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Case Report

Unusual Constellation of Anomalies in Maternal Diabetes Syndrome: Bifurcated Distal Phalanx of the Thumb, Cleft Lip, and Patent Ductus Ateriosus

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Abstract

Maternal hyperglycemia during early gestation is associated with an increased incidence of congenital anomalies. A case of maternal diabetes syndrome is presented here with a rare constellation of congenital anomalies, i.e., bifurcated distal phalanx of the thumb, patent ductus arteriosus and cleft lip.

Keywords: *Maternal diabetes, Congenital anomalies, Bifurcated thumb*

Introduction

Gestational diabetes and pre-existing maternal diabetes can cause structural defects when a developing baby is exposed to elevated levels of glucose (1, 2). The incidence of major congenital anomalies in newborns of diabetes mothers is higher than that in other newborns (3). The most common types of anomalies seen in newborns of diabetic mothers include anomalies of central nervous system, cardiovascular, skeletal, gastrointestinal, and genitourinary system, where the cardiac malformations stand in the first rank (4-6).

Maternal diabetes can lead to approximately three to five-fold increases in the risk of cardiovascular malformations (5, 7). Various types of cardiovascular anomalies have been reported in infants of diabetic mothers with a predisposition to transposition of great arteries, hypoplastic left heart, tetralogy of Fallot, and pulmonary/ tricuspid atresia (8-10). A population based study in England, Wales, and Northern Ireland revealed that in babies of diabetic mothers the most common anomalies were seen in cardiovascular system followed by musculoskeletal, nervous, urogenital, and digestive system (6). According to Macintosh et al. in this population based study, the most common cardiovascular anomalies were hypoplastic left heart, tetralogy of Fallot, transposition of great arteries, pulmonary artery stenosis, coarctation of aorta, and double outlet right ventricle. In the musculoskeletal system, caudal regression was the most common anomaly followed by diaphragmatic hernia, gastroschisis, prune belly syndrome, and scoliosis. Among 2400 infants of diabetic mothers two had cleft palate; whereas, cleft lip not seen in any of them (6).

Case

The parents are non-consanguineous and both in their early 40s without any dysmorphic feature. Mother is the known case of diabetes mellitus diagnosed approximately two year before conception. Her diabetic condition was poorly controlled before as well as during pregnancy.

The patient is 7 yr old girl and born at 40^{th} week of gestation to a diabetic mother after an unremarkable pregnancy. Her delivery was uncomplicated with normal APGAR scores. Her birth weight was 5 kg. In the physical examination she was developmentally normal with no

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neurological finding. Her right thumb was duplicated in the distal phalanx, which is compatible with Wassel type II duplication (11). The radial component of the duplicated thumb demonstrated limited range of motion (ROM) at inter-phalangial (IP) joint (symphalangism). X-ray revealed an undeveloped IP joint of radial component of the duplicated thumb (Fig. 1). Her PDA (Fig. 2) and unilateral left upper cleft lip surgically corrected (Fig. 3). The radial component of the duplicated thumb resected and the head of the proximal phalanx shaved down to the size of the base of the distal phalanx. The radial collateral ligament of the IP joint reconstructed. The final ROM of the IP joint was 0-25° in flexion.

