Neonatal Dyshormonogenetic Goitre - A Case Report

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Abstract

Dyshormonogenetic goiter resulting in neck swelling in a fetus is very unusual. We present a case of congenital dyshormonogenetic goitre in a fetus with hypothyroidism and respiratory distress at birth. He underwent planned EXIT procedure.

Key words: Dyshormonogenetic goitre, EXIT therapy, Hypothyroidism
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Introduction

Dyshormonogenetic goiter (DG) is the generic term given to the group of familial goiters owing to an inherited defect in the metabolism of thyroid hormones. There is a defect in thyroid hormone synthesis which results in prolonged thyroid stimulating hormone secretion and compensatory goiter. The prevalence of the disease is 1:50,000 live births [1]. It accounts for 10-15% of all cases of familial goiters in a neonate. A large goiter can compress the airway and cause severe complications. A well planned ex utero intrauterine (EXIT) therapy, early recognition and institution of levothyroxine will prevent severe complications and progression of the disease.

Case report

A 21-year old primigravida had a normal antenatal course until the 9th month of gestation. During the routine ultrasonographic examination at 36 weeks of gestation, it was noticed that the fetus had a soft tissue homogenous mass in the anterior aspect of the neck. The location, consistency and bilobed appearance of the mass were suggestive of fetal goiter. Fetal MRI was done. MRI showed soft tissue mass in the anterior neck. This mass appeared to be compressing the trachea and oesophagus and extending to the sternal notch. The mass had soft tissue component with no calcifications, size 4 X 6 X 5.5 cm. Mother was clinically euthyroid and had no past history of thyroid disease. Maternal thyroid function tests and ultrasound of the neck were normal. All the parameters of the fetus were normal.

Mother presented with active labour at 37 weeks and expecting a difficult airway and complications to the fetus an EXIT procedure was planned [Fig 1]. Elective partial intubation with intact uteroplacental circulation was planned and arrangement for tracheostomy and tumor resection was done. An elective lower segment cesarean section was conducted and delivered a male baby weighing 2.24Kg, with Apgar score of 8 at 5 minutes. There was mass in the anterior part of the neck with severe respiratory distress. So the child was intubated, shifted to the neonatal ICU and was immediately ventilated. Child was ventilated for a period of 72hrs and successfully extubated.

Investigations revealed haemoglobin 19.4gm%, total leukocyte count 9,200 /cmm, DC N72 L17E2 M1 and Platelet 1,52,000/cmm. Peripheral smear showed normocytic, normochromic picture with anisocytosis and mild polychromasia. Neutrophils were shifted to the left with a band cell count of 8%. Sepsis screen was negative and blood culture was sterile. Chest x ray showed cardiomegaly. ECHO study showed situs solitus with small ostium secondum atrial septal defect measuring 3.7mm.

A repeat post natal ultrasound revealed a thyroid mass. FNAC of the mass was done and showed features of dyshormonogenetic goiter. Thyroid perfusion scan was done which showed increased uptake of > 50.2U which is

about 3to 4 times the normal [Fig 2]. Thyroid function test revealed TSHof 20 µIU/ml (normal < 10 µIU/ml) with free thyroxine at 0.2 ng/dl (normal: 0.5-1.1ng/ml). X- Ray of knee showed complete absence of epiphysis at the lower end of femur.

A diagnosis of neonatal dyshormonogenetic goitre with hypothyroidism was made. The neonate was discharged on the 12th day of life with levothyroxine 50µg/day (15 µg/Kg/day). Subsequent follow up at the well baby clinic showed decrease in size of the goitre and normal growth and development.

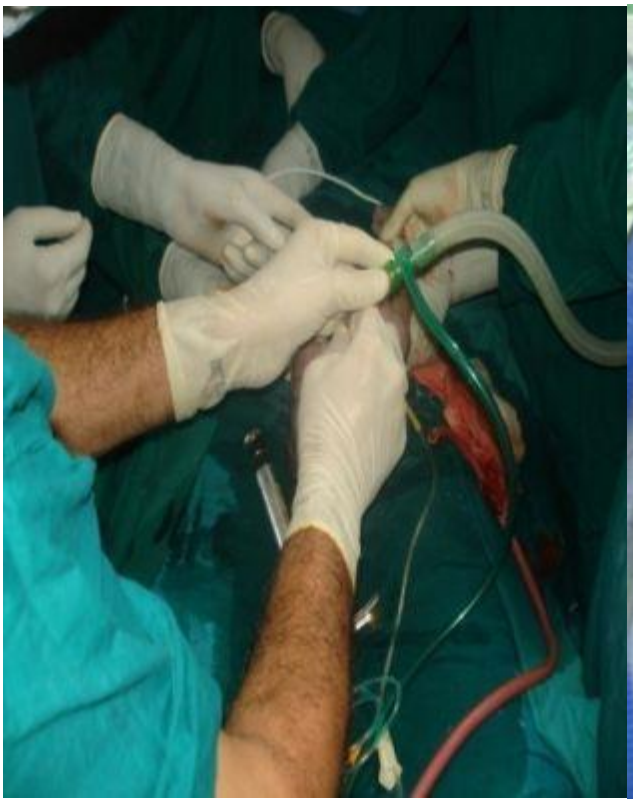

Figure 1-A

Figure 1-B

Figures 1 A and B: EXIT procedure with baby delivered partially and intubated

Figure 2: Increased uptake in thyroid perfusion scans suggesting dyshormonogenesis (image missing)

