APERT SYNDROME WITH LOBAR HOLOPROSENCEPHALY AND AGENESIS OF THE CORPUS CALLOSUM IN A PALESTINIAN NEONATE: A CASE REPORT

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ABSTRACT Background: Newborn with dysmorphic features consistent with an autosomal dominant apert syndrome which is a rarely associated with agenesis of corpus callosum and lobar holoprosencephaly. **Case summary:** Full term female newborn was a product of normal delivery, she had dysmorphic feature ;small eye sockets, proptosis, hypertelorism,down-slanted palpebral fissure, low set ears, depressed nose bridge, narrow high arched palate, trapezoid mouth, bilateral hands had complex syndactyly involving 2nd, 3rd and 4th fingers, both feet had syndactyly between 3rd and 2nd toes and webbed 1st and 2nd toes. Brain CT showed agenesis of corpus callosum and lobar holoprosencephaly and bilateral coronal synostosis. The case was diagnosed as an apert syndrome which is a rarely associated with agenesis of corpus callosum and lobar holoprosencephaly.baby was discharged home with follow up with the multidisciplinary medical team. **Conclusion:** So rare apert syndrome case with the rare association; lobar holoprosencephaly and agenesis of corpus callosum, was diagnosed since birth which resulted in appropriate medical care and multidisciplinary medical team involvement

KEYWORDS Apert syndrome, holoprosencephaly, agenesis of the corpus callosum

Introduction

Apert syndrome is a genetic disease characterised by; premature closure of cranial suture, brachycephaly, down slant palpebral fissure, proptosis, hypertelorism, depressed nose bridge, low set ears, trapezoid shaped moth, narrow high arch palate and symmetrical upper and lower limbs complex syndactyly. Apert syndrome has various CNS anomalies. The disorder is autosomal dominant, but the most cases are due to new mutations in the FGFR2 gene. The female and male are equally affected. The incidence of Apert disorder is 1:50000-1:80000 [1-2]. Our case is so interesting; she had the feature of Apert disorder and holoprosencephaly with agenesis of the corpus callosum [3].

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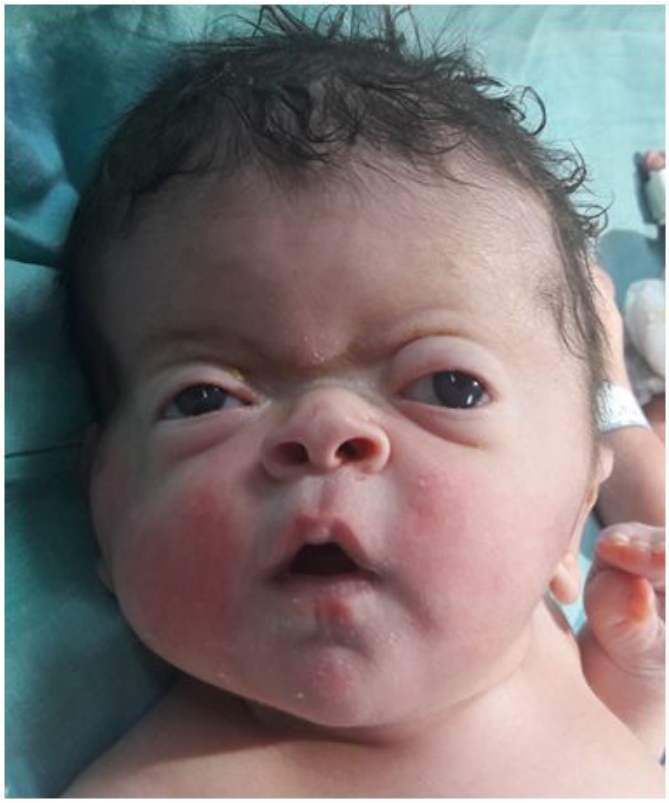
Case report

Parents were nonconsanguineous; they had a negative family history of the genetic disorder, they had three healthy female siblings. The mother had good and regular antenatal care, antenatal ultrasound one week before the labour showed fetal macrocephaly. She was full term, a product of normal delivery; birth weight was 2900 grams, Apgar scores were 7,9 at minute 1,5 respectively. On physical examination she had dysmorphic feature;brachycephaly,prominent forehead ,down slant palpebral fissure,proptosis,hypertelorism,depressed nose bridge ,trapezoid shaped mouth (Picture 1), low set ear (Picture 2), high arch palate,syndactyly involving the 2nd ,3rd and 4th fingers in both

hands[Picture 3,4],syndactyly involving 2nd and 3rd toes and there was web between 1st and 2nd toes in both feet [Picture 5,6]. Head circumference was 36 cm, and anterior fontanel was large 4×5 cm. Abdominal ultrasound and echocardiography were normal. Brain CT showed; lobar holopresencephaly[Picture 7],agenesis of corpus callosum[Picture 8],bilateral coronal craniosynostosis[Picture 9,10] , right sided choanal atresia and left sided choanal stenosis[Picture 11]. She was discharged home at the age of 7 days in good general condition with referral to the; pediatric neurosurgeon, plastic surgeon, dentist, pediatric orthopaedic, pediatric ophthalmologist, Otolaryngologist and audiologist.



