OLGU SUNUMU / CASE REPORT

Dandy-Walker Malformation with Postaxial Polydactyly: A Case Report

POSTAKSİAL POLİDAKTİLİ İLE BİRLİKTE DANDY-WALKER MALFORMASYONU: OLGU SUNUMU

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Abstract

Dandy-Walker malformation is a rare congenital malformation which involves the cerebellum, fourth ventricle and characterized by agenesis or hypoplasia of the cerebellar vermis, cystic dilatation of the fourth ventricle, and enlargement of the posterior fossa. A large number of concomitant problems may be present with the syndrome. Approximately 70-90% of patients have hydrocephalus, which often develops postnatally. Postaxial polydactyly have been reported as a new syndrome and a non-central nervous system associated malformation in the complex. Supporting this point in this case, we are describing an infant with postaxial polydactyly with Dandy-Walker malformation.

Key Words: Polydactyly, hydrocephalus, Dandy-Walker syndrome

Özet

Dandy-Walker malformasyonu, serebellar vermis agenezisi veya hipoplazisi, 4. ventrikülün kistik dilatasyonu ve posterior fossa genişlemesi ile karakterize; 4. ventrikül ve serebellumu içeren nadir konjenital bir malformasyondur. Birçok sayıda eşlik eden problem bu sendromda mevcut olabilir. Hastaların yaklaşık %70-90'ında postnatal olarak hidrosefali gelişir. Postaksial polidaktili yeni bir sendrom ve santral sinir sistemi dışı malformasyon olarak rapor edilmiştir. Bu vakada bu noktayı desteklerken, postaksial polidaktiliye sahip bir infantı tanımlıyoruz.

Anahtar Kelimeler: Polidaktili, hidrosefali, Dandy-Walker sendromu

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Dandy-Walker Malformation (DWM) can be described as a disorder in the development of the central nervous system characterized by partial or complete absence of the cerebellar vermis, posterior fossa cyst communicating with the fourth ventricle presumably representing agenesis of the foramina of Magendia and Luschka and facultative hydrocephalus.[1] The characteristic triad of Dandy-Walker malformation as consisting of (a) complete or partial agenesis of the vermis, (b) cystic dilatation of the fourth ventricle, and (c) an enlarged posterior fossa with upward displacement of lateral sinuses, tentorium, and torcular herophili. The triad typically is found in association with supratentorial hydrocephalus, which should be considered a complication rather than part of the malformation complex.

DWM can be a component of mendelian disorder or chromosomal abnormality. The chromosomal aberrations may occur as a result of teratogenic exposure with multifactorial characteristics. It may also result from environmental factors including viral infections, alcohol and diabetes.[2] As the etiology is heterogeneous, and familial occurrence has been reported the disease may rarely be inherited as an autosomal recessive pattern. A few cases resulting from autosomal recessive genes have been reported, although in most patients, the

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cause of Dandy-Walker malformation is not known. Genetic counseling is critical to estimate the risk of recurrence of genetic disorders in family members.

The incidence of DWM is about 4% among infants with hydrocephalus.[3] Besides mendelian disorders, DWM has also been described in Joubert-Boltshauser syndrome[4] and in oral-facial-digital syndrome type II or Mohr syndrome.[5] DWM can occur as a part of more complex disorders, including Meckel syndrome,[6] Ellis-Van-Creveld syndrome,[7] 3C syndrome[8] and renal hepatic pancreatic dysplasia.[9] DWM and postaxial polydactyly have been described as a probable autosomal recessive syndrome. The association of DWM with postaxial polydactyly has also been reported.[10,11]

Clinical Report

In the current case, the mother was 28 and the father was 33 years old. The parents were second-degree relatives and the presented case was their second pregnancy. The first child of the couple was a male child without any abnormality. DWM was diagnosed during routine examination with ultrasonography at 24th weeks of gestation in second pregnancy of the couple. Nuchal translucency (NT) measurement was normal (1.3 mm) at the 12th week of gestation. Ultrasonographic study performed at the 24th week of gestation, ascertained macrocephaly and mild polyhydramnios. The fetal head was enlarged with increased biparietal diameter (BPD), corresponding to 32nd week of gestation. When the condition was explained to the family and they preferred to continue the pregnancy. Ultrasonography report at 32nd weeks of gestation included polyhydramnios, cerebellar vermis agenesis, posterior fossa cyst and polydactyly of the four limbs (Figure 1,2). The BPD measurement was markedly increased (92 mm) corresponding to 38th week of gestation. Pelvic dilatation was ascertained in the left kidney. The male fetus was born by caesarean section at 32nd weeks of gestation as the mother had a previous caesarean

