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Case Reports

Nager Acrofacial Dysostosis Syndrome: A Newborn with Bilateral Hearing Impairment

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Introduction

Nager acrofacial dysostosis (NAFD) comprises defects of craniofacial region and limbs (mostly upper) with variable associated anomalies. This syndrome was recognized as a specific entity by Nager and deReynier[1], Craniofacial features include malar hypoplasia, micrognathia, low set posteriorly rotated ears, preauricular tag, atresia of external ear canal. The limb deformities in the Nager syndrome consists of hypoplasia to aplasia of thumb, with or without radius, proximal radioulnar synostosis and short forearm[2,3]. Conductive deafness usually bilateral and problems with articulation is also noted in this syndrome[2]. Occasional abnormalities may comprise genitourinary abnormality, hypoplastic 1st rib, downward pointing of ribs, lower lid coloboma and cleft lip[4].

Most reported cases have been sporadic. However, Marden et al described an infant with this syndrome having parents with age above 40 years suggesting dominant mutation[5]. Both autosomal dominant and recessive mode of inheritance are also suggested as parent to child transmission as well as more than one affected child have been reported[6].

About 70 cases have been described in the literature[7]. Early diagnosis of the syndrome and detection of hearing impairment is very important as hearing loss may cause delay in speech and language development. Hearing aid augmentation is recommended for this problem. We report this case because of its rarity as well as early detection of hearing impairment in a newborn.

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Case Report

A one hour old male baby of nonconsanguinous parents was admitted into the neonatal care unit with the complaints of prematurity, low birth weight and multiple congenital anomalies. He was born at 34 weeks and his birth weight was 1650 gm. His mother was 30 years old. She suffered from fever for 3-4 days not associated with rash during the 1st trimester and developed pregnancy induced hypertension (PIH) after 32 weeks of gestation. Baby cried immediately after birth and on admission his vital signs and colour were normal. He had facial dysmorphism comprising malar hypoplasia, micrognathia, low set ear with flat pinna of left ear and preauricular pit. Limb defects included bilateral thumb aplasia and short forearm (Fig.-1). The lower limbs were normal. Additional findings were the presence of widely spaced nipple and cryptorchidism.

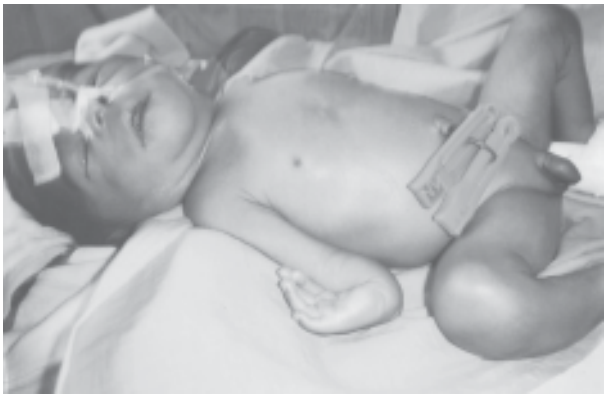

Fig.-1: *The neonate with thumb aplasia*

Complete blood count, serum electrolyte and renal function were normal. Ultrasonography of whole abdomen revealed testes in the inguinal canal and persistent processus vaginalis. Infantogram showed bilateral proximal radioulner synostosis, short left radius and bowing of right sided ulna causing madelung deformity. On both sides only 4 metacarpal bones were noted. Bowing of upper ribs were obvious on both sides of the chest (Fig.-2).

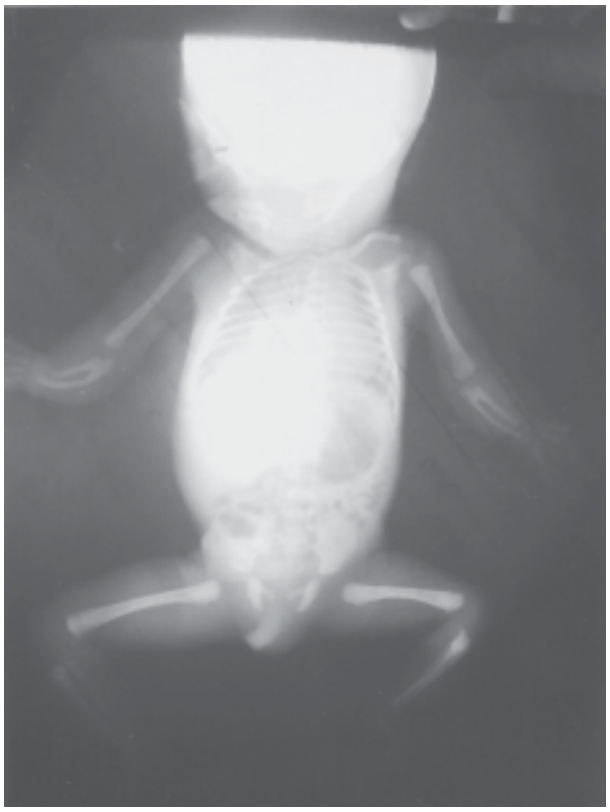

Fig.-2: *X-ray of upper limb shows bilateral radioulner synostosis with bowing of ulna causing madelung deformity*

For hearing test otoacoustic emission (OAE) was done and bilateral hearing impairment was detected and brain stem auditory evoked response (BAER) could not be tested because the baby was not sedated. Karyotype was normal (46XY).

Baby was treated with IV fluid, antibiotics and he left the hospital against medical advice.

Discussion

Nager acrofacial dysostosis (NAFDs) comprises craniofacial abnormalities[8] like malar hypoplasia, micrognathia and variable auricular defect. The most consistent feature are preaxial defects like hypoplastic/ aplastic thumb associated with radioulnar synostosis. Conductive deafness due to atresia of the external auditory canal is another characteristic feature of the syndrome[2].

Our case is characterized by craniofacial features like malar hypoplasia, low set ear with flattening of pinna associated with preauricular pits. Limb defects were thumb aplasia and short forearm with hypoplastic radius. There is cryptorchidism and slender ribs pointing downwards.

Parents look healthy. There is no consanguinity of marriage. Most of the cases of NFADs are sporadic in occurrence[3]. Mother is only 30 years old. So, it could be sporadic in occurrence. All NAFDs must be considered as genetic disorders until proven otherwise and parents deserve careful scrutiny for mild manifestations. NAFD appears to be an heterogenous entity.

Perinatal mortality is about 20%, mostly related to respiratory distress secondary to severe micrognathia. Survivors after infancy have normal intelligence and are presumed to have normal life span[3]. The syndrome can be diagnosed antenatally by ultrasonography[10] but postnatal diagnosis can be established relying upon clinical and radiological feature and with chromosomal analysis in syndromal cases.

Careful evaluation of parents and sibs is required; if one parent is mildly affected recurrence risk is 50%; if parents are apparently normal 25% recurrence risk can be excluded.

Conclusion

Mere diagnosis of Nager acrofacial dysostosis in neonatal period seeing only the craniofacial and limb defects and not performing the test for hearing may cause delayed detection of conductive deafness resulting in delayed speech and language development. Hearing aid for this problem can be proposed to the patient from early life to augment the speech development, if deafness can be detected early in the neonatal period.

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