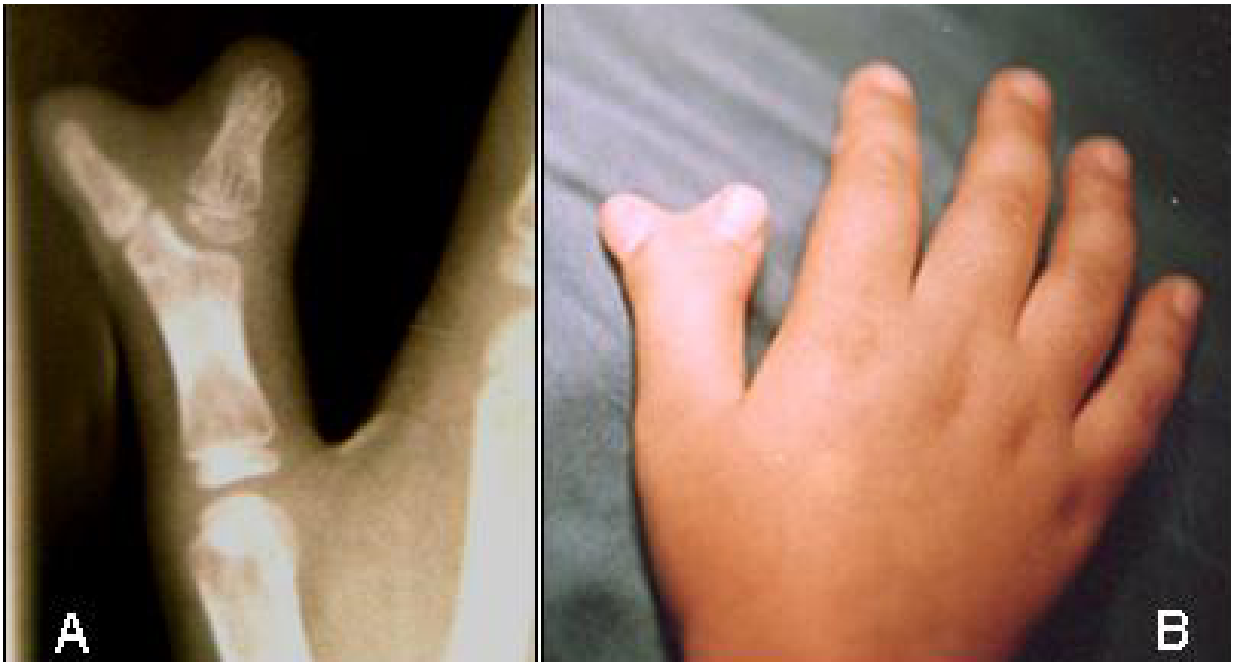

Fig. 1: A) X-Ray of the right thumb and B) photography of the right hand show bifurcation of the distal phalanx of the thumb

