



Polymastia with Diastometamyelia: Case Report

Sengul Yuksel*, Yusuf Kenan Coban**

* Inonu University, School of Medicine, Department of Medical Biology and Genetics, Malatya

** Inonu University, School of Medicine, Department of Plastic and Reconstructive Surgery, Malatya

Although possible associations between supernumerary nipples and other congenital anomalies has been emphasized, diastometamyelia and polymastia which has not been reported as an association of these two anomalies before. In this report, we describe a 11 years-old girl with diastometamyelia and polymastia and we investigated genetic etiology of her complex phenotype.

Key Words: Polymastia; Diastometamyelia; Genetics.

Diastometamiyeli Polimasti

Çoklu meme oluşumu ile diğer konjenital anomaliler arasında olası ilişkiler gösterilmiş olmasına karşın diastometamiyeli ve polimasti anomalilerinin ilişkisi daha önce bildirilmemiştir. Bu vakada, diastometamiyeli ve polimastili on bir yaşındaki bir kız hasta kompleks fenotipinin genetik etiolojisi ile birlikte araştırılarak sunuldu.

Anahtar Kelimeler: Polimasti; Diastometamiyeli; Genetik.

Ectopic breast tissue occurs in 0.4-6% of the general population. Usually these tissues develop along the embryonic milk line. Although polythelia is congenital in origin and is identifiable at childhood, polymastia may not be evident, until the influence of sex hormones during puberty.¹ The importance of possible associations between supernumerary nipples and other congenital anomalies has been emphasized by other authors.² Ferrara et al. showed a high incidence of kidney/urinary tract anomalies in children with supernumerary nipples.³ In this report, we describe a patient with diastometamyelia and polymastia which has not been reported as an association of these two anomalies before.

Case report

The patient is an 11 years- old young girl, the first of four children born to a 38-year-old mother and 52-year-old father. Information on the fertility characteristics of the mother and genetic disorders among family was gathered. According to the survey, there is no parental consanguinity and family history of similar malformations. The patient presented with thoracic deformity and right polythelia. There was no history of supernumerary nipples. The deformity included a right angled scoliosis with an evident asymmetry (Figure 1,2).

Başvuru Tarihi: 08.12.2010, Kabul Tarihi: 31.12.2010

A computer tomographic evaluation showed a normal views of trachea, bronchioli, aorta, pulmonary artery and vena cava. There was a butterfly deformity of 4.th vertebral corpus and irregular costal structures at left inferior thoracic region. Although there was no pathology in abdominal ultrasonographic examination, a CT analysis revealed L1, L5 and sacral vertebral posterior fusion defects and L2 diastometamyelia at antero-posterior level (Figure 3).

There were no renal or other intraabdominal anomalies. The left accessory polymastia was planned for breast ablation surgery. Under general anesthesia left accessory upper nipple areola and underlying mammary gland ablation was performed through a circular nipple incision. A suction drain was used at early postoperative days for removal of seroma. An uneven healing was obtained (Figure 4).

Genetic evaluation of the patient, cytogenetic study was carried out to obtain the chromosomal aberrations in this family. G-banding chromosomal analyses of the patient and the parents were performed on cultured peripheral blood lymphocytes. For chromosome analysis, whole blood (0.2 ml) from patient and the parents were taken and cultured in chromosome medium at 37 C for 72 h. The cells were exposed to colchicine (0.06 µg/mL, Sigma C9754) 2 h before harvesting. The cells were harvested by 0.4% KCl as hypotonic solution and methanol: glacial acetic acid

(3:1) as fixative. Then the cells were spread on glass slides and air dried. The slides were stained with Giemsa according to fluorescence plus Giemsa techniques. 100 well spread metaphases per donor were examined at 1000x magnification for occurrence of different types of chromosome aberrations. The karyotypes of the patient and the parents were normal.



Figure 1. Anterior view of thoracic deformity



Figure 2. Lateral view of the deformity

Discussion

Nipple excision or accessory breast ablation is performed for cosmetic reasons in this group of patients. Neoplastic and or fibrocystic developments may occur within these structures. We chose upper polymastia for ablation as the thoracic deformity guided us pointing that needed for surgery. The scoliosis was associated with diastematomyelia in our case. Diastematomyelia is a spinal dysraphism characterized by the presence of an osseous, cartilaginous, or fibrous septum that splits the spinal cord into two distinct hemicords. These split-cord malformations (SCMs) can be further classified as to whether each hemicord is contained within its own dural sac (type I) as opposed to containment of both hemicords within a single

common dural sheath (type II) A rare adult presentation of diastematomyeli with extramedullary epidermoid tumor and frontal bone agenesis in a patient of spinal dysraphism (split cord malformation hemicord with intervening bony spur) has been reported.^{4,5} SCMs are often accompanied by other cord or column anomalies, including tethered cord, dermoid or epidermoid tumors, syringomyelia, and scoliosis.⁶



Figure 3. Chest x-ray showing scoliosis and spinal dysraphism



Figure 4. After ablative surgery, the view of the patient

The aetiology of congenital anomalies and intellectual impairments are multifactorial, such that a combination of genetic and environmental factors most likely are involved, but is commonly accepted that genetic factors play an important role. We report herein a case with systemic abnormalities without associated chromosomal abnormalities. Pedigree analysis of family showed that this patient is a sporadic case. We think these

Polymastia with Diastometamyelia: Case Report

malformations became mutations or suppression on expression of development genes. The sporadic mutations can be accompanied by potential environmental risk factors, such as exposure to poison and hazardous materials, alcohol abuse, and disease during the mother's pregnancy. Further study is needed to clarify the relationship.

References

1. Fama F, Gioffre Florio MA, Cillari SA, et al. Breast abnormalities: a retrospective study of 208 patients. *Chir Ital* 2007;59(4):499-506.
2. Colombo M, Maestri L, Lambiase R, Magni LA. Polythelia as a sign of other congenital malformations: a clinical example. *Pediatr Med Chir* 1994;16(4):399-400.
3. Ferrara P, Giorgio V, Vitelli O, et al. Polythelia: still a marker of urinary tract anomalies in children? *Scand J Urol Nephrol* 2009;43(1):47-50.
4. Porensky P, Muro K, Ganju A. Adult presentation of spinal dysraphism and tandem diastometamyelia. *Spine J* 2007;7(5):622-6.
5. Nayak PK, Mohagota AK. Frontal bone agenesis in a patient of spinal dysraphism. *Pediatric Neurosurg* 2006;42(3):171-3.
6. Sheehan JP, Sheehan JM, Lopes MB, Jane JA Sr. Thoracic diastematomyelia with concurrent intradural epidermoid spinal cord tumor and cervical syrinx in an adult. *J Neurosurg* 2002;97(2 Suppl):231-4.

Correspondence Address: Sengul YUKSEL Ph.D

Inonu University, Medical Faculty,
Department of Medical Biology and Genetics,
Malatya 44069
Tel: +90-422 34106 60/1248
Fax: +90-422-341 00 36
e-mail: syuksel@inonu.edu.tr