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Case Report (Pages: 9423-9427)

Acalvaria: An Extremely Rare Congenital Skull Malformation

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Abstract

Acalvaria defined as absent skull bones, is an extremely rare congenital anomaly with only a handful of cases reported in literature. In this report we presented a male newborn case of acalvaria referred to our hospital (Shahid Motahari hospital of Urmia, Iran). The condition per se has been described as having high mortality rate. Very few living cases, less than ten, have been reported till now.

Key Words: Acalvaia, Bone defect, Child, Skull malformation.

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1- INTRODUCTION

Acalvaria is an extremely rare congenital malformation characterized by the absence of flat bones of skull, dura mater, and associated muscles in the presence of normal cranial contents and facial bones (1-4). Although its pathogenesis is unclear it has been explained by the theory of post-neurulation defect (1-3). It has been considered a fatal congenital anomaly and only a handful of cases have been reported in English literature to date (1-3). Thus, we herein present the case which referred to our hospital because of lax skull. The pertinent literature is reviewed with the aim to make physicians aware regarding the clinical features of this uncommon fatal clinical anomaly so that undue associated morbidity could be avoided by prompt antenatal diagnosis and subsequent management.

2- CASE REPORT

A two day-old male neonate referred to our hospital (Shahid Motahari hospital of Urmia, Iran) with respiratory distress and lax skull. He was born through normal vaginal delivery at 41 weeks of gestation with 10 Apgar score at fifth minute of birth. His weight was 3,800 grams and head circumference was 35 centimeters. His mother's age was 35 year-old and his father's age was 42 year-old without consanguinity. He was the third baby, having two normal sisters with 11 and 17 years old. The mother had prenatal care. Because of the results of first trimester screening tests that reported the risk of Down's syndrome greater than cut off (1/33), amniocentesis was done. The result was 46XY with no structural chromosome abnormality. Fetal ultra sound examinations revealed no major anomaly at 21 and 31 weeks of gestation. The mother used ferrous sulfate and multivitamin during pregnancy for her anemia. Her history for folic acid consumption was negative before and during pregnancy.

On physical examination he was an active male with normal neonatal reflexes. On palpation the skull was lax and frontal, parietal and occipital bones were absent. There was no associated abnormality of face, but shortness of proximal limbs was noticed in both upper and lower limbs. The roentgenogram proved the absence of bones and epiphyseal dysgenesis without any fracture (**Figures 1 and 2**).

The computerized tomographic scan and reconstructed three-dimensional-image showed the bone defects of skull (**Figure.3**). Laboratory tests were normal except for 25-hydroxy vitamin D3 (25(OH)D3), which was 16 ng/ml for baby and 12 ng/ml for his mother. The baby managed conservatively and started to be fed with breast milk after respiratory recovery and he was discharged from hospital after 6 days, with close follow up and recommendation to parents for very careful handling and protection of any head trauma.

