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A Case Study on Hereditary Spherocytosis in A 7 Year Male Child

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Abstract: ***Background: Hereditary spherocytosis (HS) is the most common inherited hemolytic anemia among individuals of Northern European descent, but it is increasingly recognized in the Indian pediatric population. However, epidemiological and genetic data from India remain limited. Objectives: This review summarizes the clinical profile, diagnostic modalities, genetic mutations, and treatment outcomes of hereditary spherocytosis in Indian children. Methods: A review of published Indian studies from tertiary care centers and genetic research institutions was conducted, emphasizing pediatric patients diagnosed with HS. Results: The clinical spectrum includes anemia, jaundice, splenomegaly, and growth failure. Mutational analysis shows ANK1 and SPTB as the most commonly affected genes. Splenectomy remains a key treatment modality, but challenges persist regarding post-operative complications and long-term growth outcomes. Conclusion: Hereditary Spherocytosis presents a significant health burden in Indian children. Improved access to molecular diagnostics and long-term follow-up is essential for optimized care.***

Keywords: Hereditary Spherocytosis, paediatric anemia India, genetic mutations ANK1 SPTB, splenectomy outcome, diagnostic challenges.

Hereditary Spherocytosis (HS) is a common inherited hemolytic anemia characterized by spherical red blood cells leading to extravascular hemolysis. While prevalent in Northern Europe, HS is underrecognized in India due to limited awareness and diagnostic facilities. This review consolidates clinical data from Indian studies, highlighting the diverse presentations, diagnostic challenges, and management approaches in the Indian context.

1. Introduction

Hereditary Spherocytosis is primarily an autosomal dominant disorder resulting from defects in red blood cell membrane proteins such as ankyrin, spectrin, band 3, and protein 4.2. In India, Hereditary Spherocytosis is often underdiagnosed, with many cases presenting with nonspecific symptoms or misdiagnosed as other hemolytic anemia.

2. Case History

A 7 year male child born out of a non -consanguineous marriage had complaint of

- Easy fatiguebility since 1 month
- Yellowish discolouration of eyes since last 15 days.
- Fever since last 7 days

3. History and Clinical Findings

- Easy fatiguability since last 1 month.
- yellowish discolouration of eyes since last 15 days which was gradual in onset and increasing.
- -Fever since last 7 days which was gradual in onset, intermittent and relieved by medication.
- No h/o breathlessness, abdominal pain, vomiting.
- No past history of blood transfusion, hospitalization, TB contact.

1) Family history

- No history of need of blood transfusion, chronic respiratory and cardiac disease in family.

2) Birth History-

- Patient was born by a Full Term Normal Vaginal Delivery with the Birth weight of 2.5kg and cried immediately after birth and no history of NICU admission.

3) Immunization history

- Completed upto 5 years of age as per national immunization schedule.

4) Developmental history

- All milestones were achieved as per appropriate for age.

5) General examination

Alert
Temperature - normal
Pulse rate -120/min
RR- 30 /min
CVS - S1, S2 present, No murmur.
CNS - NAD
RS- AEBE, clear
P/A - soft, distended, non- tender splenomegaly present upto umbilicus (Size-16cm).
Pallor- present, Icterus- present.
No cyanosis, clubbing, lymphadenopathy. No oedema over feet or any other signs of congestive cardiac failure.

4. Diagnostic focus, assessment and course of illness.

As patient had fever the symptomatic management was started and following investigations were sent

- Hemogram and peripheral Smear.

- Hb-4.6
- PCV- 15.4%
- WBC- 3300/mm3
- RBC count-235000/cmm
- Platlets - 163000/mm3
- MCV- 74 fl
- MCH- 21 pg
- MCHC-29.6
- RDW- 32.9%
- Retic count- 3.1%
- Corrected retic count- 1.31%.
- Peripheral smear- spherocytes with central pallor was seen.
- Sickling test - Negative
- Iron-37 microgram/dl (Normal- 41-141 microgram/dl
- TIBC- 224 microgram/dl (Normal- 251-406 microgram/dl)
- UIBC-187 microgram/dl (120-470 microgram/dl).
- Ferritin- 28.98 microgram/litre
- Direct coomb's test- negative
- Indirect coomb's test- negative
- Osmotic fragility test- Hemolysis begin at 0.6% NaCL and gets completed at 0.1% NaCL.
- Serum LDH -472
- ESR-67 mm
- HbsAg- negative
- HCV antibody test- negative
- HEV antibody test- negative
- EMA binding study was done:

Mean fluorscent intensity- 19926
Mean fluoroscent index- 0.7
% MFI reduced- 29.8%

Suggesting decreased binding of fluorescent dye eosin -5-Maleimide (EMA) dye study to integral red cell membrane proteins.

