Journal of Neonatal Surgery
ISSN(Online): 2226-0439
Vol. 14, Issue 7s (2025)
https://www.jneonatalsurg.com

OPEN ACCESS

Coats' Disease Masquerading as Retinoblastoma: Diagnostic Dilemmas

Septian Dwi Prabowo[1,2], Delfitri Lutfi[1,2], Dyah Fauziah[3,4]

[1]Department of Ophthalmology, Dr. Soetomo General Academic Hospital, Surabaya, Indonesia

[2]Department of Ophthalmology, Faculty of Medicine - UNIVERSITAS AIRLANGGA, Surabaya, Indonesia

[3]Department of Anatomical Pathology, Dr. Soetomo General Academic Hospital, Surabaya, Indonesia

[4]Department of Anatomical Pathology, Faculty of Medicine - UNIVERSITAS AIRLANGGA, Surabaya, Indonesia

***Corresponding Author**

Delfitri Lutfi, Department of Ophthalmology, Dr. Soetomo General Academic Hospital, Surabaya, Indonesia and Department of Ophthalmology, Faculty of Medicine - UNIVERSITAS AIRLANGGA, Surabaya, Indonesia

Email ID: delfitriua@gmail.com

Cite this paper as: Septian Dwi Prabowo, Delfitri Lutfi, Dyah Fauziah, (2025) Coats' Disease Masquerading as Retinoblastoma: Diagnostic Dilemmas. *Journal of Neonatal Surgery*, 14 (7s), 604-608.

ABSTRACT

Coats' disease is one of the most difficult differential diagnoses of intraocular mass especially retinoblastoma in early childhood. Despite comprehensive diagnostics, retinoblastoma remains possible, and enucleation appears necessary in uncertain cases with poor visual prognosis. This case, reports a 3-year-old boy with leukocoria, recurrent redness, and strabismus in the right eye for 1 year. Due to the results of ultrasonography and CT-scan examination, a retinoblastoma was suspected, so enucleation with pathology examination was performed. The findings in pathology examination were consistent with Coats' disease and negative for retinoblastoma. Although it is difficult to distinguish between retinoblastoma and coats' disease, it is necessary to initiate a comprehensive examination of clinical characteristics and ancillary test to rule out retinoblastoma, which needs to be distinguished from a variety of benign mimicking lesions to ensure the appropriate therapy. A pathologist's confirmation and discussion are required for further appropriate treatment due to the unexpected pathology result.

Keywords: *Coats disease, retinoblastoma, enucleation*

1. INTRODUCTION

Leukocoria in children poses complex diagnostic challenges. Congenital cataract, retinopathy of prematurity, coats' disease, retinoblastoma, hyperplastic primary vitreous, ocular toxocariasis, Norrie's disease, and organized intraocular hemorrhage are just a few of the various diagnoses that could be made. Retinoblastoma is the most frequent cause of leukocoria, which accounts for 60% of cases and is the biggest obstacle in ophthalmology. This highly aggressive retinal cancer typically affects young infants usually noted before 4 years of age. In a study of 500 consecutive patients, 42% of patients referred for potential retinoblastoma were found to have a mimicking condition after further examination [1,2]. Advanced coats' disease is the most common lesion mimicking retinoblastoma, commonly unilateral and usually noted in the first decade of life. However, 80% of primary ocular tumors in children under the age of 15 are retinoblastoma, 1% of all cancer-related deaths, and 17% of all newborn cancers. Although it can be difficult to distinguish between retinoblastoma and pseudo-retinoblastoma, it is necessary to initiate adequate management and comprehensive examination [3]. In the past 15 years, the clinical or pathological reliability of diagnosing retinoblastoma with 7-40% of patients who underwent enucleation had a misdiagnosis according to pathological examination. A study of Shield in 2006 reported a 42% misdiagnosis rate in 1991. In 1988, there was a 30% and 16% prevalence of clinical misdiagnosis [1,4]. A delayed diagnosis of retinoblastoma can have fatal consequences, while a false-positive retinoblastoma can result in chemotherapy and other unnecessary treatments. Given the repercussions of a misdiagnosis, the accuracy of the diagnosis of retinoblastoma has long been a source of concern. It is crucial to note that enucleation is still a valid option in retinoblastoma and pseudo-retinoblastoma [4,5].

