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Case Note

Hereditary spherocytosis

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Abstract
Hereditary spherocytosis is a congenital haemolytic anaemia due to defect in spectrin-a RBC membrane protein and is transmitted as autosomal dominant. Due to this defect there is presence of characteristic spherical cell in peripheral blood smear and osmotic fragility is increased. Haemolytic anaemia, reticulocytosis, jaundice and splenomegaly are present. This article reports a case of a 9 year old boy who presented with a history of prolonged jaundice since the age of 4 years and recurrent pain in the right upper quadrant of abdomen. Clinical examination revealed jaundice, enlarged liver and marked splenomegaly. Investigations confirmed the diagnosis of hereditary spherocytosis by the presence of spherocytes in blood smear, raised reticulocytes and increased osmotic fragility. The patient was subjected to splenectomy after vaccination against coccobacillus and was discharged after proper advice and on post splenectomy antibiotic prophylaxis.

Key words: Prolonged jaundice, microspherocytes, splenectomy, prophylaxis

Case report

A 9 year old male, the first child of non - consanguineous parents from Nawalparasi was admitted in paediatric ward of Kathmandu Medical College Teaching Hospital with complaints of yellow discolouration of eyes and recurrent pain in upper quadrant of abdomen for the last 4 years. Yellow discolouration was waxing and waning type, not associated with itching of body, normal stool colour no blood in stool, no vomiting, no history of blood transfusion or intravenous drug use in the past. There was no family history of congenital jaundice. The child had neonatal jaundice and did not require any treatment.

On examination the height was 124cm and weight 24 kg. The vital signs were normal with respiratory rate 30/min, heart beat 110/min. and blood pressure recorded was 100/60mm of Hg. There was moderate anaemia and jaundice but no oedema, ascites or lymphadenopathy. Systemic examinations were within normal limits except abdominal examination which revealed enlarged liver of 2cm and splenic enlargement of 8cm below costal margins and was non tender, firm and with irregular margin.

Test results were as follows:

Haemoglobin 5.6g/dl, white cell count 4000 with P 58, L 31, E 6, M5, RBC count 2270000/mm3, platelets 220000/mm3, ESR 67mm/hr, Retic.19%,PT 16.6 seconds, total bilirubin 4.5 mg/dl (direct – 0.3mg/dl),liver enzyme and viral markers-SGOT 351 IU/l, SGPT 40 IU/l. alkaline phosphatase 110 IU/l, HbsAg and AntiHCV both negative. Peripheral smear revealed hypochromasia, microcytes, macrocyte, anisocytosis, poikilocytes and moderate spherocytes and adequate platelets. There were no abnormal haemoglobin and direct Coombs test was negative. Urine and stool tests were normal and chest X-Ray showed no abnormality. K-39 test for Kala-azar was negative and malarial parasite was not seen in peripheral smear. Abdominal Ultrasonogram and CT Scan showed mild hepatomegaly and massive splenomegaly. Osmotic fragility was increased initially from 0.65% to a total of 0.3%. HB electrophoresis revealed HbA band. Bone marrow aspiration study showed increased erythroid series. The diagnosis of hereditary spherocytosis was made and the child had splenectomy after proper vaccination against pneumococci, meningococci, H. influenza and chickenpox. The spleen removed measured 17x11cm and biopsy showed thickened capsule with widening of splenic cords which were congested and with erythrophagocytosis. The sinuses were empty and increase in connective tissue fibres were seen focally.

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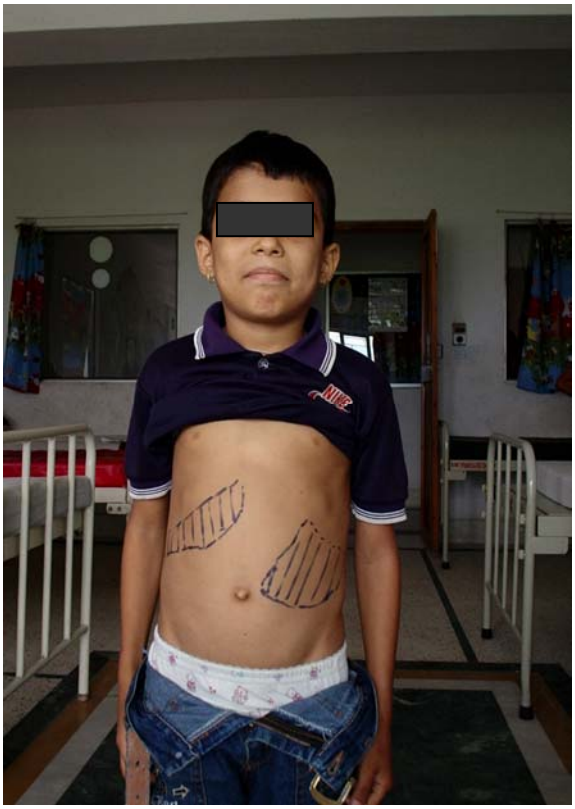

