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Case Report

Superior Eyelid Coloboma as A Manifestation of Triple X Syndrome

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Introduction

A superior eyelid coloboma is a congenital malformation affecting the skin, muscles, tarsus, and conjunctiva of the eyelid margin, usually located in the medial part, with absence of eyelashes and glands.

It is often unilateral, but bilateral colobomas are occasionally observed.

Many reports of superior eyelid coloboma describe various ocular and systemic anomalies. This case report illustrates a superior eyelid coloboma revealing a triple X syndrome.

Materials and Methods

We report a case of a superior eyelid coloboma that led to the diagnosis of a Triple X syndrome.

Results

A 5-day-old female newborn, born at term, following a poorly monitored pregnancy. Delivery was performed by cesarean section, at the maternity ward of the Mohamed V Military Instruction Hospital in Rabat. The newborn had an APGAR score of 10/10, with spontaneous crying at birth. Clinically, facial asymmetry was noted, with the mouth slightly deviating to the right but without hypoplasia. The eyelids were asymmetrical, showing a superior right eyelid coloboma **(Figure 1)**. Additional anomalies included a tubercle beneath the left nostril and bilateral preauricular appendages, although the ears were otherwise well-formed **(Figure 2)**.

Discussion

The ophthalmological examination revealed: a coloboma of the superior right eyelid, a symblepharon of the lower right eyelid extending to the inferior limbus. The corneal diameters were 11 mm in both eyes. The intraocular pressures (measured with a Perkins tonometer) were 11 mmHg in the right eye and 10 mmHg in the left eye. The corneas were normal, the anterior chambers were well-formed, the lenses were normal, and the fundus examination showed no abnormalities. The rest of the examination was unremarkable. A karyotype was requested, showing the presence of an extra X chromosome **(Figure 3)**. We concluded the diagnosis of Triple X syndrome. We opted to delay surgical intervention, instead initiating medical treat-

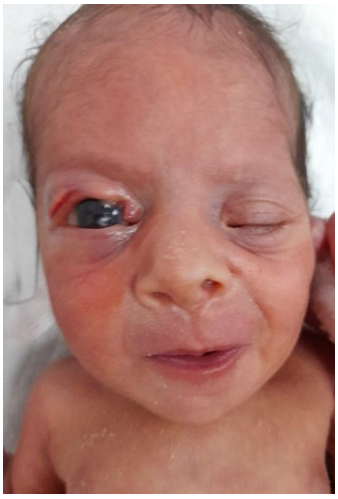
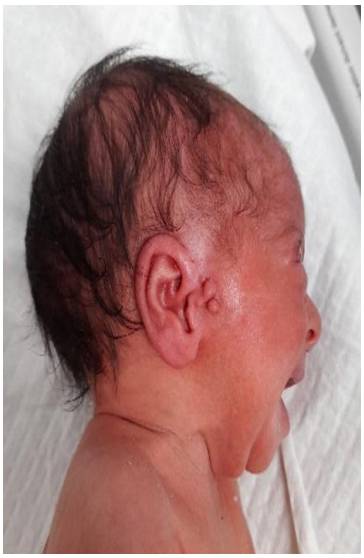


Figure1: The newborn presented facial asymmetry, with the mouth deviating slightly to the right without hypoplasia. The eyelids were also asymmetrical, showing a superior right eyelid coloboma.



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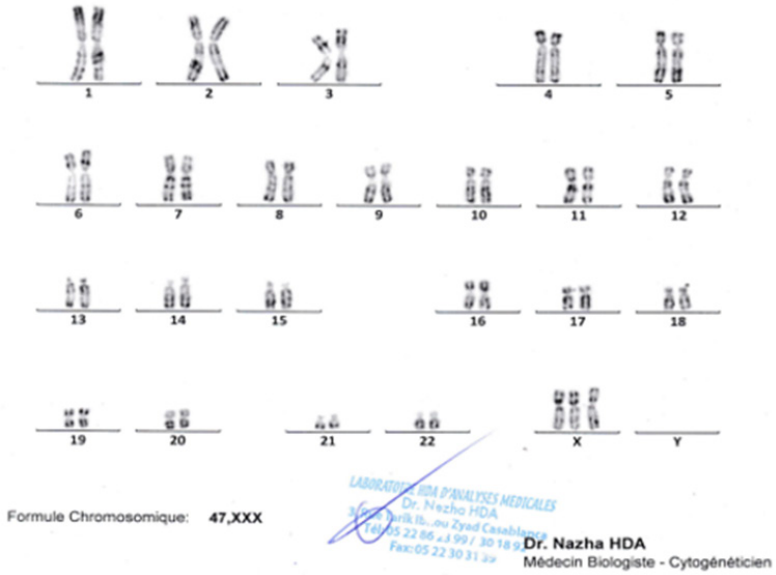

Figure 3: A karyotype showing the presence of an extra X chromosome.

ment with lubricating eye drops, awaiting surgery at the age of 3 months.

The first 47, Triple X karyotype was described by Jacobs et al. in 1959. Cases of female infants with the 47, XXX phenotype are relatively common, most of these infants have a normal phenotype. Only a few cases of X trisomy are reported in the literature with congenital malformations. Our case comes from phenotypically normal parents and presents a superior right eyelid coloboma with facial dysmorphia and multiple preauricular tubercles. In general, colobomas may be part of polymalformative syndromes such as CHARGE syndrome, Goldenhar syndrome, Goltz syndrome, etc., or be related to chromosomal abnormalities such as trisomies 13, 18, and 8, cat eye syndrome, Turner syndrome, and Klinefelter syndrome. It is very rare to find a coloboma associated with Triple X syndrome. This genetic anomaly is generally of sporadic origin, where the X chromosomes in these patients fail to separate during a process called nondisjunction. Only one of the three X chromosomes is activated, and the other two are inactivated. The variable phenotypic anomalies mentioned above are likely due to the overexpression of genes located on the extra X chromosomes that escape X inactivation.

Conclusion

Eyelid colobomas are generally part of various syndromes such as CHARGE, Goldenhar, Goltz, or related to chromosomal abnormalities such as trisomies 13, 18, and 8, Turner and Klinefelter syndromes. In very rare cases, they may be associated with Triple X syndrome.