2. PATIENT AND OBSERVATION

Patient Information: A 3-year-old boy came with his parent complaining of a white appearance in the right eye for 1 year. The parent also complained of recurrent redness and strabismus in the right eye. Prior examination was performed under anesthesia and revealed a bulging retina to the anterior segment, yet orbital imaging was not performed. The patient had a history of febrile seizures at 2 years old. The patient was not born prematurely and had no prior history of oxygen supplementation or trauma.

Clinical Findings: Visual acuity of the right eye was fixed and follow negative and the left eye was fixed and follow positive. The anterior segment examination revealed hyperemia conjunctiva, leukocoria in the right eye, and normal in the left eye (figure 1). The initial diagnosis was intraocular mass and planned for ultrasound and CT-scan examination.

FIGURE 1. Anterior segment examination revealed right eye hyperemic conjunctiva and leukocoria

Diagnostic assessment: B scan ultrasound showed echo-dense material in the right eye revealing an echogenic lesion in the form of a mass that fills >80% of the vitreous cavity with calcification. The A-scan ultrasound examination showed many spikes within the eye cavity, consistent with the vitreous, echo-spike of 50-100% RCS complex (figure 2). CT scan examination revealed an increased density of the right eye with 28HU vitreous humor oculi accompanied by retinal thickening and calcification within 105 HU, and unremarkable right bulbus oculi. These findings favored retinoblastoma (figure 3).

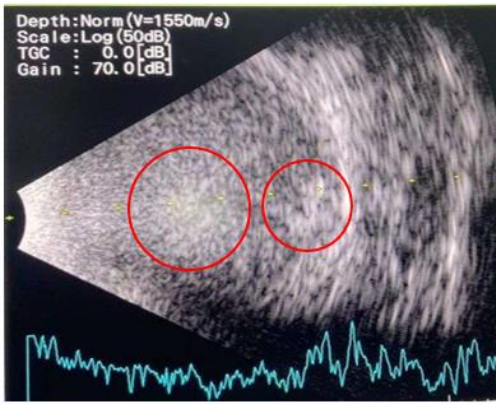

FIGURE 2. Ultrasound examination on the right eye showed an echogenic lesion typically a mass form, with 50%-100% echospike RCS complex, with 80% of calcification filling the vitreous cavity (red circles)

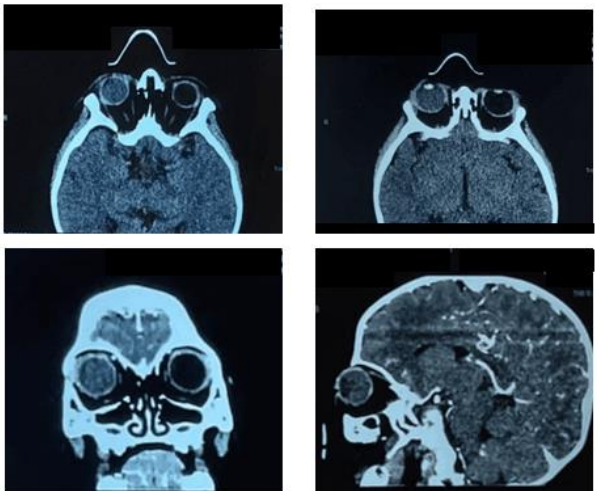

FIGURE 3. CT scan examination shows increased density of the right vitreous humor oculi (28 HU) accompanied by retinal thickening and calcification (105 HU), highly suspected retinoblastoma

Diagnosis: Retinoblastoma in the right eye was highly suspected, myoconjunctival enucleation technique was performed, followed by pathology examination.

Therapeutic interventions: Myoconjunctival enucleation was performed (figure 4). The globe transillumination test revealed a mass filling almost the entire vitreous cavity, the emissary vein was dilated, and an extra scleral mass was not found (figure 5). Left eye fundoscopy examination was within normal limits. An orbital implant and ocular prosthesis were inserted in the socket, tarsorrhaphy, and a pressure patch was maintained for one day. The enucleation specimen was sent to pathology for examination. The macroscopic examination revealed that the size of the eye globe was 2.1 cm in the largest diameter. Present retinal detachment and a yellowish jelly-like substance filled the subretinal area, but no tumor was found. The microscopic examination revealed eosinophilic amorphous material (exudate) mixed with scattered macrophages, and gliotic retinal tissue with some dilated blood vessels. There were no signs of malignancy. These pathology findings were in keeping with Coats' disease (figure 6).

