Acrorenal Mandibular Syndrome

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Introduction

Acrorenal mandibular (ARM) syndrome is rare and only one case in a male foetus is reported in literature [1]. A case of ARM syndrome in a preterm male neonate is discussed.

Case Report

A 22 year old unbooked primigravida with no significant antenatal history delivered an 850gm low birth weight preterm male neonate at 28 weeks of gestation. Despite active resuscitation, he expired after one hour. The neonate had striking anomalies of the face and feet which included low set ears, depressed nasal bridge, marked mandibular hypoplasia, cleft palate and ectrodactyle involving both the feet (Fig.1). The kiddygram revealed poorly inflated lungs, mandibular hypoplasia and split feet. The autopsy revealed bilobed hypoplastic lungs and absent kidneys while the karyotyping was normal (46, XY).

Discussion

ARM syndrome is also known as acrorenal uterine mandibular syndrome or split hand and split foot syndrome with mandibular hypoplasia. The syndrome is common in female children born out of consanguineous marriages even though it is an autosomal recessive disorder [2]. Fitch et al [3], described a similar syndrome

for the first time in a girl child with split-foot defects, bilateral renal hypoplasia, bicornuate uterus and an apparently normal jaw. Others have also reported similar cases [4,5]. Only one case of acrorenal mandibular syndrome in an 18 week old male foetus has been reported [1]. The syndrome has various deformities like split hand/feet, hypoplastic mandible, rib and vertebral anomalies, joint contractures, hypoplastic/dysplastic kidneys, diaphragmatic hernia and uterine defects. The pathogenesis has been linked to an abnormal epithelial-mesenchymal interaction during embryonic development. Parental screening may reveal skeletal, renal or uterine anomalies [5].

The male neonate in our case was a product of nonconsanguineous marriage and had typical features of ARM syndrome. The outcome of such cases with bilateral renal aplasia is uniformly fatal. Screening of parents and close relatives failed to reveal any related abnormality. Genetic counselling was carried out with an emphasis on a recurrence risk of 25% and the importance of prenatal diagnosis.

Conflicts of Interest

None identified

References

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Fig. 1 : ARM syndrome with typical facies and split feet

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