Coats disease masquerading as retinoblastoma at Kenyatta National Hospital – a case series

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Abstract

Aim: To describe 5 cases of coats disease that were referred as retinoblastoma to Kenyatta National Hospital

Study design: A retrospective case series

Study location and period: Ophthalmology Eye Unit at Kenyatta National Hospital between June 2023 and September 2024.

Methods: The files for patients who had been referred to Kenyatta National Hospital as retinoblastoma and turned out to be cases of coats disease were reviewed. Data collected included age, sex, presenting visual acuity, findings on fundus examination and other ancillary tests.

Results: The five cases presented were referred to Kenyatta national hospital with unilateral leukocoria and a tentative diagnosis of unilateral retinoblastoma. There were four males and one female and all the children were less than three years of age. All the children had profound visual loss of the affected eye at presentation. The right eye was affected in three children and left eye in two children. Even though all of the patients had an initial diagnosis of retinoblastoma at presentation, further work up at Kenyatta National hospital ruled out retinoblastoma in four of them. However, the clinical diagnosis was not clear-cut for one patient and the diagnosis of retinoblastoma was upheld and enucleation of the affected eye done. The histology of the enucleated eye however showed features of coats disease.

Conclusion: Coats disease is a rare ocular condition and it is sometimes difficult to differentiate from retinoblastoma clinically. Proper work up however can pick most cases and unnecessary enucleations can be avoided. Sometimes the diagnosis can only be confirmed histologically after enucleation.

Key words: leukocoria, retinoblastoma, coats disease

Introduction

Coats' disease is a rare, idiopathic ocular condition resulting from abnormal development of the retinal vasculature, leading to telangiectasia, exudation, and potential retinal detachment [1]. It predominantly affects young males and is usually unilateral. The disease can present at various stages, ranging from mild vascular abnormalities to severe complications such as total retinal detachment, neovascular glaucoma, and phthisis bulbi, which can lead to irreversible vision loss and, in extreme cases, necessitate enucleation. [2,3] Despite its rarity, Coats' disease is a significant cause of childhood visual impairment, necessitating early diagnosis and intervention.

Coats' disease was first described in 1908 by George Coats as a unilateral vascular disease affecting the retina of young males.[4] The clinical appearance of Coats' disease is highly variable, depending on the severity at presentation. One

of the key clinical manifestations is leukocoria (a white pupillary reflex), which is also a hallmark of other pediatric ocular conditions, including cataract, retinoblastoma, and persistent hyperplastic primary vitreous (PHPV) [5]. The presence of leukocoria often leads to a misdiagnosis, and children with Coats' disease are frequently referred for suspicion of retinoblastoma, making accurate differentiation crucial for proper management. While Coats' disease is non-neoplastic, retinoblastoma is the most common intraocular malignancy in children, requiring vastly different treatment approaches.

Differentiating Coats' Disease from Retinoblastoma

Distinguishing Coats' disease from retinoblastoma is critical to avoid unnecessary enucleation or chemotherapy. One of the most important differentiating features is the presence of intraocular calcifications in retinoblastoma, which are typically absent in Coats' disease. Additionally, Coats' disease is characterized by extensive subretinal exudation and retinal telangiectasia, whereas retinoblastoma presents as a solid intraocular mass with potential endophytic or exophytic growth patterns[6,7]. Imaging techniques such as B-scan ultrasonography, optical coherence tomography (OCT), and fluorescein angiography (FA) are essential in distinguishing between these two conditions.

The table below summarizes the similarities and differences between Coats' disease and retinoblastoma based on clinical and imaging findings.

Table 1: Similarities and differences between Coats' disease and retinoblastoma

Feature	Coats' Disease	Retinoblastoma
Laterality	Unilateral	Unilateral or Bilateral
Gender Predilection	Males > Females	No Gender Preference
Age at Diagnosis	Childhood, often 1–10 years	Infancy to early childhood (median <3 years)
Leukocoria	Present	Present
Strabismus	May be present	May be present
Retinal Findings	Retinal telangiectasia, exudation, detachment	Tumor mass, exophytic/endophytic growth
Intraocular Calcifications	Absent	Present (95%)
B-Scan Ultrasound	Retinal detachment with subretinal exudates, no calcifications	High internal reflectivity, intraocular calcifications
Fluorescein Angiography	Retinal vascular leakage, aneurysms	Tumor staining, early filling defects
OCT Findings	Intraretinal cysts, exudation	Mass lesion with shadowing
Management	Laser photocoagulation, cryotherapy, anti-VEGF therapy	Chemotherapy, enucleation (if advanced)

Treatment for Coats' disease in the past primarily involved ablative lasers, but in the last decade, the use of anti-vascular endothelial growth factors (anti-VEGF) has emerged as a promising adjunctive therapy.[8,9] Anti-VEGF agents help to reduce vascular permeability and exudation, thereby stabilizing the disease and preserving vision in less advanced cases. Combination therapy with laser photocoagulation and cryotherapy has also shown positive outcomes in preventing disease progression. Early diagnosis and intervention remain key in managing Coats' disease effectively.