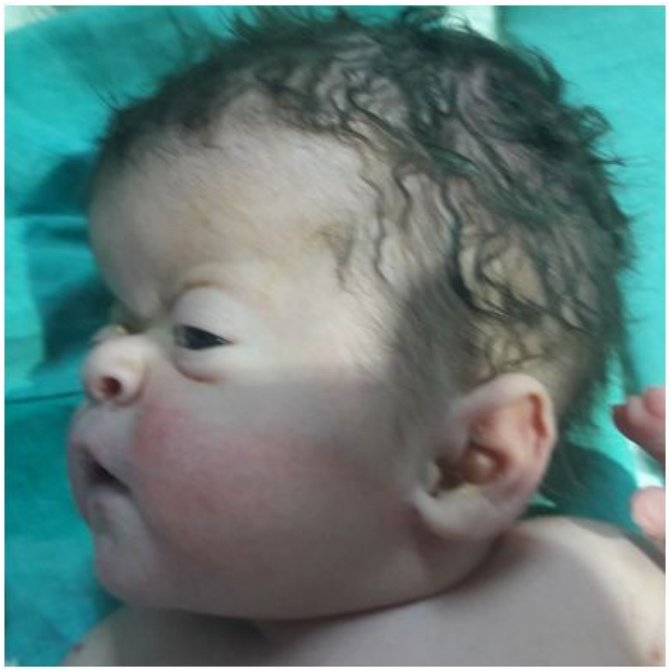
Picture 1: Dysmorphic face.

Discussion

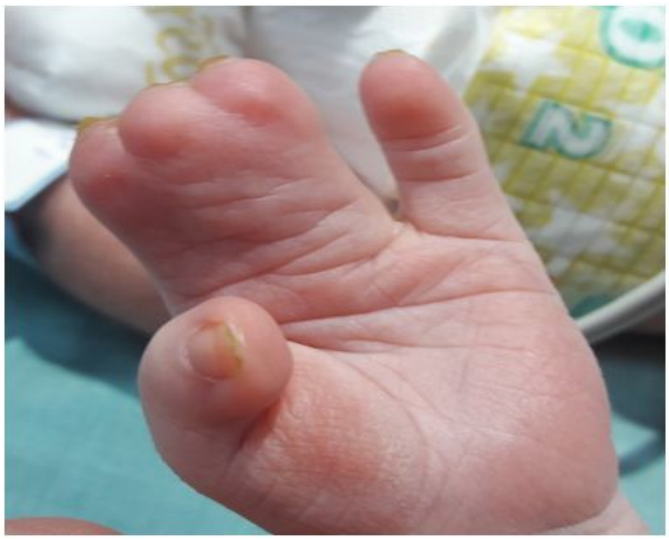
The patient had all clinical and radiologic features consistent with the diagnosis of Apert syndrome[4]. Due to limited resources, the mutation of FGFR2 gene was not done. The case is the Apert syndrome with a rare association with lobar holoprosencephaly, agenesis of corpus callosum and choanal atresia[5]. The case could be diagnosed by antenatal ultrasound examination[6]. The case was diagnosed immediately after birth, and all subspecialties were counseled which will improve the prognosis. Hereditary transmission is autosomal dominant, and the most cases are new mutations, but in the last decade, congenital anomalies incidence increased significantly especially after three war on Gaza strip[7].

Conclusion:

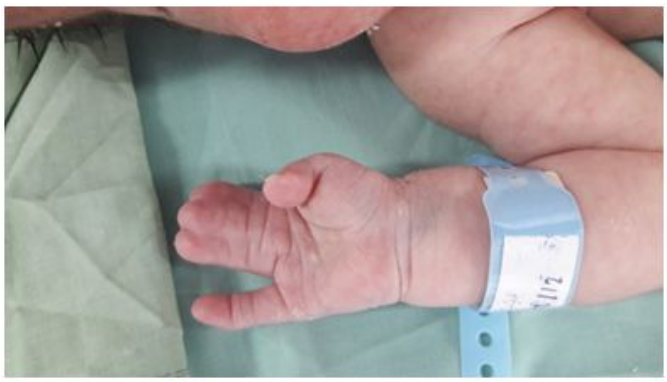
A rare apert syndrome with the rare association; lobar holoprosencephaly and agenesis of corpus callosum, was diagnosed since