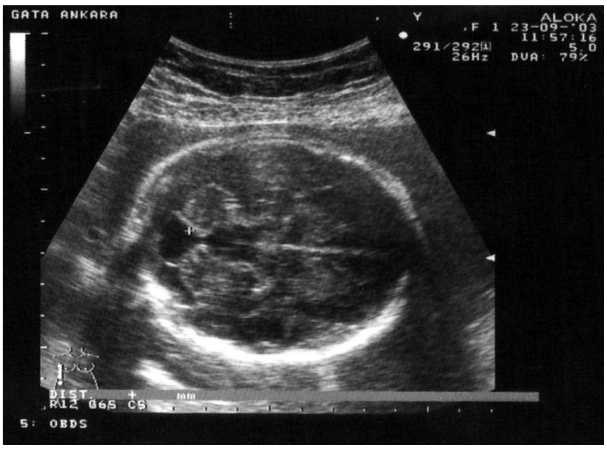

Figure 1. Cerebellar vermis agenesis, posterior fossa cyst.

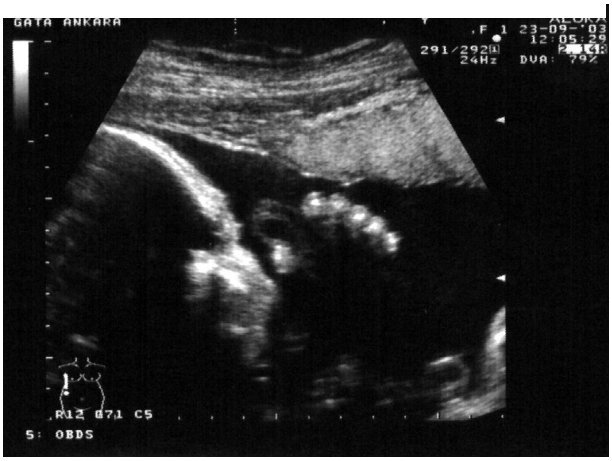

Figure 2. Polydactyly.

operation. The apgar scores were 6 and 7 at 1st and 5th minutes respectively. Assisted ventilation was performed. The birth weight was 2350 g, the height was 43 cm and a large head and postaxial polydactyly of the four limbs were noted. Head, occipitofrontal and chest circumferences were 36 mm, 42.1 cm (> 97 centile), 31.2 cm (50-75th centile), respectively. Cerebral ultrasonography of the newborn following birth demonstrated the characteristic signs of Dandy-Walker syndrome. The 4th ventricle was enlarged, cerebellum was small and the vermis was absent. Dysmorphisms including macrocephaly, large and depressed nasal

bridge, low-set ears were ascertained as facial anomaly.

A number of small hepatic cysts and mild renal pelvic dilatation were observed by the abdominal ultrasound. Karyotype analysis was normal (skin fibroblasts) (46,XX). Repeated generalized convulsions led to apnea and bradycardia on the 3th day and the infant died on the 4th day.

The basal ganglia and the thalamus appeared to be normal during the autopsy. Left ventricular wall hypertrophy was observed during the examination of the cardiovascular system. There were small, multiseptal cysts in liver. The spleen was normal. There weren't any other visceral abnormalities observed, otherwise.

Discussion

DWM is a central nervous system abnormality, which can be associated with a variety of well-defined conditions.[2] DWM with polydactyly has been described as isolated or associated with other malformations. Hart et al. in a study of 28 cases, described 3 cases of the DWM associated with polydactyly.[1] There are lots of syndromes associated with DWM. Boltshauser and Isler, Joubert et al., describe Joubert-Boltshauser syndrome, an autosomal recessive disorder that consists of episodic hyperpnea, abnormal eye movements, ataxia and cerebellar vermis agenesis associated with DWM in most cases.[4,12] Gustavson et al. describe Mohr syndrome (or oro-facial-digital syndrome type II) characterized by bilateral polysyndactyly of halluces, fleshy tumors of the tongue, and autosomal recessive inheritance which is sometimes associated with DWM.[13] Planas et al., suggest a new syndrome including mesomelic camptomelia, postaxial hexadactyly and DWM[14] Hydrolethalus syndrome described by Salonen in 24 patients is uninformly lethal and consists of many congenital malformations including polyhydramnios, severe hydrocephalus, cleft lip/plate, respiratory tract abnormalities, micrognathia, and often preaxial polydactyly in feet.[15,16] It has an autosomal recessive inheritance.[17] Meckel syndrome can be ruled out since there are no histologic renal anomalies. DWM associated with polydactyly is usually preaxial. Only two cases of postaxial polydactyly associated with DWM are reported in literature.[11,18] In this case we are supporting the conclusion that autosomal recessive syndrome including DWM with postaxial polydactyly may be a new syndrome.

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