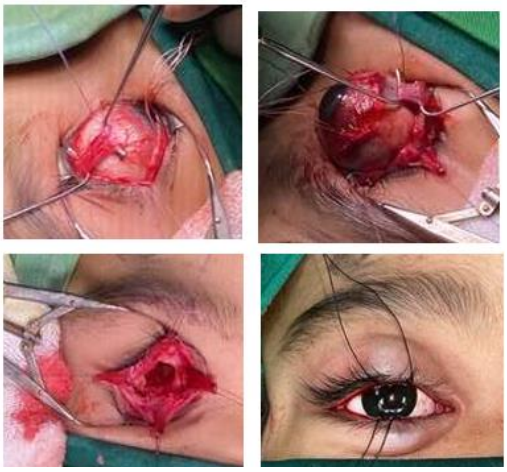

FIGURE 4. Myoconjunctival enucleation

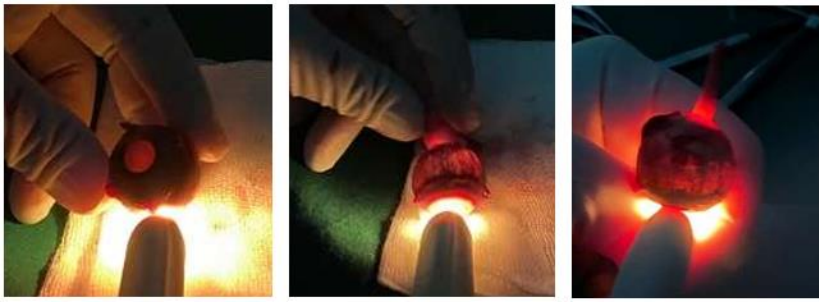

FIGURE 5. Transillumination examination during the operation revealed mass form fills the entire vitreous cavity

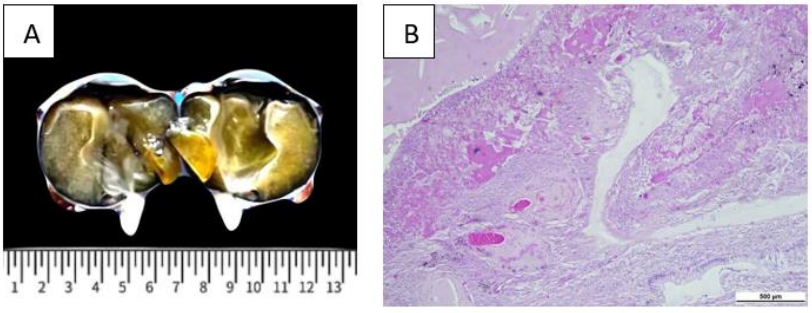

FIGURE 6. Pathology examination revealed retinal detachment and a yellowish jelly-like substance filling the subretinal area (A). The microscopic examination revealed eosinophilic amorphous material (exudate) mixed with scattered macrophages, and gliotic retinal tissue with some dilated blood vessels (B).

Follow-up and outcome of interventions: After removing tarsorrhaphy, the socket was fine-defined. In the third month, it was good and well-formed, and the motility of the ocular prosthesis was fine.

3. DISCUSSION

Leukocoria in children poses complex diagnostic challenges. Identifying and differentiating these pseudo-retinoblastomas from retinoblastoma is crucial since their care differs significantly [3,5]. In this case, the patient's mother complained about the leukocoria of the right eye and was afraid of retinoblastoma. After ultrasound and CT scan examination revealed retinoblastoma, myoconjunctival enucleation was performed, yet pathology examination revealed Coats' disease.

