CASE REPORT

POLYDACTYLY IN PFEIFFER SYNDROME II-OMIM #101600 A RARE ASSOCIATION
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ABSTRACT: Pfeiffer syndrome (PS) is a rare autosomal dominantly inherited disorder occurring in approximately 1:100,000 live births. Mutations of the fibroblast growth factor receptor 1(FGFR1) or FGFR2 gene can cause Pfeiffer syndrome. Craniosynostosis, brachycephaly, mid-facial hypoplasia, broad deviated thumbs and great toes characterize the syndrome. Pfeiffer syndrome depending on severity of the phenotype is of three types. The types 2 and 3 occur as sporadic cases and have poor prognosis. We report a case of Pfeiffer Syndrome type 2 having polydactyly, which to the best of our knowledge is first case of such an association.

KEYWORDS: Pfeiffer syndrome type-2, polydactyly, kleeblattschädel (cloverleaf) skull.

INTRODUCTION: In 1964, Pfeiffer described a rare autosomal dominantly inherited disorder that associates craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly on hands and feet.¹ Hydrocephaly may be found occasionally, along with severe ocular proptosis, ankylosed elbows, abnormal viscera, and slow development. Based on the severity of the phenotype, Pfeiffer syndrome is divided into three clinical subtypes.² Clinical overlap between the three types may occur. Pfeiffer syndrome affects about 1 in 100,000 individuals.³ The disorder can be caused by mutations in the fibroblast growth factor receptor genes FGFR-1 or FGFR-2. Pfeiffer syndrome can be diagnosed prenatally by sonography showing craniosynostosis, hypertelorism with proptosis, and broad thumb, or molecularly if it concerns a recurrence and the causative mutation was found.

CASE REPORT: A male baby was born at the 40+1 weeks of gestation to 25 years old women and her husband being 31 years old. Maternal antenatal scan showed enlarged head with deformed calvarium, hydrocephalus, dilated ventricles with colpocephaly. The parents were not consanguineous; the mother had two other healthy daughters. The baby was delivered by LSCS with a birth weight of 4,200 g, was 50 cm long, and had a head circumference of 34 cm and Apgar score of 8. The neonate had a kleeblattschädel (cloverleaf) skull (Fig-1a), brachycephaly, flat occiput, flat nasal bridge, maxillary hypoplasia, exophthalmos, proptosis, low-set ears, high arched palate. On skeletal examination radially deviated broad thumbs and medially deviated broad big toes, syndactyly in digits of upper and lower limbs, polydactyly in digits of of lower limbs were present (Fig-1b). Other system examination showed divarication of recti, undescended testes and hypospadiasis.
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USG Abdomen showing GB sludge, divarication of recti. Plain radiographs of the skull showed prominent convolutions, probably due to craniosynostosis. Three-dimensional skull CT revealed premature fusion of the bilateral coronal sutures and left lambdoidal suture, midface hypoplasia, and shallow orbits (Fig-2).

DISCUSSION: Pfeiffer syndrome was first described by Pfeiffer in 1964, who found eight cases in three generations of a family.1 It affects about 1 in 100,000 individuals, but is more rare in the Asian population.3 Pfeiffer syndrome involves the cranial bones and thumbs and great toes, which are broad and bend away from the other digits as seen in our case, PS involves craniosynostosis that is most often of the coronal and lambdoid and occasionally sagittal sutures.

Cohen in 1993 classified this syndrome into 3 clinical subtypes and suggested that these subtypes might not be classified as separate entities, even though these classifications have important diagnostic and prognostic implications.2 The classic PS is designated type I is compatible with life and consists of craniosynostosis, midface deficiency, broad great toes, brachydactyly, and variable syndactyly. Type II consists of a cloverleaf skull with Pfeiffer hands and feet, together with ankylosis of the elbows. Type III is similar to type II without the cloverleaf skull. In type III PS, ocular proptosis is severe, and the anterior cranial base is markedly short. Various visceral malformations have been found in association with type III.2 Early demise is characteristic of both types II and III, which to date has been reported to occur only as sporadic cases.4

Mutations in FGFR1, 2, or 3 genes can affect craniofacial and skeletal development. More than 60 mutations in FGFR genes, a majority of which occurs in FGFR2, are associated with craniosynostosis syndrome such as Antley-Bixler syndrome, Apert syndrome, Bearse-Stevenson syndrome, Crouzon syndrome, Muenke syndrome, and Pfeiffer syndrom.5 Pfeiffer syndrome is caused by mutations in the FGFR gene with locus heterogeneity. Mutations in the FGFR1 at chromosome 8p11.2-p12 were only detected in Pfeiffer syndrome type 2. Mutations in the FGFR2 at chromosome 10q25-q26 were reported in all three subtypes.6

The prognosis of Pfeiffer syndrome depends on accompanying anomalies, and multiple surgeries are needed to release the prematurely closed sutures.7 Craniectomy is mandatory for management of increased ICP, and a multidisciplinary craniofacial team is essential for the long term follow-up of affected children.8

CONCLUSION: This report describes a case of Pfeiffer syndrome type II as per the clinical features as well as having polydactyly with other multiple skeletal, ocular, cranial anomalies. The association of polydactyly in this case being reported for the first time.

REFERENCES:
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