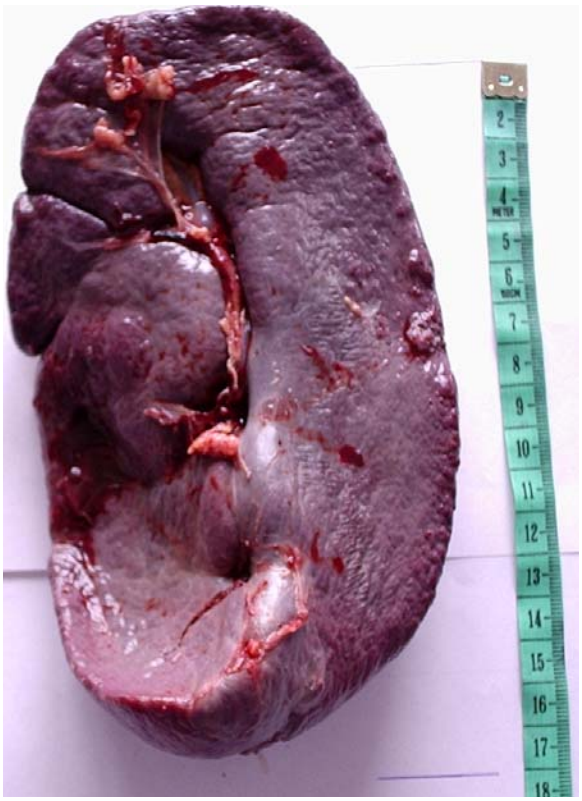

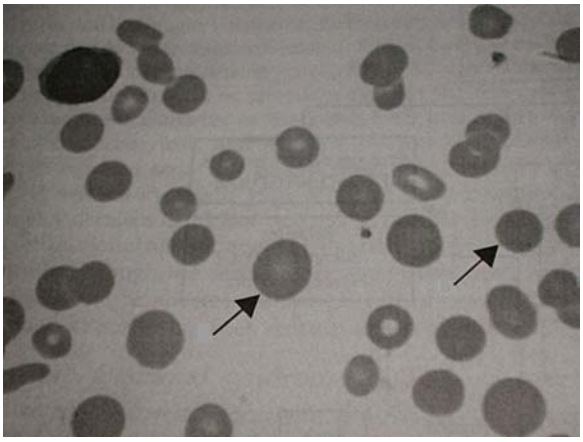

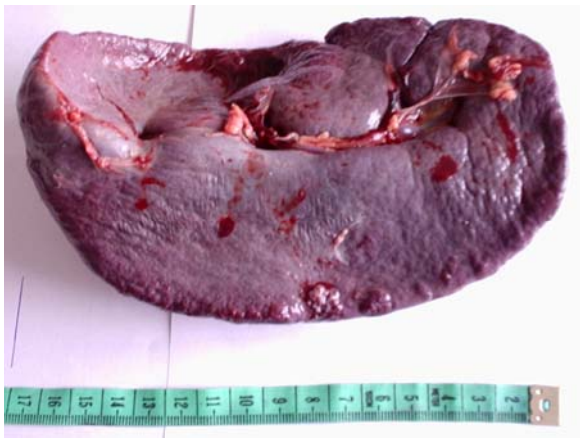

Discussion

Hereditary spherocytosis is a disorder of RBC membrane leading to chronic anaemia and a common cause of haemolysis and haemolytic anaemia. It is the most common familial and congenital abnormality of RBC membrane. The prevalence of this disease in Nepal is not known but in northern Europe prevalence is 1/5000. Most commonly it is an autosomal dominant inheritance, recessive being less frequent. In most patients spontaneous mutations are more common. Deficiency of the skeletal protein of cell membrane – spectin, ankyrin and actin one or all of them are responsible for the spheric shape of RBC.