Nearly half of all newborns who appear with a white pupil have leukocoria, which is the most common symptom (occurring 60% of the time) and is connected to retinoblastoma [5]. However, some benign disorders, known as pseudo-retinoblastoma, resemble retinoblastoma in their clinical and radiological presentations. Almost 16-22% of patients referred due to retinoblastoma suspicion have mimicking lesions. The two disorders that most frequently mimic retinoblastoma are Coats' disease (40%) and persistent fetal vasculature (26%), which often occur in young children with leukocoria as the first symptom [4,5].

A child with leukocoria needs to be evaluated systematically. The primary purpose is to rule out retinoblastoma. Retinoblastoma needs to be distinguished from a variety of benign mimicking lesions to ensure the appropriate therapy. The diagnosis needs to be made as soon as possible for maximum ocular salvage. Imaging is advised to identify extraocular extension, optic nerve infiltration, choroidal infiltration, cerebral metastases, or a secondary tumor, even if the diagnosis is made with an ophthalmoscope which is frequently reliable. It also aids in distinguishing from other lesions that mimic others [6].

A leukocoria may be due to exudates, but if there is retinal detachment, this will appear greyer. Clinical traits include three main features of massive yellowish exudates within and beneath an edematous retina, sometimes giving a pseudo-tumor appearance, and exudative, occasionally total retinal detachment [6,7]. Retinoblastoma is almost always associated with vitreous and subretinal seeding as well as calcifications. However, the diffuse infiltrating form may eventually reveal as a pseudo-hypopyon and leukocoria, appearing as a greyish retinal coating without increased mass or calcifications [5]. Distinguishing coats' disease from retinoblastoma is crucial, and imaging can be beneficial [4,5].

Imaging is frequently effective in diagnosing intraocular tumors. Particularly, in very young patients, ultrasound examination, CT scan, and MRI are useful for ruling out additional potentially lethal conditions such as retinoblastoma. However, because calcifications can be present in both situations, it can be challenging to distinguish rely on imaging. Intraocular mass and calcification, signs of exophytic retinoblastoma, can be found with ocular ultrasound examination [7].

In this case, CT-scan examination revealed an increasing density of the right eye in vitreous humor oculi accompanied by retinal thickening and calcification. Due to the intraocular calcification, CT-scan is a beneficial diagnostic imaging, calcification can be clearly seen in more than 90% of advanced retinoblastoma with a high resolution of thin section. Retinoblastoma appears as a hyperdense mass due to its hypercellularity compared with the vitreous body. The sensitivity of CT-scan in identifying calcifications in retinoblastoma ranges from 81% to 96% with even higher specificity [5]. It is challenging to distinguish between Coats' disease and unilateral non-calcifying retinoblastoma on a CT scan. The latter is helpful in the late stages of Coats' disease because it can indicate retinal detachment as greater attenuation in the eye globe. Although calcification is not a characteristic of Coats' disease, a sub-macular calcified nodule may be noted in as many as 1/5 of advanced Coats' disease. These show the retinal pigment epithelium's exuberant proliferation [4,6]. In advanced stages, the subretinal exudate is detected and is hyper-dense on CT-scan due to the high protein concentration [5].

Retinoblastoma and Coats' disease, which manifests as significant subretinal exudation from leaking telangiectatic retinal blood vessels, are challenging to distinguish by ultrasonography. In this case, ultrasonography showed an echo-dense material mass filling >80% of the vitreous cavity with calcification. Given that Coats' disease primarily affects young men, this led to incorrect diagnosis. However, the highly reflective lesion in mass could be interpreted as a calcification which is featured as retinoblastoma [6,8].

Retinoblastoma has changed from a fatal childhood cancer to a largely curable cancer over the past 40 years. The goal of current management is to save the eye and maintain vision. At present, laser treatment, cryotherapy, and enucleation are among the therapeutic options. The treatment options are selected for each individual's instance such as the risk of metastatic disease, second cancers, systemic condition, laterality of the disease, size and location of the tumor, and estimated visual prognosis. Enucleation is the preferred treatment for advanced unilateral retinoblastoma or the worse eye in bilateral cases. The stage of Coats' disease determines the visual result after treatment. Although the general visual prognosis is poor, the prognosis varies according to the stage of the disease at which the patient appears. Even with treatment, Coats' disease generally progresses, and when it reaches an advanced stage, the affected eye typically becomes blind. With more advanced

stages indicating the necessity for primary enucleation and worse final visual acuity, the Shields categorization system helps direct treatment and forecast ocular and visual outcomes [6,9]. The main indication for enucleation in Coats' disease is poor visual prognosis, painful eye due to secondary glaucoma, and pathological diagnosis due to highly suspected retinoblastoma [9,10].

