IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 20, Issue 8 Ser.11 (August. 2021), PP 43-48
www.iosrjournals.org

A puzzling diagnosis mimicking retinoblastoma: Coats disease: Case report.

Ibtissam Razzouki[1], Meriem Regragui[1], Mehdi Karkouri[1]
Salma Belaaroussi[2] , Asma Bamoussi[2], Asmaa Elkettani[2]
[1](Department of anatomical pathology, University Hospital Center of Ibn Rochd, Casablanca, Morocco)
[2](Department of pediatric ophtalmology, 20 August Hospital , University Hospital Center of Ibn Rochd, Casablanca, Morocco)

Abstract:
Background*: Coats disease is an idiopathic eye disease, primarily affecting young children.*
Its most important differential diagnosis is unilateral retinoblastoma, whose prognosis and therapeutic management are different, hence the importance of the anatomo-clinical correlation.
observation*: This is a 13-month-old boy who presented with leukocoria in his left eye for 4 months.*
The initial ophthalmologic examination showed the negativity of the pursuit, threat, glare and photomotor reflexes.
Examination of the anterior segment revealed edema of the cornea with decreased depth of the anterior chamber.
The pupil was in semi-mydriasis with the presence of a whitish glow of the pupillary air.
Fundus ocular tone was <6mmHg associated with corneal edema in the anterior segment and chamber.
Ultrasound showed the presence of retro-lenticular intravitrous tissue structures extended towards the head of the optic nerve, vascularized by Doppler, suggesting a persistence of the primitive vitreous.
Cranio-orbital magnetic resonance imaging (MRI) showed the presence of a left intraocular mass involving the right temporal and nasal V-shaped fields, without extra scleral or neural extension.
The surgical procedure consisted of enucleation of the left eyeball.
Macroscopic examination showed the presence of a friable yellowish lesion occupying the vitreous body and reaching the posterior chamber.
The optic nerve was without macroscopic peculiarity.
The histological examination objectified, in intra-retinal, areas of necrosis comprising crystals of cholesterol as well as layers of foamy histiocytes and calcifications.
The riddled slide and the optic nerve showed no histologic lesions.
The retina presented telangiectatic vessels and subretinal serosities.
The morphological appearance was therefore compatible with Coats disease.
discussion*: Coats disease is one of the most difficult differential diagnoses of retinoblastoma.*
Patients may present with a variety of signs, such as reduced visual acuity, strabismus, and leukocoria.
Although the clinical examination alone may be sufficient to make a diagnosis, other paraclinical means are useful, namely fluorescein angiography and ultrasound which makes it possible to demonstrate subretinal opacities and detachment of the skin. the retina.
Computed tomography and MRI can exclude retinoblastoma by objectifying calcification.
When it is difficult to decide, diagnostic enucleation is indicated.
Thus, in order to differentiate between Coats disease and retinoblastoma, numerous morphological, ultrasound, radiological and histological criteria are useful.
Conclusion: *Coats disease is a diagnostic trap and its differentiation from retinoblastoma can be difficult.*
General pathologists generally do not encounter this disease because it is diagnosed at an early stage which helps save the eyeball.
Therefore, it is essential for the pathologist to be familiar with the histopathological features of retinoblastoma and to distinguish it from Coat's disease.
Key Word*: Coasts disease, telangiectasis, exsudative retinal detachment*

Date of Submission: 10-08-2021 Date of Acceptance: 25-08-2021

I. Introduction

Coats disease is an idiopathic, non-hereditary eye disease, affecting primarily young children, first described by Coats in 1908 [1].
The main telltale symptoms are early onset reduced vision, unilateral vision loss, strabismus, and leukocoria.
The most important differential diagnosis is unilateral retinoblastoma [2], the prognosis and treatment of which are different, hence the importance of the anatomo-clinical correlation.
In this study, we report a case of Coat's disease in an infant and we evaluate the histopathological and clinical findings.