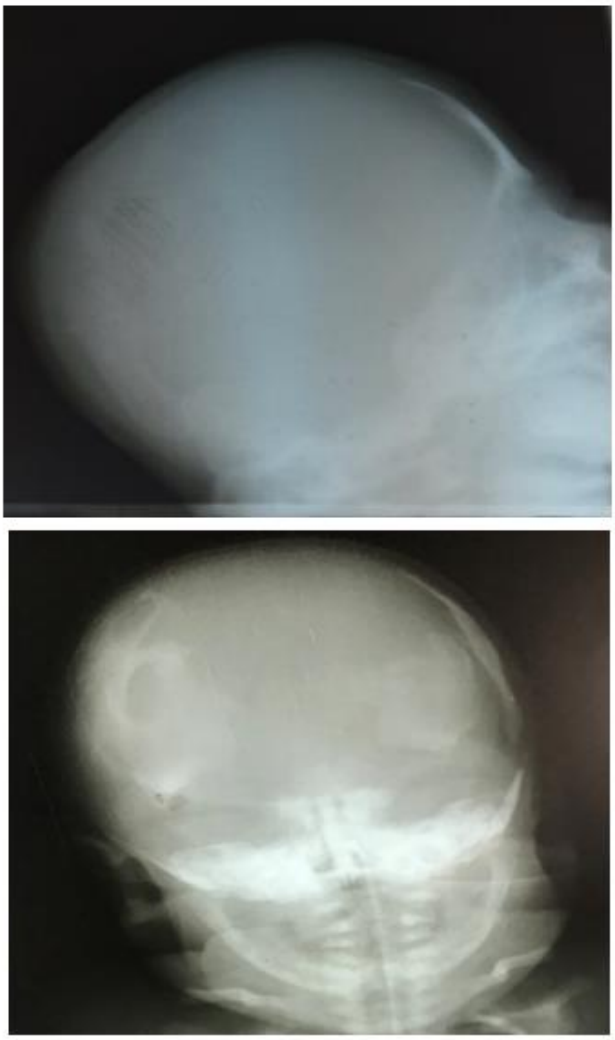

Fig.1: Skull X- ray, Lateral and AP views, showing absence of large parts of skull bones. AP: Anteroposterior.

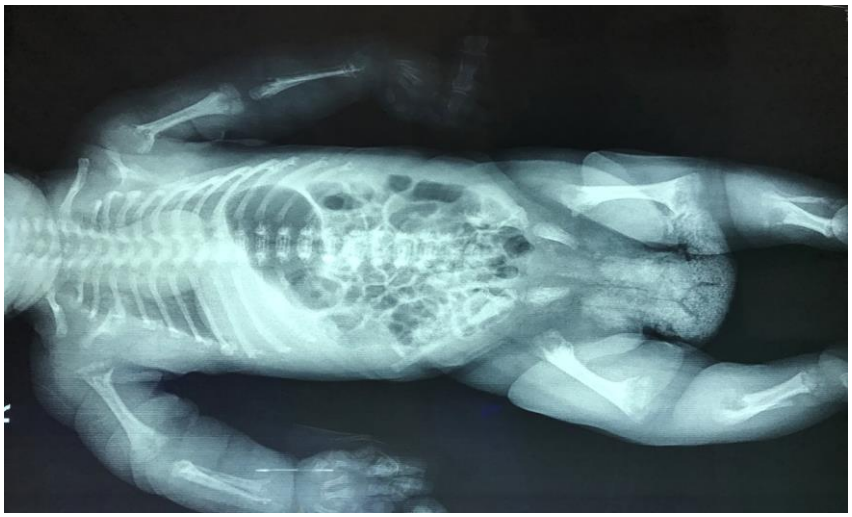

Fig.2: Baby gram, showing epiphyseal dysgenesis and shortness of limbs.

