LETTER TO EDITOR (VIEWERS CHOICE)

A RARE CASE OF SEVERE COMBINED IMMUNO DEFICIENCY WITH SITUS INVERSUS TOTALIS

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A ten month old unimmunized boy, born of a 3rd degree consanguineous marriage, presented withrecurrent episodes of fever, cough and diarrhea since birth. His five elder siblings (four males and one female)had died of similar illnesses in infancy. On examination, weight was 5 kg (<5th centile), temperature was 102^0 F, he had tachycardia and marked tachypnea. Pallor, oral thrush, atrophic tonsils,bilateral purulent ear discharge, peri-anal excoriation, and intertrigo were present. There was dextrocardia with bilateral crepitations.Complete blood count revealed hemoglobin of 11.6gm/dl, white cell count of 7270/ cumm with 61% neutrophils, 22% lymphocytes (absolute lymphocyte count-1599/cumm),16% monocytes and 1% eosinophils. Ultrasonography (USG) showed an absent thymus, liver on the left and stomach on the right side. Echocardiography revealed small ostium secundum atrial septal defect (ASD). Flow cytometric lymphocyte subset analysis revealed $CD19^+$B lymphocytes of 416 cells/cumm, $CD3^+$ Tlymphocytes of 272 cells/cumm, $CD3^+/CD4^+$ Th lymphocytes of 208/cumm, $CD3^+/CD8^+$Tc lymphocytes of 32/cumm and $CD3^+/CD16^+56^+$ NKof 815/cumm. Nephelometric immunoglobulin analysis showed normal levels of IgG (2gm/dl), IgM (1.1gm/dl), IgA (1gm/dl) and IgE (3.87 IU/ml).A diagnosis of severe combined immunodeficiency (SCID) of $T^+B^+NK^+$ variety, most likely due to IL-7Rα deficiencywas considered. Features of situs inversus totalis (SIT) are commonly associated with immotile cilia syndrome (we could not do nasal cilia testing in our patient due to non-affordability) and not usually seen with SCID as was seen in our patient.He improved considerably with antibiotics. He was immunized with killed vaccines and started on cotrimoxazole prophylaxis. He required irradiated blood transfusionas he developed anemia during prolonged stay in the ward. Hematopoietic stem cell transplant has been advised but is proving to be difficult due to high cost.

This specific subtype of SCID is transmitted in an autosomal recessive fashion, as is SIT, although X linked pattern has been found rarely in SIT. The incidence of SCID is around 1:50,000-75,000 births. (1) It is a pediatric emergency and most patients present by 3 months of age with inevitability of death by 2 years of age. (2) Stem cell transplant is the treatment modality.Incidence of SIT is about 1:10,000. It is commonly associated with immotile cilia syndrome in which affected individuals suffer from chronic respiratory tract infections, a variable combination of infertility (males) and chronic ear infections. (3,4)

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References :

1 Puck JM. Population-based newborn screening for severe combined immunodeficiency: steps toward implementation. J Allergy Clin Immunol. 2007;120:760-768
2. Rosen FS. Severe combined immunodeficiency: a pediatric emergency. J Pediatr. 1997;130:345-346
3. Sharma S, Chaitanya KK, Suseelamma D. Situs Inversus Totalis (Dextroversion) - An Anatomical Study. Anat Physiol.2012; 2:112
4. Schidlow DV. Primary ciliary dyskinesia (the immotile cilia syndrome). Ann Allergy. 1994;73:457-68

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