To the best of our knowledge, Coats' disease has not been reported before in a Kenyan child. Our aim in this case series is to report five cases of Coats' disease diagnosed in Kenyan children, highlighting their clinical presentations, imaging findings, and management strategies. By presenting these cases, we aim to contribute to the global understanding of Coats' disease and emphasize the importance of considering this condition in the differential diagnosis of childhood leukocoria.

Case reports

Patient 1

A 10-month-old male was referred to the pediatric ophthalmology clinic from a peripheral facility with a diagnosis of retinoblastoma in the right eye. The child had a history of a white reflex in the right eye since birth, associated with poor vision in the same eye. Otherwise, the child was healthy, with no complaints of any systemic diseases.

At presentation, the visual acuity of the right eye was not fixing and following light, while the left eye was fixing and following light. The intraocular pressure of the right eye

was 68 mmHg, whereas it was within normal limits in the left eye. A relative afferent pupillary defect (RAPD) was present in the right eye.

Further examination of the right eye revealed leukocoria, and fundoscopy showed a yellow-whitish mass filling the vitreous cavity. An ocular ultrasound of the right eye was performed, revealing a hyperechoic mass filling the vitreous cavity with an attached retina. Based on these findings, an impression of right eye retinoblastoma was made.

The patient was admitted, and an examination under anesthesia (EUA) was performed for both eyes. The right eye was enucleated, and intraoperatively, the right optic nerve was subjectively noted to be thickened.

Due to the subjective thickening of the optic nerve, the child was started on intravenous high-dose vincristine, etoposide, and carboplatin (HDVEC) while awaiting histology results.

Histology later revealed a detached retina with telangiectatic vascular channels associated with eosinophils and exudates containing foamy histiocytes. No evidence of malignancy was found, and the final diagnosis was Coats' disease.

Chemotherapy was subsequently stopped, and the child is now under follow-up in the eye clinic.

Diagnosis: Features of coats disease

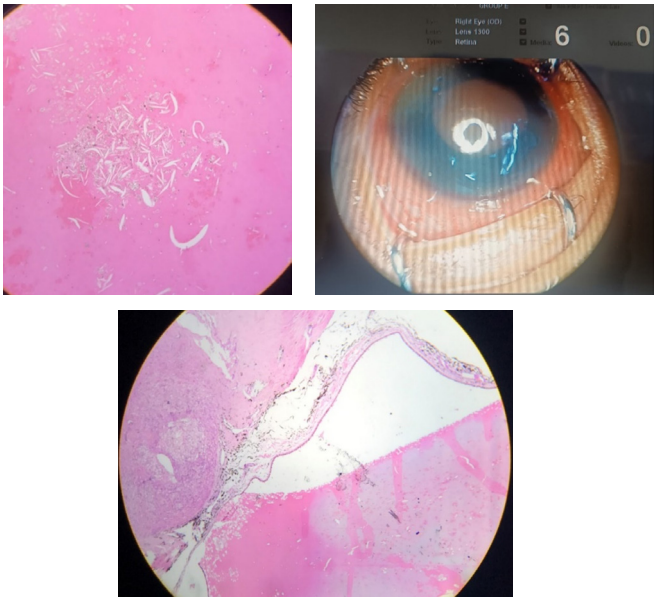


Figure 1: Histopathology indicated subretinal fluid with cholesterol clefts and lipid-laden macrophages and retinal detachment. Clinical photography, showed Conjunctival hyperemia and leukocoria. © Dr. Weheliye Abdinoor

Patient 2

A child aged 2 years and 10 months presented to the pediatric ophthalmology clinic with a complaint of a white pupillary reflex in the left eye for the past three months.

In his past medical history, the child had been treated for bilateral undescended testes, and orchidopexy had been performed at the age of 2 years and 7 months. Additionally, he had a history of recurrent chest infections.