II. observation

This is a 13 month and 22 day old male child who was brought to the outpatient department at the age of 9 months with a telltale sign of leukocoria in the left eye.
On initial ophthalmologic examination, the pursuit, threat, photomotor and glare reflexes were negative. Examination of the anterior segment revealed edema of the cornea with decreased depth of the anterior chamber. The pupil was in semi-mydriasis with the presence of a whitish glow of the pupillary air.
The fundus obtained after sedation showed ocular tone <6mmHg, slight corneal edema in the anterior segment and anterior chamber, and the absence of iris rubeosis.
Intraocular pressure was <6 mm Hg.

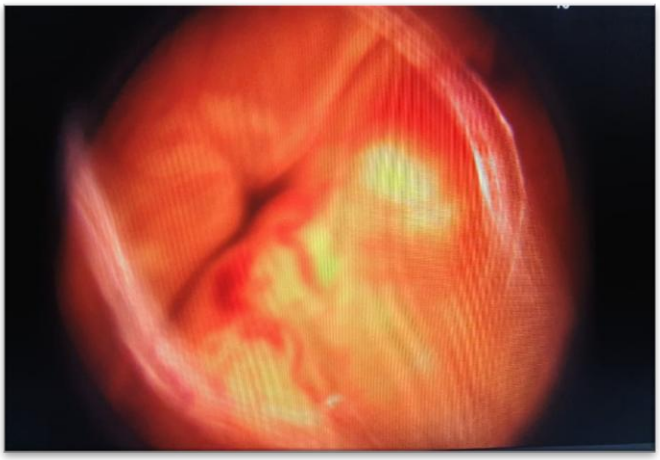

Figure 1: Fundus image taken with a "Retcam" wide-field digital camera showing total retinal detachment

Ultrasound showed the presence of intra-vitreous retro lenticular tissue structure producing a triangular aspect extended towards the head of the optic nerve, vascularized by Doppler, suggesting a persistence of the primitive vitreous.

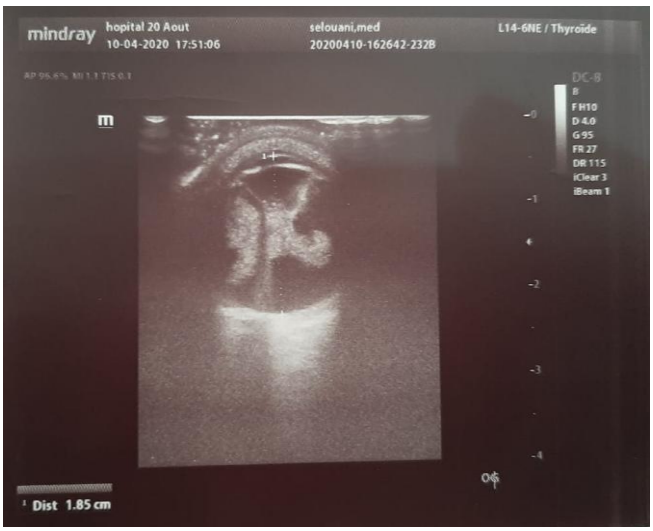

Figure 2: Ultrasound aspects showing the presence of an intra-vitreous lenticular tissue mass producing a triangular appearance (arrow)

Cranio-orbital MRI showed the presence of a left intraocular mass involving the right temporal and nasal V-shaped fields, without extra scleral or neural extension.
The surgical procedure consisted of enucleation of the left eyeball with placement of a silicone ball.
The eyeball was stored for formalin fixation for 2 days.
It weighed 5g, and measured 2x1.8x1.8cm.
The optic nerve was resected over 1 cm.
When cut, it was noted the presence of a friable yellowish lesion, occupying the vitreous body and reaching the posterior chamber, measuring 1.5x1.5x1.5cm.
The optic nerve was without macroscopic peculiarity.
Histological examination of the various samples taken showed that the yellowish lesion described above corresponded to areas of necrosis comprising crystals of cholesterol as well as layers of foamy histiocytes.
Calcifications were also observed.
This necrosis was located intra-retinal, respecting the choroid and the anterior segment.
The riddled slide as well as the optic nerve showed no histological lesion.