There is a lack of studies that provide a detailed discussion of histological features of Coats' disease. Some pathological insight has been gained from the few occurrences of enucleation in Coats disease. In this case, microscopic examination showed eosinophilic amorphous material (exudate) mixed with scattered macrophages, and gliotic retinal tissue with some dilated blood vessels. Variable retinal detachment, often bullous extending to the lens, is the most common symptom of Coats' disease. Advanced cases may include hemorrhage, retinal detachment, anterior chamber angle obstruction, and lens iris diaphragm displacement. The subretinal space is filled with thick and yellow lipoprotein exudates, sometimes by yellow material clumps. The histopathological findings of the retina in the enucleated Coats' disease included bullous total retinal detachment with deep and subretinal exudates, massive gliosis, retinal disorganization, fresh and or old hemorrhages, ghost cells, and cholesterol crystals [9,10].

4. CONCLUSION

Although it's challenging to distinguish retinoblastoma and advanced coats disease, it is necessary to initiate a comprehensive examination of clinical characteristics and ancillary test to rule out retinoblastoma, which needs to be distinguished from a variety of benign mimicking lesions to ensure the appropriate therapy. A pathologist's confirmation and discussion are required for further appropriate treatment due to the unexpected examination.

REFERENCES

[1] Shields CL, Yusuf U, Raquel B, Ralph CE, Barry M, Jerry AS. Retinoblastoma in an Eye with Features of Coats' Disease. Journal of Pediatric Ophthalmology and Strabismus. 2006;43:313-315.

[2] Mandura EA, Abdullah SA. Coats' Disease Diagnosed During Adulthood. Cureus. 2021;13(7):e16303.

[3] Jansen RW, Christiaan M, Hervé JB, Paolo G, Liesbeth C, Sophia G, Philippe M, Nathalie C, Arnaud G, Sabrina S, Theodora H, Francis LM, Jonas AC, Paul V, Annétte CM, Marcus C, Pim G. MR Imaging Features to Differentiate Retinoblastoma from Coats' Disease and Persistent Fetal Vasculature. Cancers. 2020;12:3592.

[4] Ghassemi F, Fatemeh B, Ali M. Lesions Simulating Retinoblastoma at a Tertiary Care Center. Journal of Ophthalmic and Vision Research. 2015;10(3):316-319Chaurasia S, Ramappa M, Murthy SI, Vemuganti GK, Fernandes M, Sharma S, et al. Chronic conjunctivitis due to Mycobacterium tuberculosis. Int Ophthalmol. 2015;34(3):655–660.

[5] Sanchez N, Kranz P, Constenia D. Leukocoria in Children: Findings on CT and MRI of the Principal Causes. Neurographics. 2022;12(4):192-202.

[6] Sen M, Carol LS, Santosh GH, Jerry AS. Coats Disease: An Overview of Classification, Management, and Outcomes. Indian Journal of Ophthalmology. 2019 Vol 67(6);763-771.

[7] Sigler EJ, John CR, Jorge IC, Matther WW, Barrett GH. Current Management of Coats' Disease. Survey of Ophthalmology. 2014(59)30-64.

[8] Zaini LM, Siti H, Annisa MB. Coats' Disease in Children: A Review of Clinical Manifestation, Diagnosis and Management. International Journal of Clinical Pediatrics and Child Health. 2020;2(1):17-23.

[9] Helmi HA, Abdullah J, Azza MM, Saleh A, Hind MA. Clinicopathological Correlations in Enucleated Globes of Late-Stage Coats' Disease with A Review of the Literature. Journal of Epidemiology and Global Health. 2022(12):496-503.

[10] Asari WA, Supriya R, Nirali PS. A Prospective Study of Myoconjunctival Enucleation for Enhanced Implant Motility. International Journal of Research in Medical Sciences. 2021;9(12):3547-3552.