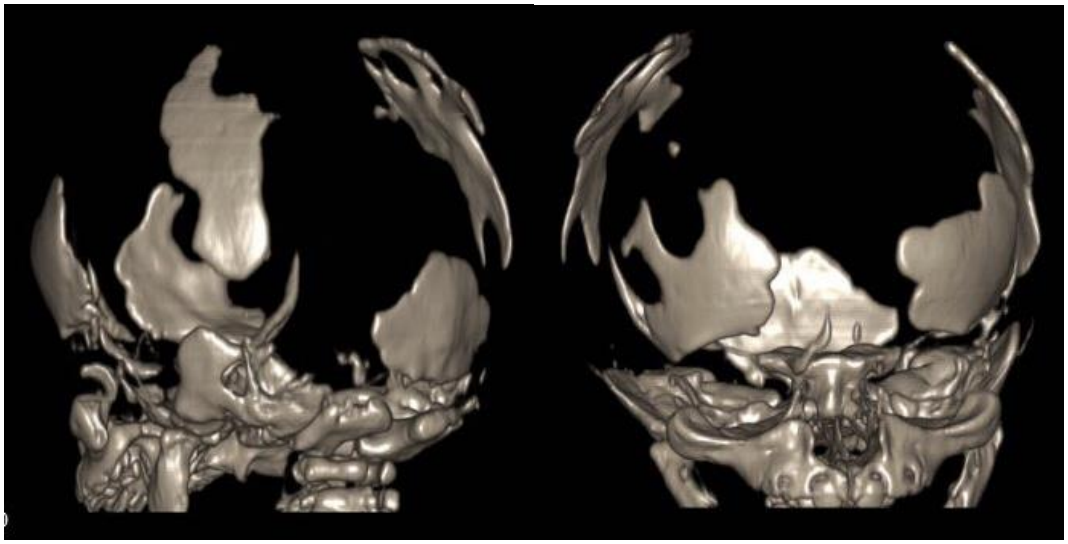

Fig.3: CT- scan of skull, showing absence of large parts of frontal, occipital and parietal bones.

3- DISCUSSION

Acalvaria is an extremely rare congenital malformation with incidence of less than 1 in 1,000,000 and has been sparingly reported in literature with only a handful of case reports (1, 2). Its causation is poorly understood and no concrete etiology has been described. This disorder, showing some female predilection (3), has generally been reported as a fatal congenital malformation with only rare reports describing extended survival (4). Acalvaria is characterized by the absence of flat bones of the skull, dura mater and associated muscles with the presence of normal cranial contents and facial bones (5). Although some cases may show abnormal development of the brain tissue, it is usually normally developed in the majority of cases (5). The etiopathogenesis of acalvaria is still unknown (1-3). A review of literature suggests that different investigators have explained the embryological basis of this rare malformation by theory of post-neurulation defect (2, 4). Normally, around 4 weeks of gestation and after closure of anterior neural pore, there is migration of mesenchymal tissue under ectoderm which forms skin and scalp, but the mesenchymal tissue develops into skull bones and associated muscles (1, 2). Thus, faulty migration of mesenchymal tissue in presence of normal development of ectoderm results in an absence of flat bones of skull and associated muscles (1, 2). Although not widely accepted, some investigators still consider acalvaria to be a part of spectrum of anencephaly, whereas others explain its pathogenesis on the basis of failure of primary closure of neural tube (1, 2). The clinical presentation as experienced in present and reported cases is heralded by the presence of soft, lax skin covering the skull due to the absence of flat bones of cranial vault and associated muscles. The facial bones and cranial contents are usually normal, although some neuropathological abnormality has also been reported (2, 4). Folic acid intake has not been shown to prevent acalvaria, distinguishing this pathology from neural tube defects (1). It occurs rarely and has no specific recurrence risk (2). There have been no chromosomal abnormalities associated with acalvaria, but other congenital anomalies have been found to be associated with it. These include holoprosencephaly, hydrocephalus, and micropolygyria, hypertelorism, and cleft lips or palates (2). Some differential diagnoses for acalvaria include severe osteogenesis imperfecta and congenital

hypophosphatasia. The prognosis is poor in fetuses with acalvaria. The initial treatment of acalvaria is mainly conservative aimed at supportive care and management of any associated anomalies if present (3). There are currently no surgical procedures to correct the bone defect. Therefore, these pregnancies are usually terminated. Management of acalvaria is also not well defined as in its etiology no two cases are the same (6, 7). It has been managed both conservatively and with extensive surgical reconstruction (2, 4, 6, 7). The first ever surviving case was reported 23 years back in Japan. He was treated by surgical closure of the scalp defect. Also, there was associated hydrocephalus so a shunt surgery was also performed. On follow- up he was noted to have severe developmental delay with mental retardation (4). He was last reported alive at the age of 11 years and was attending a special school (4).

4- CONCLUSION

Acalvaria is a very rare fatal anomaly. It occurs after neurulation, distinguishing it from neural tube defects. It consists of an absence of the flat bones of the cranium, the dura mater, and scalp muscles. Since the only living case of this fatal anomaly with long-term follow-up is severely mentally retarded, so we conclude that antenatal diagnosis of this fatal anomaly by a vigilant sonographic evaluation is extremely important so that the morbidity, mortality, and the associated psychological trauma resulting at the time of birth and on subsequent management could be prevented.

5- CONFLICT OF INTEREST: None.

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