography done was normal. Mutational analysis for cystic fibrosis was negative. HIV ELISA done was negative. Chest x-ray and Mantoux test were negative. Serum immunoglobulin assay revealed pan-hypogammaglobulinemia with IgG <2.24 g/L (normal range 3.45-12.36), IgM 0.48 g/L (0.5-2), IgA< 0.1g/L (1.4-2.5), IgE< 0.10 IU/mL (0.1-200). (3) Serum total protein was 5.8g/dL and albumin was 3.8g/dL. Protein losing enteropathy was ruled out as there was no hypoalbuminemia and upper gastrointestinal endoscopy with biopsy was normal. There was no proteinuria. Nitroblue tetrazolium test was normal. Serum complement assay revealed C3, C4 and CH50 levels of 84 mg/dl, 68 mg/dl and 76 U/l respectively. Flow cytometric lymphocyte subset analysis revealed $CD3^+$T cells of 3320/cumm, $CD19^+$B cells of 1080/cumm, $CD4^+$T cells of 2150/cumm, $CD8^+$T cells of 1248/cumm and $CD3^+/CD16^+56^+$ NK cells of 550/cumm. Light microscopy of scalp hair shaft revealed melanin aggregates distributed in the medullary area. He was treated with 400 mg/kg/dose of human intravenous immunoglobulin (IVIG). Urinary infection was treated with cefoperazone-sulbactam and amikacin for 10 days. Parents were counselled regarding monthly IVIG therapy, supplementation of nutritious diet and vaccination specifically to avoid live vaccines. He was started on oral cotrimoxazole prophylaxis. Currently he is under regular follow up and is asymptomatic. As per dermatology advice regarding albinism, parents were instructed to avoid sunburn.

Diagnosis of CVID was entertained in the presence of hypogammaglobulinemia with normal lymphocyte subset on flow cytometry. Griscelli syndrome is characterized by partial albinism with variable cellular immunodeficiency. Hypogammaglobulinemia is usually not the primary feature in Griscelli syndrome. However it can occur in the accelerated phase of Griscelli syndrome characterized by lymphohistiocytic infiltration of various organs with lymphadenopathy and hepatosplenomegaly, which was absent in our case. Characteristic immunologic abnormalities include absent delayed-type cutaneous hypersensitivity and impaired natural killer cell function. Griscelli syndrome was not considered in our case as there was no organomegaly or lymphadenopathy with normal blood counts and flow cytometry. Bone marrow examination was not performed as parents did not give consent. Chediak Higashi syndrome was ruled out as hypogammaglobulinemia is not a feature and by the absence of giant neutrophilic granules. (4) Children with CVID require immunosuppressive therapy in addition to IVIG in the presence of autoimmune manifestations. (5)

Thus it is important to suspect primary immunodeficiency in a child with partial albinism in the background of failure to thrive and recurrent infections. Early diagnosis is required to prevent complications.

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