- Hb Electrophoresis- Normal.
- G-6PD enzyme activity – Normal.

5. Radiological Investigations

- Ultrasonography (Abdomen and pelvis: Hepatomegaly >14 cm and splenomegaly of >16cm.
- Portal vein doppler- liver: hepatomegaly 13 cm and shows normal shape and texture.
- Spleen- gross splenomegaly >16cm, normal in shape and echotexture.

- As patient had severe anemia PCV transfusion was done.
- Folic acid supplementation was started.
- Growth monitoring and hemogram were planned monthly and iron studies 3 monthly.

Planned for splenectomy if growth faultering occurs or if patient develops transfusion dependent anaemia.

6. Discussion

Epidemiology

- Hereditary Spherocytosis has an estimated prevalence ranging from 1:2000 to 1:5000. Approximately 75% of the patients have autosomal dominant pattern of inheritance and the remaining comprise of recessive mutation and de novo mutation.

Pathophysiology

- The normal red cell membrane can be divided into two parts. The outermost part is a lipid bilayer in which proteins like palladin, glycophorin are embedded. The other part of RBC membrane is composed of a helical cytoskeleton of alpha and beta spectrin which is like a meshwork. Ankyrin attaches the lipid bilayers to the helix of alpha and beta spectrin.
- In hereditary spherocytosis, Ankyrin deficiency is most common cause of HS. It could be inherited as an autosomal dominant or a autosomal recessive manner. Beta spectrin is also a common cause of autosomal recessive mutation. Alpha spectrin causes autosomal recessive mutation. The loss of structural proteins leads to weak vertical intercations and / or decreased lipid anchoring. On continual loss of membrane, the RBC loses its biconvave shape and becomes a sphere. These rigid spherocytes then undergo further membrane loss and ultimately hemolysis occur in spleen resulting in shortened RBC life span.

7. Clinical Presentation

a) Clinical manifestations are variable and may include:
b) Hemolytic anemia
c) Jaundice
d) Splenomegaly
e) Gallstones (pigment stones)
f) Children are often affected by the crisis which are of the following types:
   - Aplastic Crisis- Generally caused by Parvovirus B19, and can lead to sudden and severe anaemia and reticulocytopenia and thrombocytopenia. A bone marrow shows presence of giant normoblast.
   - Megaloblastic Crisis- Have higher requirement of folate.
   - Hemolytic Crisis- During the episode of any viral syndrome, in a child less than 6 years with Hemolytic anaemia, a sudden increase in spleenomegaly along with jaundice anaemia and reticulocytosis is observed.

8. Diagnosis

1) Laboratory Findings
- Complete blood count (CBC): Normocytic or slightly macrocytic anemia
- Peripheral blood smear: Spherocytes (small, round RBCs lacking central pallor)
- Reticulocytosis
- Elevated mean corpuscular hemoglobin concentration (MCHC)
- Elevated lactate dehydrogenase (LDH) and indirect bilirubin
- Decreased haptoglobin

2) Confirmatory Tests
- Osmotic fragility test (traditional, now largely replaced)
- Eosin-5-maleimide (EMA) binding test via flow cytometry – highly sensitive and specific

- Genetic testing – increasingly used for definitive diagnosis and family screening

3) Differential Diagnosis
- Autoimmune hemolytic anemia (AIHA)
- Congenital dyserythropoietic anemia
- Glucose-6-phosphate dehydrogenase (G6PD) deficiency
- Pyruvate kinase deficiency

-The presence of a negative direct antiglobulin test (DAT) helps distinguish HS from AIHA.

4) Management

a) Supportive Care
- Folic acid supplementation
- Monitoring for hemolysis and anemia

b) Splenectomy
- Indicated in moderate to severe cases. It reduces hemolysis and anemia but increases the risk of sepsis, particularly from encapsulated organisms. Postsplenectomy care includes:
- Preoperative vaccination (pneumococcus, meningococcus, Haemophilus influenzae type b)
- Long-term antibiotic prophylaxis

c) Cholecystectomy
- May be indicated for symptomatic gallstones, often performed concurrently with splenectomy.

9. Prognosis and Complications

- With appropriate management, individuals with HS have a good prognosis. Complications may include:
- Iron overload (especially with frequent transfusions)
- Aplastic crisis (commonly from parvovirus B19)
- Splenic sequestration (rare but potentially life-threatening in children).

10. Conclusion

- Hereditary spherocytosis is a common and well-characterized hemolytic anemia with distinct clinical and laboratory features. Advances in diagnostics and genetics have improved patient outcomes. Diagnosis is often delayed allowing progressive fall in hemoglobin leading to compromised quality of life and also leading to big splenomegaly which may pose many other complications.
- Therefore, there is need for increased awareness for timely diagnosis and planning long term management.

References

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