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Chorioretinal Coloboma Cyst: Case Report

I.Chaouche -M.Haloua-N.BOUARDI-B.Alami-Y.Lamrani- M.Maaroufi -M.Boubbou.

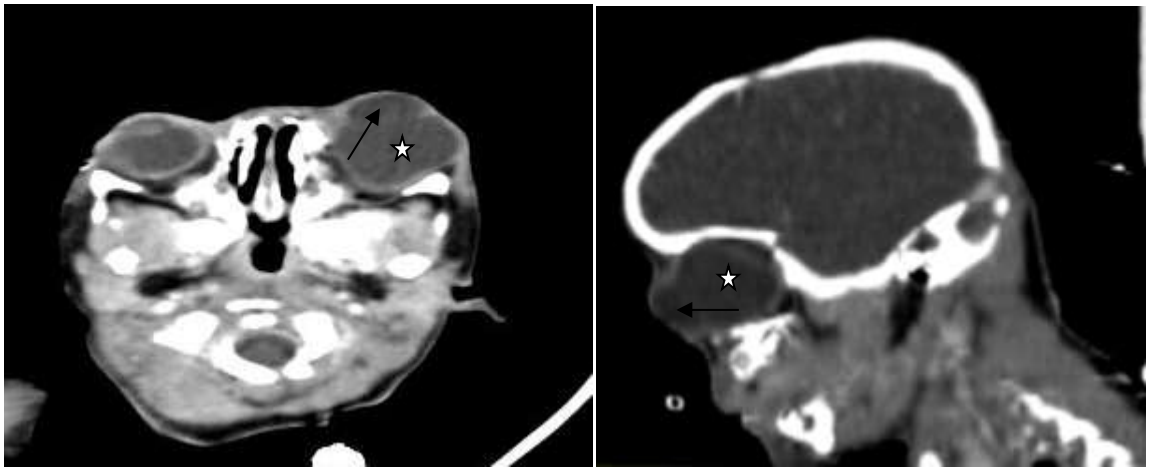
Radiology Service –UH Hassan II-FES-Morocco.
Faculty of medicine and pharmacy-Fez
University sidi mohammed benabdellah

***Abstract**: Chorioretinal coloboma cyst it's a rare congenital pathology, associated with microphthalmia. Coloboma is an embryological defect in the formation of the iris or choroid. The coloboma cyst is a cystic formation through an embryological defect of the eyeball. The treatment is based on surveillance in small cysts and surgery in large cysts.*

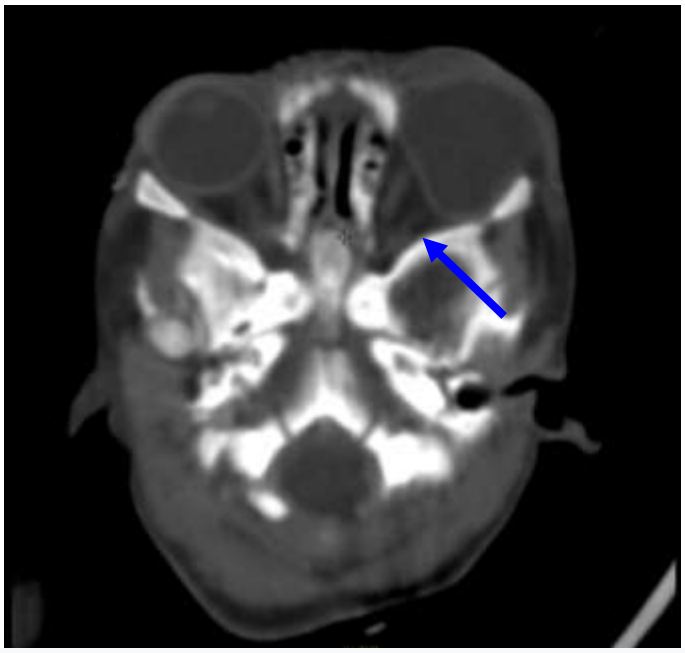
Keywords: Cloboma cyst- Ct-scan- genetic

INTRODUCTON:
The coloboma cyst of the orbit is a rare, non-hereditary anomaly. It creates a cavity lined with neuro-ectrodermic tissue communicating with the vitreous cavity [1].
The diagnosis is evoked clinically and confirmed radiologically by B-mode ultrasound, CT scan and MRI [1].
In our case we report a form of coloboma cyst with chorioretinal location and microphthalmia.

CASE REPORT:
In our case, we report a 30-day-old little girl, having consulted in the pediatric emergency for left orbital tumefaction since birth, which had gradually increased in size.
The pregnancy was uneventful with delivery at term.
The clinical examination objectified an orbital tumefaction with proptosis without inflammatory signs.
We performed an orbito-cerebral scanner with injection of contrast product.
On the CT-scan we objectified the presence of a voluminous retroocular cystic lesion, communicating with the vitreous cavity, which is oval in shape, with well-defined regular contours, measuring 20x22mm. Compresses and pushes the eyeball forward, causing grade III proptosis
On the scanner we also objectified the absence of visualization of the lacrimal gland and the lens
In the case of our patient, there was no indication for immediate surgery because the current standard of care is observation of orbital cysts to allow the orbit to reach approximate adult volume.



Axial section and sagittal reconstruction: retroocular cystic formation ( ) communicating with the vitreous cavity with microphthalmia ( ) and proptosis grade III



Axial section: retroocular cystic formation and optic nerve hypotrophy

DISCUSSION:

Microphthalmia with orbital cyst is a rare colobomatous manifestation of failed closure of the orbital fissure.
The diagnosis of colobomatous cyst of the orbit can be suspected clinically when it is large or associated with microphthalmia or papillary coloboma [2]. Elsewhere the diagnosis is based on imaging by the ultrasound B, CT and MRI [3].
Cyst coloboma can be part of a hereditary polymalformative framework, the most frequent are CHARGE (Coloboma, Heart defect, Atresia choanae, Retarded growth, Genital anomalies, Ear anomalies), Rubinstein-Taybi, Goldenhar, Goltz, Lenz, Warburg, Aicardi syndromes also be related to chromosomal abnormalities: trisomies 13, 18 and 8; triploidy; cat's eye, Turner, Klinefelter syndromesTheir research imposes a complete general assessment [4].
On the ultrasound, the coloboma cyst is manifested by an anechoic formation, rounded, of regular contours with hypoplasia of the optic nerve.
On CT scan, the coloboma cyst is manifested by a hypodense cystic formation without contrast enhancement
In MRI the morphological sequences are sufficient for the diagnosis; a coloboma cyst is described in hypo signal T1 hyper signal T2, without contrast enhancement.
The communication with the vitreous cavity is necessary to make the diagnosis [3].
As iris colobomas do not cause visual disturbance, they will not be touched unless the eye needs to be operated on for a cataract or a corneal transplant. Chorioretinal coloboma cysts cause severe visual acuity deficit, whereas iris coloboma cysts can remain asymptomatic.Chorio-retinal colobomas cannot be improved, only the associated retinal detachments can be operated. These are often fragile eyes that make any surgery delicate [3,5].

CONCLUSION:

The colobomatous cyst of the orbit is a rare condition that is often part of a polymalformative syndrome. The therapeutic decision depends on the size of the cyst as well as the condition of the eyeball.

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