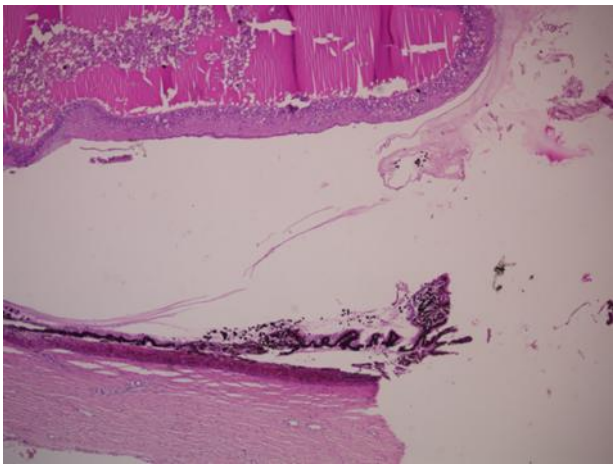

Fig 3: Coats disease: histopathological results. Total exudative retinal detachment (H&E).

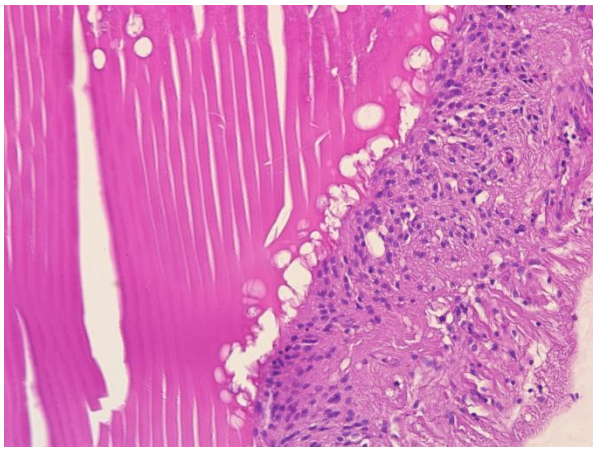

Fig 4: Coats disease: histopathological results. Subretinal fluid with cholesterol clefts and lipid-laden macrophages (H&E. Original magnification × 200).

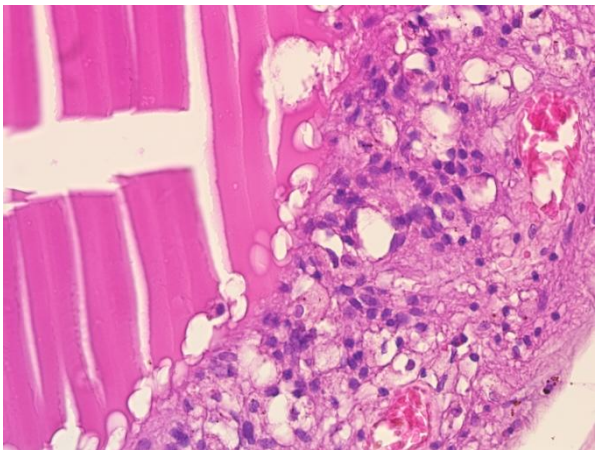

Fig. 5: Coats disease: histopathological results. Subretinal fluid with cholesterol clefts and lipid-laden macrophages (H&E. Original magnification × 400).

The retina presented telangiectatic vessels with the presence of subretinal serosities.
The morphological appearance was therefore compatible with Coats disease.

III. Discussion

Coats disease is one of the most difficult differential diagnoses of early childhood retinoblastoma.
A study [3] carried out on 486 children, the diagnosis of retinoblastoma was made in 408 children (84%) while 78 children presented with another pathology (16%).
In the 78 children with another diagnosis, including Coats disease which was observed in 20 cases (25%) with 2 cases requiring histological confirmation after enucleation,
It is a non-hereditary unilateral disease of uncertain etiology. Although an associated gene has been identified on chromosome 4, it mainly affects boys in infancy (mean: 5 years) [4,5, 6] , but possibly also seen in adults.
Smithen et al. [10] reported Coats disease in 13 patients with an average age of 50 years.
Although these patients have symptoms similar to those of children, the authors said that the disease detected in older patients progresses more slowly than that seen in children and that this may lead to a later diagnosis.