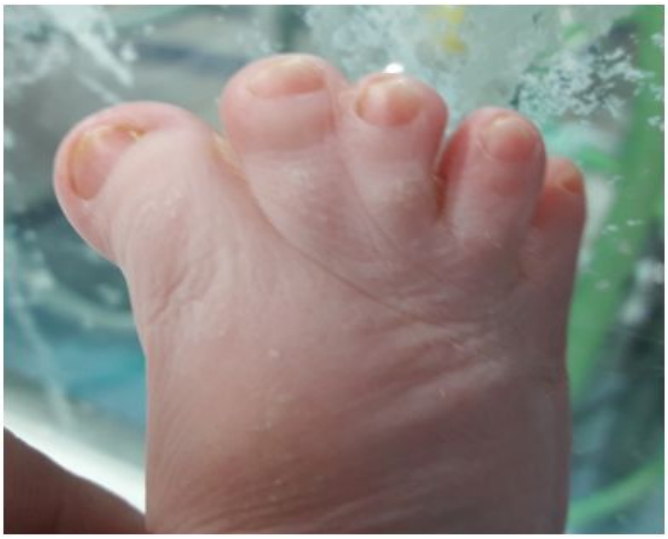
Picture 2: Low set ears.



Picture 3: Syndactyly involving the 2nd, 3rd and 4th fingers (Left hand).

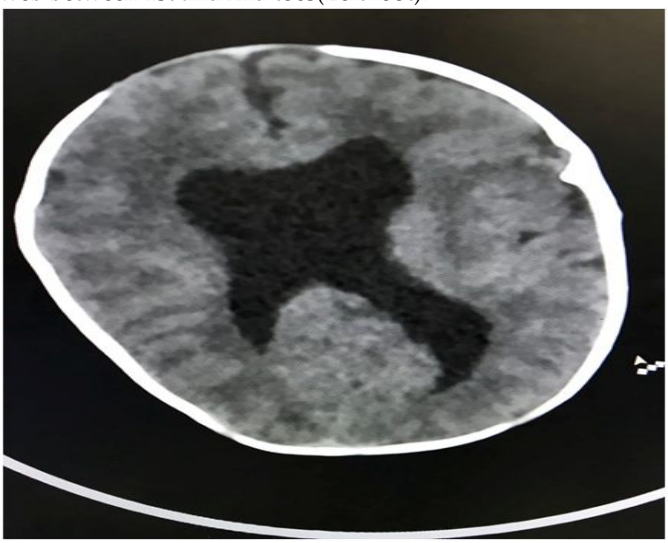


Picture 4: Syndactyly involving the 2nd, 3rd and 4th fingers (Right hand).

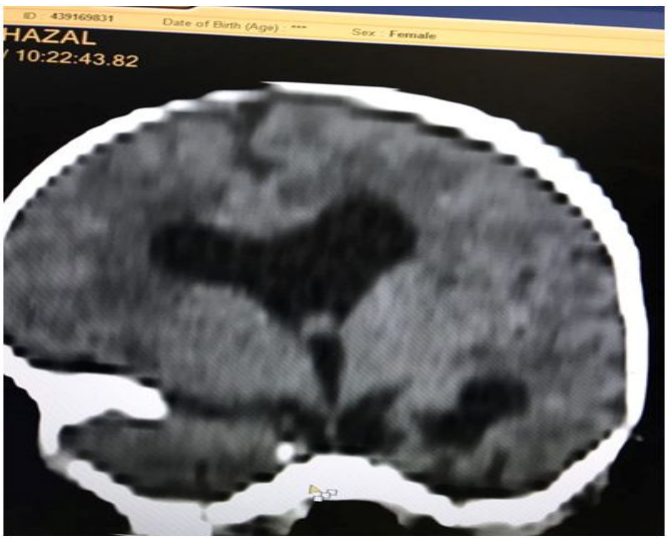


Picture 5: Syndactyly involving 2nd and 3rd toes and there was web between 1st (Right foot).

