Demirel et al., J Genet Disor Genet Rep 2018, 7:3
DOI: 10.4172/2327-5790.1000178

Journal of Genetic Disorders & Genetic Reports

Case Report

A SCITECHNOL JOURNAL

Cerebellar Hypoplasıa as a Manıfestatıon of 6q25 Deletıon in a Preterm Newborn

Demırel G[1*], Vatansever B[1], Karavar H[1], Gundogdu S[1], Ertan G[2] and Tastekın A[1]

[1]*Division of Neonatology, Istanbul Medipol University, Istanbul, Turkey*

[2]*Department of Radiology, Istanbul Medipol University, Istanbul, Turkey*

[*]**Corresponding author:** Gamze Demirel, Division of Neonatology, Faculty of Medicine, Istanbul Medipol University, Istanbul, Turkey, Tel: +90 212 4707000-8545; E-mail: fgdemirel@medipol.edu.tr

Received Date: August 04, 2018; **Accepted Date:** September 07, 2018; **Published Date:** September 14, 2018

Abstract

Deletions of chromosome 6q are rare and almost all have some form of craniofacial dysmorphisms and structural brain malformations such as corpus callosum agenesis, colpocephaly, polymicrogyria and hydrocephalus. Here we report a preterm infant with cerebellar and pontine hypoplasia as a presentation of terminal deletion of 6q25.

Keywords: 6q25 deletion; Cerebellar hypoplasia; Preterm infant

Introduction

Deletions of chromosome 6q are rare and almost all have some form of craniofacial dysmorphisms [1,2]. Short neck, broad nose, low set ears, hypertelorism, downslanting palpebral fissures are some of them. In postnatal cases, brain abnormalities, intellectual disabilities, hypotonia are also seen. Structural brain malformations including venticulomegaly, corpus callosum agenesis, colpocephaly, polymicrogyria and hydrocephalus associated with 6q deletions have been reported [3]. Here we report a preterm infant with cerebellar hypoplasia as a presentation of terminal deletion of 6q25.Materials and Methods

Case Report

Female, born at 30 2/7 gestational weeks, by ceserean section, with a weight of 1200 g, length of 34 cm and head circumference of 26 cm. She was the 5th infant of nonconsangious marriage. Prenatal ultrasonography (USG) revealed cerebellar hypoplasia and enlarged cysterna magna. She was admitted to neonatal intensive care unit with diagnosis of prematurity, and respiratory distress syndrome. Intratracheal surfactant was administered. She presented the following main dysmorphic features on physical examination: narrow forehead, downslanted palpebral fissures, hypertelorism, broad nose, low set ears and flat filtrum (Figure 1). Kranial USG showed bilateral hypolasia of cerebellar hemispheres and vermiş and enlarged cysterna magna. Cranial magnetic resonance imaging (MR) showed hypoplasia of pons and cerebellum (Figure 2). Echocardiography was performed due to heart murmur and showed perimembranous ventricular septal defect and secundum atrial septal defect. Lisinopril and furosemid treatment was started. Renal USG was normal. On 10th day of life, acute renal insufficieny began, due to unresponsiveness to medical therapy, peritoneal dialysis was performed. The patient was entubated and connected to mechanical ventilator due to respiratory insufficiency. Eye examination revealed bilateral zone 2, grade 2 retinopathy due to prematurity, optic nerve was pale. On follow up, the patient has abdominal distension, biliary residues via feeding catheter, and persistant intestinal segments on abdominal graphy. On operation necrotic intestinal segments was seen, two drenous catheters was installed. After few weeks, barium graphy showed normal intestinal passage and feeding was started and increased to full enteral amounts. Steroid therapy was started for bronchopulmonary dysplasia but it had no benefit. We consider cerebellar and pontine hypoplasia was the cause of respiratory insufficiency and trakeostomi was planned. On chromosomal analysis of peripheral blood tis 46, XX, del(6) (q25) (Figure 3). Parental chromosomal analysis was planned.

Figure 1: Dysmorphic facial appearance of the infant: narrow forehead, downslanted palpebral fissures, hypertelorism, broad nose, low set ears and flat filtrum.