Some cases of Coats disease are asymptomatic, and patients can present with a variety of signs, the most common being reduced visual acuity, strabismus, and leukocoria.
Other signs that may be present in patients with Coats disease are pain, iris heterochromia, and nystagmus.
Shields et al [7] evaluated a series of 150 cases and determined that 34% had low vision, 23% strabismus and 20% leukocoria.
In a study of 97 cases [8], about half had low vision, followed by strabismus and leukocoria.
Although the extent of symptoms varies, Coats disease is primarily a progressive disease, which can be asymptomatic at an early stage and diagnosed during routine eye examinations.
Clinically, patients with Coats disease are characterized by retinal telangiectasias, dilated capillaries, micro aneurysms and arteriovenous malformations [5]. Intra and subretinal lipid exudates are also characteristic [5].
.
The list of differential diagnoses includes retinoblastoma, primary persistent hyperplastic vitreous (PHPV) and toxocariasis [11].
In a study of 500 patients seen for leukocoria at an ophthalmic oncology center in Philadelphia, the cause of leukocoria was 58%, PHPV in 27.8%, and Coats disease in 16% [6].
Meier et al [9] analyzed data from 83 eyes of 58 children with leukocoria up to 7 years of age.
Retinoblastoma was present in only 6% of cases, Coats disease with 2 affected eyes only in 2.4% of cases.

Although clinical examination alone may be sufficient to make a diagnosis of cost's disease, other paraclinical means can also be helpful and are often necessary. To quote, the ultrasound which makes it possible to demonstrate subretinal opacities and retinal detachment.
Fluorescein angiography allows clear visualization of vascular changes.
Retinal ectasia and rarefaction of the peripheral capillary bed are observed, and are characteristic of the disease.
The aneurysms are clearly visible.
In advanced cases, a CT scan shows lipid exudates as a hyperdense area in the eye socket.
MRI has a low contribution in early situations, and its interest lies mainly in advanced cases.

Invasive diagnostic modalities such as fine needle aspiration cytology (FNAC) are not recommended. However, FNAC can be used to confirm the diagnosis when non-invasive modalities are not contributing.
However, FNAC is contraindicated in cases of total retinal detachment or in cases of strong clinical suspicion of retinoblastoma. [12] [7]

Thus, and in order to differentiate between Coats disease and retinoblastoma, numerous morphological, ultrasound and radiological criteria are useful.

In some situations, it is difficult to decide between retinoblastoma and Coat's disease, so diagnostic enucleation is indicated.
Macroscopically: Coats disease takes on a "buff yellow" appearance while retinoblastoma has a "snow-white" appearance.
But reliable differential diagnostic differentiation is still not easy, mainly in the advanced stages of the disease.
In advanced stages, exudative retinal detachment and secondary glaucoma can also occur.
In retinoblastoma, on the other hand, there are usually uniform vasodilations; subretinal lipid exudates are virtually non-existent in retinoblastoma, and subretinal exudates are typical purely serous [13]. Unlike the calcified shadows characteristic of retinoblastoma, intraocular calcifications are only very rarely found in Coats disease [14]. The calcifications are mainly found at the level of the RPE.

Histopathology (Fig. 4) [15], [16], [17] shows three characteristic changes of the disease:
• Vascular anomalies, telangiectasias, with an alteration of the endothelial coating and a thickening of the basement membrane (disappearance of endothelial cells and pericytes or endothelial proliferation), leading to a rupture of the hematoretinal barrier;
• Diffuse Periodic Acid Schiff (PAS) positive exudates in and under the outer layers of the retina, altering the architecture of bipolar and sensory cells;
• Cholesterol crystals and histiocytes with foamy cytoplasm (macrophages), full of cholesterol, ghost cells, mainly in the subretinal exudate.
This microscopic evaluation ensures an unambiguous diagnosis, revealing these very characteristic histopathological signs of Coats disease [18], while Flexner-Winter Steiner rosettes, Homer Wright rosettes, necrosis and hyperdense, round to spindle-shaped, pathognomonic histologic signs of retinoblastoma were not present.