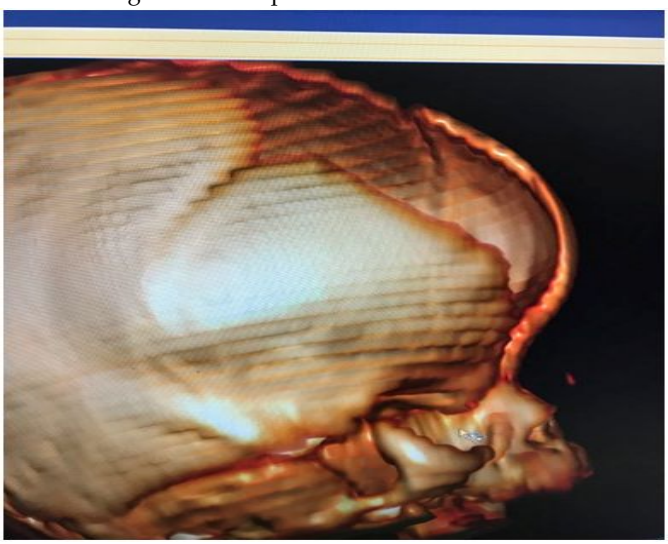
Picture 6: Syndactyly involving 2nd and 3rd toes and there was web between 1st and 2nd toes(Left foot).



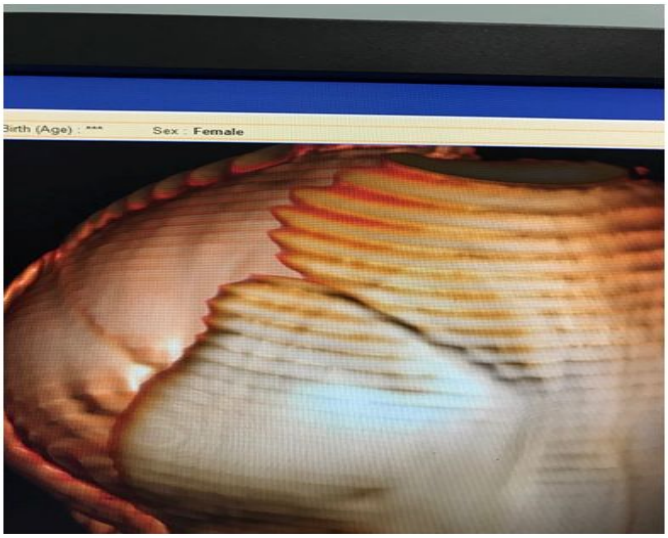
Picture 7: Lobar holopresencephaly.



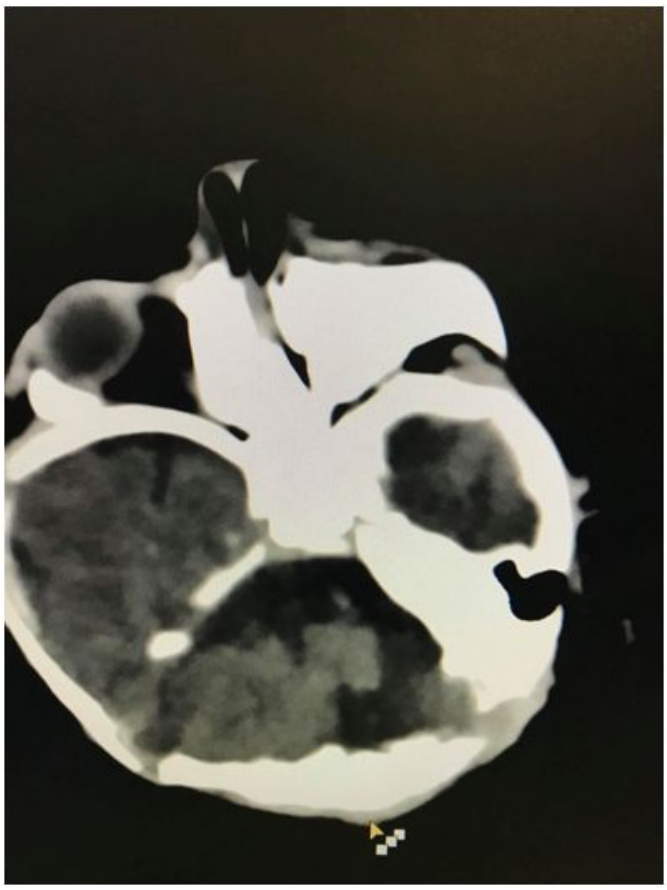
Picture 8: Agenesis of corpus callosum.



Picture 9: Right-sided coronal craniosynostosis.



Picture 10: Left-sided coronal craniosynostosis.



Picture 11: Right-sided choanal atresia and left-sided choanal stenosis.

birth which was resulted in appropriate subspecialty counselling and multidisciplinary medical team involvement.

Take home messages

The 1-Agenetic laboratory is needed as soon as possible in Gaza strip which will help in diagnosing so many congenital disorders in turn; it will help in genetic counselling and decrease the incidence of the hereditary disorder.

2-More studies are needed to find out the association between the incidence of congenital anomalies and the environmental factors.

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Authors' Statements

Competing Interests

The authors declare no conflict of interest.

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