A rare co-existence of two autosomal recessive conditions: Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (CYP21A2 mutation) with Beta thalassemia major

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INTRODUCTION

- 21-hydroxylase deficiency is the most common cause of congenital adrenal hyperplasia (CAH). It is an autosomal recessive condition due to CYP21A2 mutation leading to 21-hydroxylase enzyme deficiency in adrenal gland which leads to decrease synthesis of cortisol and aldosterone and increased synthesis of androgens as shown in figure 1.

- Beta thalassemia is an autosomal recessive condition caused by defective beta-globin chain synthesis and accumulation of unbound alpha globin chains leading to ineffective erythropoiesis.

- We are reporting a case with rare co-existence of CAH due to 21-hydroxylase deficiency with beta thalassemia major.

CASE SUMMARY

Initial Presentation

- A 20-day old neonate presented with complaints of failure to thrive and repeated vomiting. He was born to consanguineous parents with two more siblings (one sibling known to have thalassemia major). There was no history of infant’s death, ambiguous genitalia and CAH in family.

- On examination baby was severely dehydrated with normal male like genitalia (bilateral testis palpable in scrotum) as shown in figure 2.

Figure-2 Showing abnormal male like genitalia

Initial workup & Diagnosis

- Initial investigations revealed severe hyponatremia, hyperkalemia, metabolic acidosis with raised 17-OH progesterone (table-1), all suggestive toward salt looser CAH.

Table-1 showing initial investigations

<table>
<thead>
<tr>
<th>Investigations</th>
<th>Results</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sodium</td>
<td>128</td>
</tr>
<tr>
<td>Potassium</td>
<td>8.8 mmol/L</td>
</tr>
<tr>
<td>17-OH Progesterone</td>
<td>&gt;320 ng/L</td>
</tr>
</tbody>
</table>

- Hb electrophoresis done suggestive of beta thalassemia major as shown in table-2

Table-2 Showing Hb Electrophoresis

<table>
<thead>
<tr>
<th>Hb Electrophoresis</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>HbA1</td>
<td>14.6%</td>
</tr>
<tr>
<td>HbA2</td>
<td>4.9%</td>
</tr>
<tr>
<td>HbF</td>
<td>80.5%</td>
</tr>
</tbody>
</table>

- He received a transfusion and was started on folic acid supplementation with proper follow up for need of repeated transfusions and chelation

DISCUSSION

- This is very rare coexistence of two different autosomal recessive conditions.

- Literature review just reveal only one such case reported in India in 2012 by Yewale etal.

CONCLUSION

- We are reporting a very unusual co-existence of two different autosomal recessive conditions: 21-hydroxylase deficiency and Beta thalassemia Major.

- It might be due to increased penetrance of Thalassemia major and CAH (21-hydroxylase deficiency) in our population with increasing trend of consanguineous marriage.

REFERENCES


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