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EC OPHTHALMOLOGY
Case Report

Edwards Syndrome (Trisomy 18) with Eye Disorder

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Abstract

Edwards Syndrome or trisomy 18 is a chromosomal disorder due to an extra copy of chromosome 18, mosaic trisomy or partial 18q trisomy. The syndrome is associated with multiple malformations involving any organ or system and have been considered lethal owing to its poor prognosis. In trisomy 18 generalized abnormalities affect all systems. However, ocular findings in this entity are rarely reported. Here, we presente a case with confirmed trisomy 18 with ophthalmologic examination revealing corneal opacities and palpebral abnormalities.

Keywords: *Trisomy 18; Newborn; Corneal Opacity; Eye Disorder; Case Report*

Abbreviation

ES: Edwards's Syndrome

Introduction

Edwards syndrome (ES), also known as Trisomy 18, is a rare chromossomal disorder with a broad clinical picture, with involvement of multiple organs and systems, being the second most frequently observed autosomal trisomy at birth, after only to Down syndrome (trisomy of the chromosome 21). It was first reported in 1960 by Edwards., *et al*. who reported a newborn with multiple congenital malformations and cognitive deficit [1]. The disorder occurs in approximately 1 to 3600 - 8500 live births and more commonly affects females than males, in the ratio of almost 3:2. However, the fetal loss is higher in males than females, and females had better survival than males. It is clinically characterized by a broad clinical picture, with more than 130 different findings already reported, involving various organs, and a prognosis that is considered poor. Sometimes, ES is not suspected during pregnancy. Most fetuses diagnosed during gestation are spontaneously aborted and, among those that are born alive, most die within the first week. Less than 10% survive the first year [2].

The ES phenotype results from full, mosaic or partial trisomy 18q. Full trisomy 18 is the most common form occurring in almost 95% of cases and every cell contains 3 full copies of chromosome 18. Mosaics or partial trisomy can occur in 5% of cases in where some cells are normal with 46 chromosomes and others have the extra chromosome [3]. The clinical presentation of ES is characterized by low birth weight, craniofacial and skeletal abnormalities, congenital heart anomalies, gastrointestinal abnormalities, urogenital abnormalities,

neurological problems and mental deficiency. However, none of the clinical features is pathognomonic [1-3]. Ocular findings may accompany this syndrome [4]. The common eye-related disorders include the anomalies of palpebra, cornea, anterior uveal tract, lens, retina, and optic disc. We reported here a case of ES with bilateral cloudy cornea due to its rarity.

Case Report

A female neonate was born by cesarean section to a healthy 30-year-old woman, gravida 3 para 3, at 38 weeks and 06 days of gestational age. Birth weight was 3135g. The mother had received regular prenatal care, with no complications. Family history was unremarkable for congenital abnormalities, and the older siblings were normal. The child was born hypotonic and cyanotic, with Apgar scores 4 at 1 minute and 7 at 5 minutes. She was transferred to our neonatal intensive care unit for respiratory distress and heart disease murmur suggesting the presence of malformation. Her length was 49 cm, head circumference was 35 cm, and chest circumference was 33 cm. Physical examination revealed multiple congenital anomalies, including low-set ears, associated with preauricular appendages on the left, micrognathia, cryptorchidism, polydactyly in the hands, and a prominent occiput. Ocular examination revealed ptosis, long and sparse eyelashes, small palpebral fissures, and bilateral corneal opacities (Figure 1). The lens was clear without cataracts. No iris coloboma was noted. Central nervous system examination revealed hypotonia. Ultrasound of the brain revealed thinning of the corpus callosum and cerebellar hypoplasia. Echocardiography showed a ventricular septal defect, obstruction of right ventricular outflow tract, aortic root overriding the ventricular septum, and right ventricular hypertrophy, compatible with fallot tetralogy. Ultrasonography of the liver, spleen, and kidney were normal. The karyotype result confirmed the diagnosis of ES with complete trisomy (47, XX, +18 chromosomes). The infant was mechanically ventilated and she died of cardiopulmonary failure at 44 days of age. An autopsy was not performed.

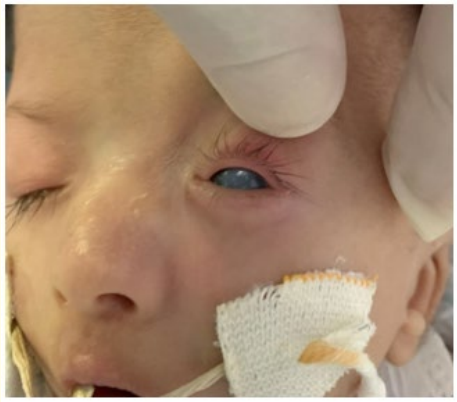

Figure 1: *Facial appearance of our patient. Micrognathia, long eyelashes, corneal opacity, low-set ears with prearicular appendages can be observed.*

Discussion

ES is a chromosomal lethal condition with about one-third dying in the neonatal period. The findings ranges from mild presentation with no internal organ malformation to the classic presentation of this syndome. Risk factors for the disease include a positive family history in close relatives and rising maternal age [5-7]. In this case were observed abnormalities in cardiovascular system, musculoskeletal, craniofacial, urogenital, and central nervous system. The full trisomy of chromossome 18 was found in karyotype analysis.

The clinical pattern of ES is well defined and rarely misdiagnosed. Most cases in the developed countries are suspected prenatally based on screening by maternal age, detection of sonographic abnormalities during second or third trimester, and measurement of

nuchal translucency in the first trimester. After birth, it is diagnosed by peripheral blood karyotype. The differential diagnosis includes trisomy of chromossome 13 or chromossome 9, Werding-Hoffman's disease, CHARGE syndrome, Pena-Shockeir syndrome, and VACTERL association [2]. The main causes of death include central apnea, heart failure or respiratory faiure or a combination of these factors.

In addition to many severe systemic malformations, some ocular findings have been reported, but in less than 10% of trisomy 18 cases. Most common described anomalies of the eyes involve the ocular adnexae, especially eyelids and orbits. However, serious ocular involvement as corneal opacity, microphthalmia, cataract, strabismus, congenital glaucoma and optic atrophy have been reported clinically [2,3].

Many ocular pathological findings have been reported [4,7,8]. Iris stromal hypoplasia, abnormal lens shape, and decreased ganglion cells in the retina in a case described by Calderone., *et al*. [4]. Bilateral completely opaque corneas, elevated intraocular pressure, and various other anomalies were found in siblings with trisomy 18 [7]. Velzeboer., *et al*. [8] reported the ophthalmic histopathology in three cases of trisomy 18. In two cases the cornea was normal, while in one case hypercellularity of the stroma was found and Bowman's and Descemet's layers were absent. A minimal cataract was also found in two cases. Retinal folds were a common finding in the posterior region in all three cases. In one case, a partial coloboma of the optic disc was present in both eyes.

Conclusion

This case is one of the rare reports of ptosis, long and sparse lashes, and corneal opacities in ES. Although ocular findings are not diagnostic for this syndrome, ophthalmologic evaluation may be helpful in differentiating trisomy 18 from other ilnesses, and to provide more precise information. Given the limited life expectancy for most patients with trisomy 18, aggressive medical and surgical ophthalmic care is generally not advised and palliative/supportive care options are preferred. However, in patients with better prognoses, interventions may be performed.

Conflict of Interest

Authors report no conflict of interest.

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