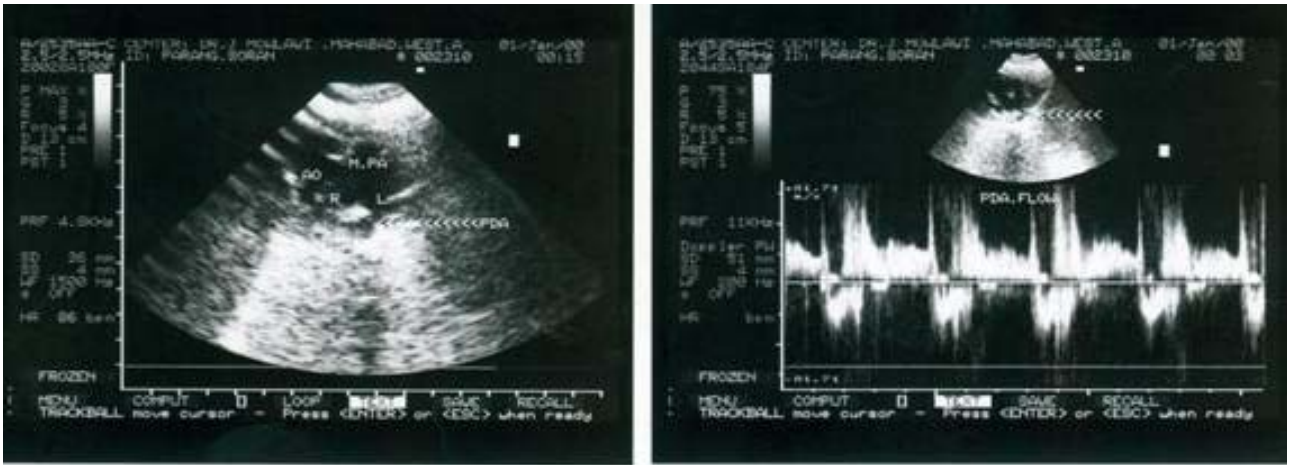

Fig. 2: Sonographic detection of PDA before surgical correction of the defect

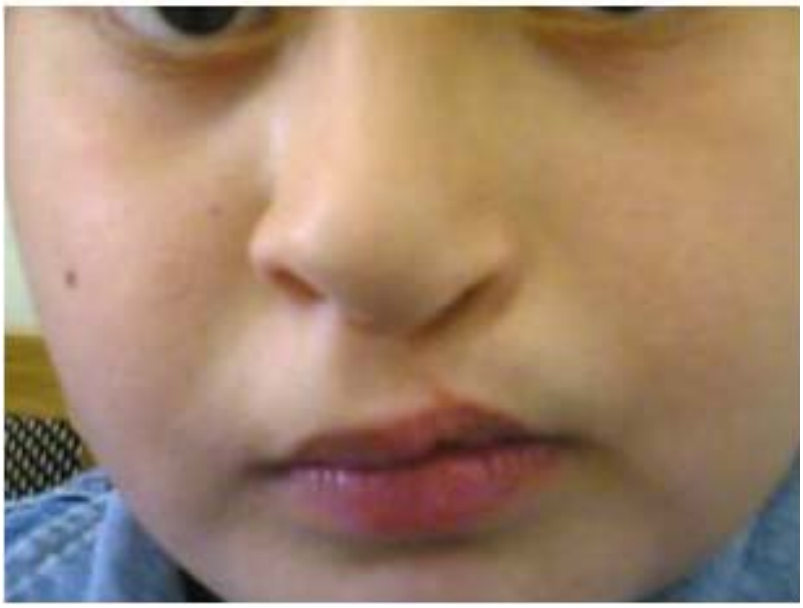

Fig. 3: Unilateral left upper cleft lip after surgical correction

Discussion

Infants of diabetic mothers are at increased risk for major congenital malformations and the teratogenic effects of maternal diabetes occur in multiple organs and systems. No single anomaly is pathognomonic, but several are much more frequent including cardiovascular, skeletal, CNS, genitourinary, and gastrointestinal system (3-5). Our case was macrosomic infant of a diabetic mother considering the birth weight of 5 kg and according to Yaseen et al. definition (12). Macrosomia could be due to mitogenic and anabolic effects of insulin. After scrutinized examinations, we could not find any structural anomaly other than duplicated distal phalanx of the thumb, cleft lip, and patent ductus arteriosus. To the best of our knowledge this case is the first reported combination of duplicated distal phalanx of the thumb, cleft lip, and patent ductus arteriosus in a maternal diabetes syndrome. This kind of combination is also seen in Robinow syndrome, however, many other features which were not seen in our case, are associated with this rare syndrome (13). In a population based study by Macintosh et al, there was no case of cleft lip in infants of diabetic mothers; however, isolated cleft palate was seen in two out of 2400 infants of diabetic mothers, which is approximately two times of expected rate in general population (6). Duplicated distal phalanx of the thumb is not specified in infants of diabetic mothers in the literature. An echocardiographic study within 48 h of birth on 100 neonates of diabetic mothers revealed that patent ductus arteriosus was the most common echocardiographic finding followed by patent foramen oval, that is, 70% and 68%, respectively (14).

Since organogenesis can be affected by maternal hyperglycemia, hypoglycemia and ketosis, careful attention to pre-conception control of diabetes and treating gestational diabetes decrease the risk of birth complications and anomalies (15, 16). And also detailed sonographic studies should be carried out on high-risk pregnancies for early detection of congenital anomalies. This case could be unique due to: 1) there are a few reports (17) that have described the symphalangism in a duplicated thumb, 2) constellation of duplicated thumb, cleft lip, and patent ductus arteriosus has not been reported in the maternal diabetes syndrome.

Acknowledgements

The authors declare that they have no conflict of interests.

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