TEACHING FILE

GRAND ROUNDS

Ira Shah

THROMBOCYTOPENIA IN AN INFANT

Case:- A 4½ month old boy born of 3rd degree consanguineous marriage presented with intermittent fever and gradual distension of abdomen since 2 months and self limited oral ulcers 7 days back. He was an elective caesarean section delivery in view of breech presentation with a birth weight of 2.7 kg and no antenatal or post natal complications. He is on exclusive breast feeds and immunized till date. He has achieved milestones normally. Father had tuberculosis 8 years back. On examination, the child was well nourished (Weight = 5.75 kg, Height = 62 cm) and had no lymphadenopathy, jaundice or edema. On systemic examination, he had splenohepatomegaly. Other examination findings were normal.

Investigations showed:
- Hemoglobin = 11.1 gm/dl
- WBC count = 7,800/cumm [polymorphs 14%, lymphocytes 80%)
- Platelet count = 83,000/cumm
- SGPT = 49 IU/L
- Total proteins = 6.3 gm/dl, Albumin = 3.6 gm/dl
- S. Bilirubin = 0.6 mg/dl
- USG Abdomen = Splenohepatomegaly
- TORCH titres = Negative
- X-Ray Chest = Normal
- Bone marrow examination – Megakaryocytes reduced
- HBsAg, Anti HCV, VDRL = Negative
- Bone marrow G-CSF study = Normal
- Karyotype = 46 XY (No breaks)

What further tests should be done to diagnose this child ?

Expert's opinion:- This child has presented with splenohepatomegaly since 2½ months of age and has thrombocytopenia. Hence, one would consider a diagnosis of
- Congenital infection
- Leukemia
- Storage disorder

The most likely malignancy that would present with a chronic course of 2 months with predominant splenomegaly and thrombocytopenia would be Juvenile Meta Myeloid Leukemia (JMML) However, in this child, bone marrow examination as well as bone marrow G-CSF studies are normal. Hence JMML is ruled out. Other possibility that may be associated is monosomy 7 that can present the same way which also has been ruled out in view of normal karyotype.

A storage disorder such as Gaucher's disease and Niemann Picks disease can present with splenohepatomegaly. However, bone marrow examination would have picked up the foam cells/ storage cells which in this child were absent and hence storage disorder also seems unlikely.

Congenital infections can present with bone marrow involvement as well as organomegaly at this age group. However in this child TORCH titres, HBsAg, Anti HCV and VDRL were negative ruling out the common causes of congenital infections. However, HIV is one infection that can cause thrombocytopenia as well as hepatosplenomegaly. Infact, it has been found that almost 8-10% of patients with Idiopathic thrombocytopenic purpura (ITP) have HIV infection. Hence, this child should be tested for HIV.

The parents were counseled for the same and HIV DNA PCR was positive in this child with a viral load of 1,20,000 copies/ml. Both parents were also subsequently tested and found to be HIV infected.

This child was started on HAART and after 6 months, his viral load was undetectable.

Hence, in a child with hepatosplenomegaly and thrombocytopenia, always rule out HIV infection.

E-published: October 2011. **Art**#69

A 10 MONTH OLD WITH RECURRENT INFECTIONS

Case:- A 10 month old girl born of non consanguineous marriage presented with not gaining weight, recurrent loose motions and fever since 3 months of age. She has been treated for the same several times but there has been no response. She was a full term home delivery and was on exclusive breast feeds till 5 months of age following which weaning was started. At present child is on breast feeds and normal diet. She has delayed milestones and has achieved only head holding and social smile. She has an elder brother who is 2 years hold and is normal. On examination, she was wasted (weight = 3.5 kg, length = 60 cm), had oral thrush and loss of subcutaneous fat. There were no lymphadenopathy and tonsils were absent. Systemic examination was normal.

What is the diagnosis?

Expert's opinion: This child has failure to thrive with recurrent infections. Hence an underlying immune dysfunction should be ruled out such as primary immunodeficiency and HIV. Since the onset of problems has been since 3 months of age, it suggests a problem in the T cells. (B cell defects usually present after 6 months of age due to relative protection by maternally acquired antibodies). T-cell defects present in the peri-neonatal period. Acquired immunodeficiency is a possibility but they usually have other features such as lymphadenopathy and hepatosplenomegaly. In this child absence of tonsils and Lymphnodes suggests poor developed centers for T cells and a T cell defect such as severe combined immunodeficiency (SCID) is a possibility. Agammaglobulinemia can also present with absent tonsils and lymph nodes. SCID can be

confirmed by absent thymus and decreased T cells on flow cytometry (poor CD3, CD4 and CD8) cells lineage. In this child CD3, CD4 and CD8 were deficient with decreased CD19 levels suggestive of T^-B^- SCID.

Severe combined immunodeficiency syndrome (SCID) is a rare genetic disorder characterized by defective or absent T cell and B cell function. They usually present in the first 6 months of life with sepsis, disseminated tuberculosis following BCG vaccine, candidiasis, pneumocystis carinii pneumonia, severe viral infections, chronic diarrhea, failure to thrive and malabsorption. It is classified into 2 major groups: those without T cells and B cells (T^-B^-) and those with B cells (T^- B^+). Most patients with SCID have thymic hypoplasia and small, poorly developed lymph nodes and tonsils and thus absent tonsils as well as poorly formed lymphnodes could be clinical markers of SCID in an infant with recurrent infections. SCID is often fatal within the first year of life unless rescued with bone marrow transplant or curative hematopoietic stem cell transplant.

E-published: November 2011. **Art**#76

HERPES ZOSTER

Case Report:- A 9 year old boy born of non consanguineous marriage presented with herpes zoster over left hand since 5 days. He had renal calculi at 5 years of age. He had no other problems since then. Mother had died immediately post his birth and the father was asymptomatic. He had an older sister aged 11 years who had TB a few years back. On examination, he was averagely nourished (weight = 20.5 kg and height = 121.8 cm), had vesicular lesions over left hand and other systems were normal.

What further tests are required?

Expert's opinion: Herpes zoster is caused due to reactivation of varicella virus. It is commonly seen in geriatic population or patients with debilitaling disease with fall in immunity. It is not seen commonly in children. If herpes zoster occurs in a child, then one must suspect a poor immunity and workup the child for the same. Among various immunodeficiencies; acquired immunodeficiency is still the commonest. Congenital immunodeficiencies usually present in infancy. Herpes zoster being a virus depends on T cell mediated immunity and all congenital T-cell mediated immunodeficiencies present early in life and not at 9 years of age. Also with a family history of death of the mother and tuberculosis in elder sister, the child should be screened for acquired immunodeficiency. This child's HIV ELISA was done which was positive and reconfirmed by another kit. The sister was also HIV infected. Thus, always rule out an underlying immune disorder in a child if there is herpes zoster especially if it is multi-dermatomal.

E-published: December 2011. **Art**#81

From: Medical Sciences Department, Pediatric Oncall, Mumbai.

Address for Correspondence Dr Ira Shah, 1/B Saguna, 271, B St Francis Road, Vile Parle (W), Mumbai 400056.