IV. Conclusion

Coats disease is a serious disease of unknown etiology, involving a functional visual prognosis.
General pathologists usually do not encounter this disease because it is diagnosed at an early stage by clinical and radiological examination, which helps save the globe and useful vision.
The differentiation between retinoblastoma and Coats disease can be difficult. Coats disease, on the other hand, remains a diagnostic trap, especially during the advanced stages of the disease. Enucleation appears to be indicated in uncertain cases due to the reduced visual prognosis and the risk of secondary complications in advanced Coats disease.
Therefore, it is essential for the pathologist to be familiar with the histopathological features of retinoblastoma and to distinguish it from Coat's disease.

References

[1]. Coats G: Forms- of retinal disease with massive exudation. R Lond Ophthalmol Hosp
[2]. B. Förl • I. Schmack • H.E. Grossniklaus • K. Rohrschneider Universitäts-Augenklinik Heidelberg Morbus Coats , Wichtige Differenzialdiagnose zum Retinoblastom.
[3]. Vahedi, L. Lumbroso-Le Rouic, C. Levy Gabriel, F. Doz, I. Aerts, H. Brisse, O. Berges, M.T. Iba Zizen, L. Desjardins Diagnostic différentiel du rétinoblastome : étude rétrospective de 486 cas
[4]. Shields JA, Shields CL, Parsons HM (1991) Review: Differential diagnosis of retinoblastoma. Retina 11: 232–243
[5]. Shields JA, Shields CL (2001) Differentiation of Coats‘ disease and retinoblastoma. J Pediatr Oph¬thalmol Strabismus 38: 262–266
[6]. Shields JA, Shields CL, Honavar SG et al. (2001) Classification and management of coats disease: the 2000 Protocor Lecture. Am J Ophthalmol 131: 572–583
[7]. Shields JA, Shields CL, Honavar SG, Demirci H. Clinical variations andcomplications of Coats disease in 150 cases: the 2000 Sanford Gifford Memorial Lecture. Am J Ophthalmol. 2001;131:561-571.
[8]. Al-Qahtani AA, Almasaud JM, Ghazi NG. Clinical Characteristics and Treatment Outcomes of Coats Disease in a Saudi Arabian Population.Retina. 2015;5:2091-2099
[9]. Meier P, Sterker I, Tegetmeyer H (2006) Leukokorie im Kindesalter. Klin Monatsbl Augenheilkd 223: 521–527
[10]. Smithen LM, Brown GC, Brucker AJ, Yannuzzi LA, Klais CM, Spaide RF.
[11]. Coats' disease diagnosed in adulthood. Ophthalmology. 2005;112:1072-1078.
[12]. Shields JA, Shields CL. Review: Coat's disease: The 2001 Luesther T. Mertz lecture. Retina 2002;22:80-91.
[13]. Senft SH, Hidayat AA, Cavender JC (1994) Atypical presentation of coats disease. Retina 14: 36–38

[14]. Shields JA, Shields CL. Review: Coat's disease: The 2001 Luesther T. Mertz lecture. Retina 2002;22:80-91.
[15]. Tripathi R, Ashton N. Electron microscopical study of Coat's disease. Br J Ophthalmol 1971;55:289–301.
[16]. Apple DJ, Naumann GO. Spezielle Pathologie der Retina. In: Naumann GO, editor. Pathologie des Auges. Berlin: Springer-Verlag; 1980. p. 577–666.
[17]. Tarkkanen A, Laatikainen L. Coat's disease: clinical, angiographic, histopathological findings and clinical management. Br J Ophthalmol 1983;67:766.
[18]. Yanoff M, Fine BS (2002) Retinoblastoma and Pseudoglioma. In: Yanoff M, Fine BS (eds) Ocular Pathology. 5th edn. Mosby, Inc, St. Louis, MO, USA, pp 704–709, 717–719

Ibtissam Razzouki, et. al. "A puzzling diagnosis mimicking retinoblastoma: Coats disease: Case report.'' *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*, 20(08), 2021, pp. 43-48.