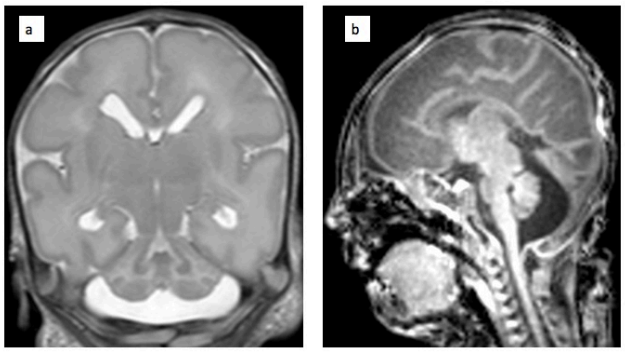

Figure 2: (a) Coronal T2-weighted. **(b)** Sagittal T1-weighted image show hypoplasia of the cerebellar hemispheres and pons.

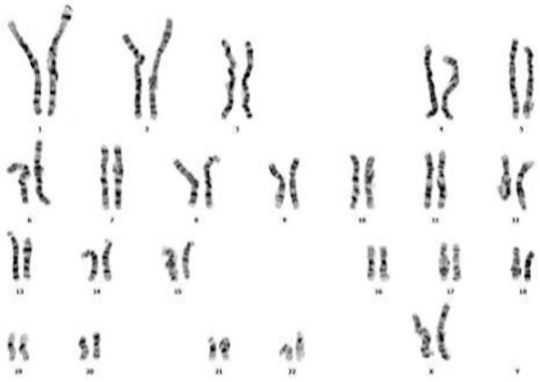

Figure 3: Karyotype analysis of the patient.

Discussion and Conclusion

Isolated 6q subtelomeric deletions are relatively rare and very few cases have been reported [3-6]. The most common clinical features include dysmorphic features, microcephly, facial dysmorphism, cardiac defects, seizures, hemivertebra and brain abnormalities such as hydrocephalus and abnormal corpus callosum [4,6-8]. On nadir reports on this subject, cases are usually diagnosed after infancy period, while evaluating for dysmorhic features, seizures or brain malformations [4]. In a unique case report, prenatal evaluations of chromosomal analysis after USG findings such as structural brain abnormalities like agenesis of corpus callosum, hydrocephaly and ventriculomegaly stated 6q25 deletion on karyotype analysis [2].

In our case, during neonatal period kranial MR revealed hypoplasia of pons and cerebellum. To our knowledge our case is the first case with 6q deletion associated with cerebellar and pontine hypoplasia. Previously reported cases and our case indicate that structural brain abnormalities might be manifestation of deletions of chromosome 6q. Combination of dysmorphic facial appearence and MR findings showing brain abnormalities such as pontine and cerebellar hypoplasia are an indication for testing subtelomeric rearrangements.

References

1. De Vos IJ, Stegmann AP, Webers CA, Stumpel CT (2017) The 6p25 deletion syndrome: An update on a rare neurocristopathy. Ophthalmic Genet 38: 101-107.
2. Cetin Z, Sanhal C, Guzel BN, Toru S, Yakut S (2013) Prenatal diagnosis of isolated ventriculomegaly as a second trimester manifestation of de novo terminal 6q25 deletion. Genet Couns 24: 427-429.
3. Michelson M, Ben-Sasson A, Vinkler C, Leshinsky-Silver E, Netzer I, et al. (2012) Delineation of the interstitial 6q25 microdeletion syndrome: Refinement of the critical causative region. Am J Med Genet A 158: 1395-1399.
4. Elia M, Striano P, Fichera M, Gaggero R, Castiglia L, et al. (2006) 6q terminal deletion syndrome associated with a distinctive EEG and clinical pattern: A report of five cases. Epilepsia 47: 830-838.
5. Eash D, Waggoner D, Chung J, Stevenson D, Martin CL (2005) Calibration of 6q subtelomere deletions to define genotype/phenotype correlations. Clin Genet 67: 396-403.
6. Lee JY, Cho YH, Hallford G (2011) Delineation of subtelomeric deletion of the long arm of chromosome 6. Ann Hum Genet 75: 755-764.
7. Wadt K, Jensen LN, Bjerglund L, Lundstrom M, Kirchhoff M, et al. (2012) Fetal ventriculomegaly due to familial submicroscopic terminal 6q deletions. Prenat Diagn 32: 1212-1217.
8. Li Y, Choy KW, Xie HN, Chen M, He WY, et al. (2015) Congenital hydrocephalus and hemivertebrae associated with de novo partial monosomy 6q (6q25.3-->qter). Balkan J Med Genet 18: 77-84.