Congenital Hypopituitarism Presenting Like Sepsis: A Diagnostic Challenge

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Abstract

Congenital hypopituitarism is characterized by multiple pituitary hormone deficiencies which can be potentially fatal in the newborn period but treatable if diagnosis is made early. We report a neonate who presented with sepsis-like symptoms, hypothermia, jaundice, and repeated episodes of severe hypoglycemia. The patient was managed initially on the lines of sepsis however, later on investigations including MRI revealed hypopituitarism, which was corrected promptly.

INTRODUCTION

Hypopituitarism is defined as deficiency in pituitary hormone production(1). The onset can be at any time in life. Congenital hypopituitarism is a rare condition associated with deficiency of all six major hormones (LH, FSH, GH, thyrotropin, corticotrophin, and prolactin(2). Even rarer is the successful diagnosis and management of this condition in the neonatal period, especially when accompanied by other comorbid conditions. This case highlights the clinical presentation and management of such a neonate.

CASE REPORT

An eleven days old male neonate presented in pediatric emergency department of Government Medical College, Amritsar with history of refusal to feed, lethargy, failure to gain weight and jaundice for last 3 days. He was born at 39+1 weeks of gestation by normal vaginal delivery with birth weight of 2.5 kg. The neonate was born with a normal Apgar score and was transferred to the postnatal ward with his mother. His mother was 19 years old and the marriage was non consanguineous. The maternal medical, family and antenatal history were unremarkable and so were antenatal scans and maternal serology for intrauterine infections.

On examination, the patient was in shock, hypothermic, hypotonic and hyporeflexic. Sepsis work up was done and patient was put on antibiotics and ionotropic support. Hypoglycaemia was corrected with dextrose infusion. As sepsis markers came out to be normal and blood culture also showed no growth, antibiotics were stopped. His clinical condition started improving and breastfeeding was established but even on full feeds, the patient was having repeated episodes of hypoglycaemia. He also had persistant jaundice with indirect hyperbilirubinemia (8.16 mg/dl) with total serum bilirubin of 10.04 mg/dl. T4 – 2.4 mcg/dl (9.8-16.6) as well as TSH - 0.2 mIU/L (1.7-9.1) were low. Low level of T4 with low TSH suggested us the diagnosis of congenital hypopituitarism. Other anterior pituitary hormones like ACTH - 5 mcg/l (>10), Cortisol at 8 am - 45 nmol/l (55-304), GH - 0.82 ng/ml (1-20), FSH - 0.04 IU/L (0.3-4.2), LH - 0.07 IU/L (0.1-13.4), Prolactin - 3 ng/ml (7-40) also came out below, however ADH was normal. MRI of brain showed pituitary hypoplasia (0.6cm, 0.37cm and 0.26cm in its transverse, anteroposterior and superoinferior dimensions respectively). Posterior pituitary lobe was in normal position and showed normal hyperintense signal. Ophthalmic examination was normal. Replacement doses of hydrocortisone and l-thyroxin were started. These improved his clinical status as he became more active with normal tone. Now the child is 2 months old, active and gaining weight adequately.
DISCUSSION

Congenital hypopituitarism may be mistaken for sepsis, which can present with similar features of hypothermia, hypoglycemia, lethargy and poor feeding, as in this neonate (2-5). Affected newborns are usually of normal size and weight at birth. Prolonged jaundice is common and some infants show evidence of neonatal hepatitis syndrome but the relationship is obscure (1). Unilateral or bilateral optic nerve hypoplasia is common (1, 2, 6). Definitive diagnosis rests on demonstration of absent or low levels of pituitary hormones. MRI is indicated in all patients with hypopituitarism (1, 7). The initial investigations and management should be aimed at stabilizing the baby. Presence of any one or all of the above clinical features should be a trigger for the investigation of hypopituitarism.

Congenital hypopituitarism may be the result of complications around delivery, or may be the result of insufficient development (hypoplasia) of the gland, sometimes in the context of specific genetic abnormalities like mutations in HESX1 and SOX2 (8). Pituitary hypoplasia may be isolated or associated with other defects like septo-optic dysplasia and midline brain defects (9). In our case there is isolated pituitary hypoplasia mainly involving anterior lobe of pituitary.

By far anterior pituitary is the most common lobe to be involved, but both anterior and posterior pituitary may be affected. Adrenocorticotropic hormone (ACTH), thyroid stimulating hormone (TSH), follicle stimulating hormone (FSH), luteinizing hormone (LH), growth hormone (GH), and prolactin may be deficient along with partial or complete absence of vasopressin. This may cause diabetes insipidus, although our patient did not have this condition. Less commonly, hypothalamus may also be involved. Provocative testing may be required if the clinical features and biochemical abnormalities are subtle. Children with congenital hypopituitarism need to be followed up regularly for ongoing management.

In our patient, the presentation of repeated episodes of hypoglycemia and hypothermia were very suggestive of congenital hypopituitarism. Though micropenis is also one of the common findings in hypopituitarism but it was not present in our case (1, 2). Early diagnosis meant that treatment was instituted early.

CONCLUSION

Congenital hypopituitarism, a rare entity in newborns, can present like sepsis. Clinical presentation is variable, depending on the type and severity of deficiencies and on the age at diagnosis. Early diagnosis and treatment of this
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disease leads in decreasing morbidity and mortality of affected newborns. High index of suspicion is needed for diagnosis and appropriate management of affected newborns.

References
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