Discussion

Congenital dyshormonogenetic goitre represents about 10-20% of all cases of congenital hypothyroidism, and most of the neonates exhibit a very large goitre. There have been major advances in screening of congenital hypothyroidism in neonates and in understanding the thyroid system ontogenesis and diagnostic approach to fetal thyroid dysfunction particularly the dyshormonogentic group. The two most common causes for dyshormonogenesis are the defective organification of iodine, frequently the result of mutations in the TPO gene, and the defective synthesis and secretion of thyroglobulin synthesis. As a result of impaired thyroid hormone synthesis thyroid stimulating hormone secretion is increased and results in compensatory goitre [2]. Prenatal diagnosis and treatment with levothyroxine in such cases is advocated as fetus with such large goitre can compress the trachea and obstruct respiration and also cause mechanical problems during delivery. Abuhamad et al. advocated the intramniotic injections of levothyroxine and showed normalization of fetal thyroid function and decrease in size of the fetal goitre by cordocentesis at 35 weeks of gestation [3].Davidson et al during their study showed the total dose of levothyroxine required to reduce the goitre was in the range between 250µg-1000µg [4]. Noia et al showed that even a single intramniotic injection of levothyroxine resulted in a rapid decrease in the fetal goitre size [5]. The results from long

term follow up in children with levothyroxine have shown normal mean IQ values, satisfactory school performance and minimal motor dysfunction. However speech defects and minimal CNS defects have been reported [6].

In cases of large goitre causing tracheal obstruction and mechanical problems during the process of delivery ex utero intrauterine or EXIT procedure is followed. It allows controlled delivery and intrapartum assessment strategy to treat foetuses with life threatening diseases. By maintaining utero placental circulation and partial delivery of the infant, crucial time is provided to perform procedures critical to the survival of the infant like intubation, tracheostomy, tumor decompression and resection [7].

In our case, goitre was suspected when antenatal ultrasound at 36 weeks of gestation showed soft tissue mass in the neck. A difficult airway was anticipated and after discussion with team of doctors comprising of gynaecologist, pediatric surgeon, oto-rhinolaryngolist and anaesthetist an EXITprocedure was planned. Neonate with soft tissue neck mass with respiratory distress was intubated, shifted to intensive care unit and ventilated. There was improvement after 72hrs and was successfully extubated. FNAC of the neck mass, ultrasound, thyroid perfusion scan and thyroid profile confirmed it to be a case of dyshormonogenetic goitre with hypothyroidism. Neonate was discharged on the 12th day with levothyroxine. There was a gradual reduction in the neck swelling on subsequent follow up and the growth and development of the child was normal. Early recognition and intervention in case of congenital goitrous hypothyroidism can prevent complications and progression of the disease.

References

1. Fisher DA. Fetal thyroid function diagnosis and management of fetal thyroid disorders. Clin Obstet Gynecol 1997; 40(1):16-31.
2. Medeiros-Neto GA, Stanbury JB. Inherited disorders of the thyroid system. Boca Raton, FL: CRC press, 1994: 1-218.
3. Abuhamad AZ, Fisher DA, Worsof SL, Slotnick RN, Pyle PG, Evans AT, et al. Antenatal diagnosis and treatment of fetal goitrous hypothyroidism: case report and review of the literature. Ultrasound Obstet Gynecol 1995; 6(5): 368-371.
4. Davidson KM, Richards DS, Schatz DA, Fisher DA. Successful in utero treatment of fetal goiter and hypothyroidism. N Engl J Med 1991; 234: 543-546.
5. Noia G, De santis M, Tocci A, Maussier ML, Caruso A, Mancuso S et al. Early prenatal diagnosis and therapy of fetal hypothyroid goiter. Fetal Diagn Ther 1992; 7: 138-143.
6. Illig R, Largo RH, Qin Q, Torresani T, Rochiccioli P, Larson A. Mental development in congenital hypothyroidism after neonatal screening. Arch Dis Child 1987; 62 (10): 1050-1055.
7. Mychaliska GB, Bealer JF, Graf JL, Rosen MA, Adzick NS , Harrison MR. Operating on placental support: The EX Utero Intrapartum treatment (EXIT) procedure. J Pediatr Surg 1997; 32(2): 227-31.

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