

CASE REPORTS

CONGENITAL HYPOTHYROIDISM WITH BILATERAL ANOTIA IN A NEONATE OF DIABETIC MOTHER

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Abstract

We report a case of bilateral anotia in a newborn of diabetic mother with congenital hypothyroidism. Infants of diabetic mother have increased incidence of external ear anomaly due to defective embryogenesis. Bilateral anotia may be associated with congenital hypothyroidism as both thyroid gland and external ear develop from first and second branchial arches.

Introduction

The term microtia means abnormality of the size, shape and location of the pinna and ear canal. (1) Anotia indicates complete absence of the pinna and ear canal. External ear abnormality is usually associated with serious renal malformation, mandibulofacial dystocia, hemifacial microsomia and other craniofacial malformations. (2,3) Infants of diabetic mothers are at increased risk of developing external ear malformation than general population. (4) Thyroid gland and external ear both develop from first and second branchial arches. (5) For this reason one can suspect congenital thyroid gland abnormality in patient with bilateral or unilateral microtia or anotia. Here we are reporting a case with bilateral anotia with congenital hypothyroidism born to a diabetic mother.

Case Report

A second born male child, delivered at 36 weeks of completed gestational age by normal vaginal delivery presented with bilateral absence of pinna. Ear cartilages and external auditory meati were absent on both sides and no skin tag was present (Figure 1 and 2). Mother was a known case of Type 1 Insulin Dependent Diabetes Mellitus (DM) and was on daily subcutaneous insulin. The mother's pre-pregnancy Body Mass Index (BMI) was 23.41 kg/m² and her glycosylated hemoglobin (HbA1c) level was 6.2% just before delivery. There was no history of antenatal or puerperal fever, exposure to drugs, radiation during the antenatal period. There was no history of thyroid dysfunction in the family. On examination, birth weight of the baby was 2.35 kg (between 25th -50th percentile), length was 49 cm (75th percentile) and head circumference was 36 cm. Anterior fontanelle measured 1.5cm x 1cm. Posterior fontanelle admitted only tip of little finger. There were no other dysmorphisms and other examinations were normal. Though the baby passed meconium within the first 48 hours, he was constipated with bowel movements once in 4 days. The child developed icterus on day 4 of life and the icterus was prolonged till 17 days of life. However, there was no evidence of ABO incompatibility, of G6PD deficiency and the baby did not require phototherapy. The child had a weak but hoarse cry and there was generalized hypotonia. The deep tendon reflexes showed delayed relaxation. CT scan of the brain with cuts of internal ear was done and it showed normal petrous temporal bone, ossicles and internal ear structures. Brainstem evoked response audiometry (BERA) showed mild bilateral conductive

deafness. Based on clinical suspicion, thyroid function tests were done on day 9 of life which showed T3 : 8 pg/dl (normal : 200-600 pg/dl), Free T4 0.42 ng/dl (normal : 2.0-4.9 ng/dl), TSH 251.12 mIU/ml (normal : 1.7-9.1 mIU/ml), confirming a diagnosis of hypothyroidism. Ultrasound (USG) abdomen was normal and renal function tests were normal. USG neck did not show thyroid gland. TORCH screening was negative.

Chromosomal analysis revealed 46XY. Child was started on levo-thyroxine supplementation at 5 mcg/kg/day; the dose was increased to 10 mcg/kg/day after 2 weeks. Currently the child is being reviewed for reconstructive ear surgery after parental counseling.



Figure 1: Absent pinna and external auditory canal on left side



Figure 2: Absent pinna and external auditory canal on right side

Discussion

Microtia and anotia can be grouped into four types according to the level of affliction. (6) Type I: The external ear is small and the auricle retains most of its normal structure. The external auditory

meatus is usually present. Type II: The external ear is moderately anomalous. The auricle can be hook-, S-, or question mark shaped in appearance. Type III: The external is a rudimentary soft tissue structure with no cartilage; the auricle does not have a normal appearance. Type IV: Anotia; all external ear structures are absent. Our patient falls into the fourth category. It can occur unilaterally (79-93% of cases) or bilaterally; in unilateral cases the right ear is more often affected (7). There are few published studies on microtia-anotia frequency. The birth prevalence estimates vary greatly among countries ranging from 0.8 to 17.4 per 10,000. (2) Anotia accounts for 13-22% of all cases of microtia and anotia combined. (8) It can be an isolated finding or it may be associated with Goldenhar syndrome (most severe manifestation of oculo-auriculo-vertebral spectrum), Treacher Collins syndrome, congenital rubella syndrome (5) trisomy 21 (9), retinoic acid embryopathy (10), oto-mandibulo-facial dysostosis, Meir Gorlin syndrome, thalidomide embryopathy (2), fetal alcohol syndrome, diabetic embryopathy (11), holoprosencephaly, Fryns syndrome (12) etc. Inheritance can be multifactorial or autosomal dominant. Preauricular pits and sinuses, and a combination of pits, preauricular appendages (skin tags), cupping deformity, and deafness are all hereditarily dominant (13). Both dominant and recessive characteristics have been revealed in deafness associated with several auricular abnormalities (14). In our case we did not find any associated feature suggesting any syndrome or family history suggesting any particular pattern of inheritance. But there was maternal history of diabetes.

Maternal diabetes is known to have teratogenic effects (4). Infants of diabetic mothers are at increased risk of Oculo-Auriculo-Vertebral Syndrome than in the general population associated with hearing loss, athymia, and cardiac, renal, and limb malformations.

Embryologically the thyroid gland arises from the first and second pharyngeal arches at 5 weeks, and its descent through thyroglossal duct is completed by 7 weeks. The external ear arises from a group of elevations called "auricular hillocks" around the first and second pharyngeal arches at around 7 weeks of age and is completed by 7 months of gestation. Therefore anomalies of the external ear may co-exist with anomalies of the thyroid gland. In the developing countries like India thyroid screening is not routinely done in all infants. But, in cases of bilateral anotia we must look for thyroid gland abnormality even though there may not be any symptom of congenital hypothyroidism. There are few case reports including one of them from India which support our conclusion (15).

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