Clinical feature of hereditary spherocytosis

Milder types are asymptomatic till adulthood where as the severe type presents with signs of haemolytic anaemia in newborn with anaemia and hyperbilirubinemia in the first 24 hrs of life.

Severe anaemia, jaundice, splenomegaly (only after infancy) and cholelithiasis (pigmentary gall stone, as early as 4-5 yrs. in approx 50% of the patient), are the clinical features of this disease.

They are more susceptible to aplastic crisis due to any infection and more so with parvo viral infection. Chronic haemolysis results in formation of cholelithiasis.

RBC is spherical in shape and smaller in diameter than normal, hyperchromic as a result of higher haemoglobin concentration, more rigid and less deformable than normal RBC with increased permeability of cell membrane to cations.

Haemoglobin is decreased in the range of 6-10 gm/dl but may be normal.

ESR may be normal or increased because of anaemia.

Reticulocyte count ranges to 6- 20% (mean 10%) indicative of haemolysis

MCV is normal and MCHC is increased (36–38 gm/dl).

In the peripheral smear reticulocyte are seen (polychromatophilic) and the shape of spherocytic RBC are predoninent. Platelet count and prothrombin time are within normal limits.

Direct Coombs test is negative in hereditary spherocytosis and this rules out autoimmune haemolytic anaemia.

Bone marrow is consistent with anaemia because of erythroid hyperplasia. High RBC turnover is reflected by the increased erythroid series in the bone marrow.

In osmotic fragility test spherocytic RBC, when exposed to hypotonic saline, swells up easily and lyses more readily than normal. Spherocytes tolerate less well to hypotonic solution with consequences of lysis than normal biconcave RBC.

Liver function tests shows unconjugated hyperbilirubinemia with normal liver enzymes Urobilinogen may be increased in the urine, which also indicates presence of unconjugated hyperbilirubinemia.

Sonogram and CT scan of the abdomen and pelvis shows splenomegaly, mild hepatomegaly and in few cholelithiasis.

Treatment

In newborn period the treatment is phototherapy and repeated blood transfusion to the children presenting with severe anaemia.

Splenectomy is the definitive treatment of this disease, which should be performed after the age of 5 years to avoid increased risk of post splenectomy sepsis. Cholecystectomy is also indicated at the same time of splenectomy as the patient is very prone to develop cholelithiasis.

Indication for splenectomy

1. Severe anaemia
2. Reticulocytosis >10%
3. Repeated hypoplastic or aplastic crisis
4. Faltering growth

Management of the splectomised patient

1. Vaccination with multivalent pneumococcal and haemophilus vaccines, at least 2-3 weeks before elective splenectomy, is given. Meningococcal A C vaccination is only recommended for those living in areas with high endemic infection rates.
2. Life-long penicillin V 250 mg 12-hourly is recommended to protect against bacterial strains not covered by the vaccines. If allergic to penicillin a macrolide is considered as alternative to it.
3. Wherever possible, splenectomised patients should carry a card or bracelet indicating the date of their last vaccinations. In the event of overwhelming sepsis, this card may be life saving in unconscious patients by administrating rapid IV administration of appropriate antibiotics.

4. Splenectomised patients admitted with septicaemia should be resuscitated and given intravenous antibiotics to cover pneumococcus, haemophilus and meningococcus.
5. Animal bites should be promptly treated with local disinfections and antibiotics to prevent serious soft tissue infection and septicaemia.

Conclusion

Hereditary spherocytosis is a rare haemolytic anaemia in Nepal. Its incidence has not been derived so far. Milder type may be asymptomatic and can have normal life but their haemoglobin level will remain on the lower side of normal limits, whereas severe type of hereditary spherocytosis needs definitive treatment in order to avoid chronic anaemia. Repeated blood transfusion is the treatment till the final decision of the splenectomy is taken. It has to be preceded by series of vaccinations. After the splenectomy the child needs life long prophylaxis against the encapsulated organism.

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