TEACHING FILE

GRAND ROUNDS

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HEMOPHILIA

Clinical Problem: A 10 years old boy who is a diagnosed case of hemophilia (Factor VIII levels <1 percent) not on regular Factor VIII therapy presented with abdominal pain and black stools since 1 day. There is no fever or bleeding from any other site. On examination, he was pale, had tachycardia (Heart rate = 130, min) with bilateral basal crepitations and hypotension (blood pressure = 80/60 mm of Hg). He had tender hepatomegaly and no bruises or deep tissue bleeding. Other examination findings were normal. Investigations showed hemoglobin of 3.7 gm/dl, WBC count of 22,700/cumm (70 percent polymorphs, 30 percent lymphocytes) and normal platelet count. Partial thromboplastin time (PTT) was 87 seconds and prothrombin time (PT) was normal. He was treated with IV Fluids and dobutamine. Factor VIII concentrate could not be given due to unaffordability.

To stop bleeding, what can be given – cryoprecipitate or fresh frozen plasma?

Expert Opinion : This child was severe hemophilia and for abdominal bleedings leading to congestive cardiac failure, ideal replacement should be recombinant factor VIII concentrates (50 IU/kg). However if recombinant factor VIII is not available, alternative in form of blood component therapy should be used. Cryoprecipitate is the preferred blood component as it is rich in Factor VIII whereas fresh frozen plasma (FFP) is not useful as it contains decreased Factor VIII. Thus giving FFP is not going to make much of a difference.

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CATARACTS

Clinical Problem : A 3 year old male child born of non consanguineous marriage presented with bilateral cataracts since 10 months. Birth history and milestones were normal. There was no jaundice, failure to thrive or dysmorphic features. On examination, apart from bilateral white reflex, other systems were normal.

How to approach such a case?

Expert Opinion : Causes of cataracts in a child are varied and include intrauterine infections, genetic disorders, metabolic disorders, hypoparathyroidism and even prematurity.

This child has no prematurity, dysmorphic features or delayed milestones. Thus, most likely cause of cataract in this child would be metabolic disorder. Common metabolic disorders leading to white cataract are:

- Hypoparathyroidism
- Galactosemia
- Lowe's syndrome
- Diabetes mellitus

In this child, since there is no jaundice or hepatomegaly, galactosemia type 1 and 3 seem unlikely. Lowe's syndrome is associated with RTA and mental retardation. Diabetes mellitus would have additional features of polyuria, polydipsia. Thus in this child, one must rule out Hypoparathyroidism. The calcium, phosphorus, alkaline phosphatase in this child was normal. Galactosemia type 2 is a possibility and one must do the galactokinase enzyme levels. Galactosemia workup for galactokinase deficiency was positive.

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ENCEPHALOPATHY

Clinical Problem : A 10 month old boy born of non consanguineous marriage presented with fever for 5 days, refusal of feeds for 3 days and drowsiness for 1 day. On examination, vital parameters were normal. He had pallor, drowsiness and hepatosplenomegaly. There was no icterus. Deep tendon reflexes were brisk and tone was normal. There was no focal neurological deficit, meningeal signs and other systems were normal. Investigations showed:

- Hemoglobin = 8.4 gm/dl
- WBC = 5,500/cumm (32 percent neutrophils, 67 percent lymphocytes, 1 percent eosinophils)
- Platelet count = 57,000/cumm
- S.electrolytes = Normal
- SGOT = 244 IU/L, SGPT = 138 IU/L
- Bilirubin = 0.8 mg percent
- Total proteins = 5.8 gm percent (albumin = 2.8 gm percent)
- No acidosis
- Blood sugar = 45 mg percent
- Serum ammonia = 465 µg/dl
- CRP = Negative
- Urine = Normal. No Ketones
- PT = 16.2 sec (elevated), PTT = more than 2 min.

Patient was started on IV Fluids, lactulose and metronidazole and responded within 24 hours. His sensorium improved and abnormal parameters improved in next 3 days. However, hemoglobin dropped from 8.4 to 7.5 gm percent and platelet count dropped to 8000, cumm.

What is the diagnosis?

Expert Opinion : This child has presented with an encephalopathy (altered sensorium). She has fever for 5 days; one must consider an acute infective cause. The child does not have any meningeal signs, hence meningitis seems unlikely. One would consider a diagnosis of encephalitis (especially

viral) as a possibility. However the child also has hepatosplenomegaly, thus other differential diagnosis such as cerebral malaria, hepatic encephalopathy and Reye's syndrome should be considered. Bilirubin is normal in this child and there is hypoglycemia with elevated liver enzymes and hyperammonemia leading to suspicion of Reye's syndrome. Reye's syndrome may be seen with metabolic disorders such as Fatty acid oxidation defects or can also occur due to viral infections. This child has prolonged prothrombin time and partial thromboplastin time along with thrombocytopenia and hemoconcentration (Hemoglobin dropped from 8.4 to 7.5 gm percent after starting IV fluids). Hence one must consider viral hemorrhagic fever such as Dengue in this child which leads to hepatitis as well as encephalopathy especially DEN-2 (Dengue 2) virus is hepatotrophic). In this child, Dengue IgM was positive.

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