A 3 YEAR OLD GIRL WITH CHRONIC DIARRHEA
A 3 year old girl born of non-consanguineous marriage presented with watery diarrhea since 1 month. The diarrhea initially was blood stained and then foul smelling, greasy with a frequency of 12-15 times/day. There was a weight loss of 2 kg in last one month. She had history of right-sided otorhoea 1½ year back. There was no history of similar complaints in the past, no contact with tuberculosis and similar history in the family. She was treated for same with oral antibiotics but there was no response. On examination, she was malnourished (Weight = 8 kg, less than 5th centile, Height = 87 cm (less than 5th centile)) with no significant examination finding. Her investigations showed:

- Stool examination – fat globules
- Urine examination – Normal
- Mantoux test – Negative
- HIV ELISA – Normal
- S. electrolytes, blood gases – Normal
- Hemoglobin = 8.2 gm/ dl
- WBC count = 21,600/ cumm (59 percent neutrophils, 40 percent lymphocytes)
- ESR = 6 mm at end of 1 hour
- Stool for cryptosporidia – multiple oocysts seen

She was treated with Nitazoxanide to which she responded.

How should she be investigated further?

Expert Opinion: Cryptosporidiosis is usually a protozoal organism that causes disease in an immunocompromised person. It can also occur in immunocompetent children. It can lead to persistent unremitting diarrhea which may be difficult to treat. Whenever cryptosporidium are found in a patient, the patient should be investigated for an underlying immunodeficiency. This child’s HIV ELISA is negative. Cryptosporidium is usually seen with T-cell defects. Hence this child should undergo a CD lymphocyte analysis and serum immunoglobulins. The immunoglobulins were normal in this child. CD lymphocyte subset showed normal T-cell counts.

Cryptosporidium is a parasite that is acquired by drinking contaminated water. Chlorination does not eradicate the organism and thus it is commonly seen in lakes, rivers, recreational pools and swimming pools. Drinking this contaminated water leads to ingestion of the oocyst that will invade the jejunum and release sporozoites that act on the intestinal mucosa leading to watery diarrhea. Cryptosporidium is usually eradicated by boiling and filtering the water.

Treatment is Nitazoxanide. However it may not be very useful in immunocompromised individuals. In such patients, the treatment of choice is immune reconstitution. Other drugs that have been tried are Pararomycin and azithromycin.

FEVER, COUGH, CONVULSION AND HEPATOSPLENOMEGALY
Clinical Problem: A 4½ months old boy born of non consanguineous marriage presented with fever and cough for 15 days. He was treated with IV antibiotics and nebulization for same. He also had an episode of uprolling of eyes 3 days ago for which he was treated with Diazepam. His eldest sister had died at 18 years of age due to fever and cough 4 months ago. The child was a full term normal delivery with birth weight of 4 kg on exclusive breast feeds and had received only oral polio vaccine. On examination, the child had tachycardia (heart rate = 120/ min), respiratory rate = 44/ min with subcostal and intercostals retractions and chest indrawing. Weight was 7 kg and height was 63 cms. On respiratory system, there was bilateral rhonchi with minimal chest findings. He had hepatosplenomegaly. Other systems were normal.

What is the diagnosis?

Expert Opinion: This child has presented with fever, cough for 15 days suggestive of a respiratory infection. Respiratory distress with indrawing suggests hypoxia. Thus the child has an infection that is leading to hypoxia. Thus most common cause would be either in the alveoli or the parenchyma leading to V/Q mismatch and hypoxia. Thus, one would consider pneumonia or interstitial lung disease. Since chest findings are minimal on examination it would suggest and interstitial lung disease. Common interstitial lung diseases, one would consider are:

- Miliary TB
- Pneumocystis carinii pneumonia
- Fungal infections
- Interstitial pneumonitis like UIP, LIP, DIP
- Sarcoidosis
- Histiocytosis

This child also has a convulsion. The convulsion could be due to infection in the brain such as meningitis or due to hypoxia. With meningitis one would consider tuberculosis as an etiology. Hepatosplenomegaly would
be seen with TB, and sarcoidosis or histiocytosis. Thus, one would consider a differential diagnosis of disseminated TB, histiocytosis or sarcoidosis in this child. On investigation, Chest X-Ray shows miliary reticulonodular shadows, Mantoux test was positive and CSF showed 45 cells with 100 percent lymphocytes and sugar of 60 mg percent. Thus, child was diagnosed as Tuberculosis and treated with 4 drugs ATT and steroids.

**What is the possible diagnosis?**

This child has presented with recurrent seizures. In addition, the child has dysmorphic features such as microphthalmia, microcornea and microcephaly suggestive of either an intrauterine infection or genetic disorder. The commonest IU infection in this child would be congenital rubella and seizures could be due to brain calcifications, malformations. In this child, urine for rubella was positive suggestive of diagnosis of congenital rubella. Echocardiography was normal. ECG showed sinus bradycardia.

**MICROPHTHALMIA, MICROCORNEA, LOW SET EARS**

Clinical Problem : A 4 year old girl presented with recurrent seizures since 1 year of age which are generalized tonic clonic convulsions with clenching occurring once in 2-3 months. Mother had appendicitis in first trimester of pregnancy. A milestone in form of speech is delayed. The child is a full term normal delivery with birth weight of 2.2 kg. On examination, the child has microphthalmia, microcornea, low set ears, microcephaly (head circumference = 44 cm). There was an ejection systolic murmur in pulmonary area. Other systems are normal.