Examination Findings

Head and Neck: The child had sparse hair, brachycephaly, low-set ears, midface hypoplasia, and ear lidding.

Central Nervous System (CNS): The child was alert, and there was a relative afferent pupillary defect (RAPD) in the left eye.

Cardiovascular Examination: A pansystolic murmur was heard at the apex.

Abdominal and Perineal: The abdomen was normal, and orchidopexy scars were noted on the perineum.
The rest of the systems essentially normal.

Ocular Examination:

- Visual acuity at presentation:
  - Right eye: 3/3
  - Left eye: No perception of light (NPL)
- Intraocular pressure: Within normal limits in both eyes.
- Fundoscopy:
  - Left eye: A yellowish mass in the vitreous with retinal detachment.
  - Right eye: Normal findings

Imaging and Investigations

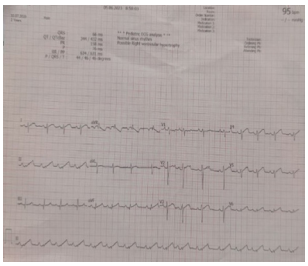
Several imaging studies were conducted, including a 2D echocardiogram, ECG, and ocular ultrasound.

Cardiac Investigations:

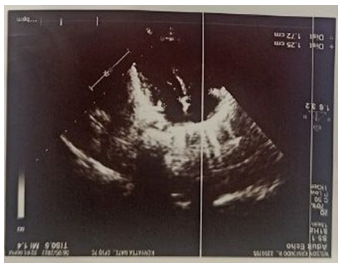
- 2D Echocardiogram:
  - Mild right atrium and right ventricle dilation.
  - Supravalvular stenosis of the pulmonary artery.
  - Mild pulmonary stenosis.
  - Pressure gradient: 30 mmHg.
- ECG:
  - Normal sinus rhythm.
  - Possible right ventricular hypertrophy

Ocular Imaging:

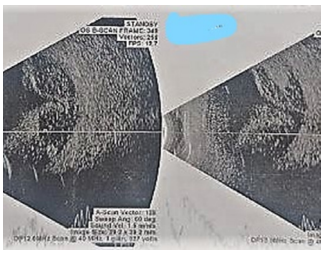
- Ocular ultrasound findings:
  - Right eye: Normal, with an echolucent vitreous and no retinal detachment.
  - Left eye: Funnel-shaped retinal detachment
- Widefield imaging (RetCam):
  - Left eye: Diffuse exudation and a convex-shaped retinal detachment



ECG



2D Echocardiogram



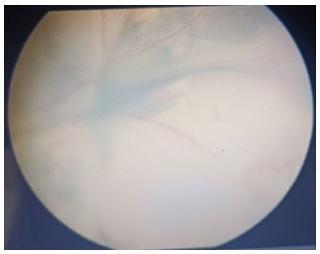


Figure 2: Ocular ultrasound showed a funnel shaped retinal detachment. Widefield imaging, diffuse exudation and a convex-shaped retinal detachment © Dr. Arthur Korir

An impression of syndromic child with stage 3 coats disease was made. The child was therefore referred to cardiology clinic and the child is on follow up in the eye clinic.

Patient 3

A 3-year-old girl was referred to our pediatric ophthalmology clinic with a diagnosis of right eye retinoblastoma for examination under anesthesia (EUA) and further management. She presented with a 9-month history of outward deviation of the right eye, associated with a white pupillary reflex.

Her past ocular and medical history were unremarkable, and systemic examination was also unremarkable.

Ophthalmic Examination

- Visual Acuity:
  - Right eye: Unable to fixate or follow light.
  - The right eye could pick 100/1000 objects at 33 cm.
- Extraocular Motility: Free in both eyes.
- Hirschberg Test: 7.5-degree exotropia in the right eye.
- Fundoscopy:
  - Right eye: A whitish, vascularized, retrolental mass inferiorly.
  - Left eye: Normal anterior and posterior segments

Imaging and Investigations

- Ocular Ultrasound (Right Eye):
  - Hyperechoic mass filling the vitreous, attached to the retina with moderate spikes.
  - No calcification was noted.

Diagnosis and Management

The child was admitted with a provisional diagnosis of right eye retinoblastoma. Examination under anesthesia (EUA) was performed for both eyes.

- Widefield imaging under anesthesia (Right Eye):
  - Superonasal retinal detachment with diffuse exudation and telangiectasia
  - No retinoblastoma masses were identified

A final diagnosis of right eye Stage 3 Coats' disease was made. The child is currently on follow-up in the eye clinic.

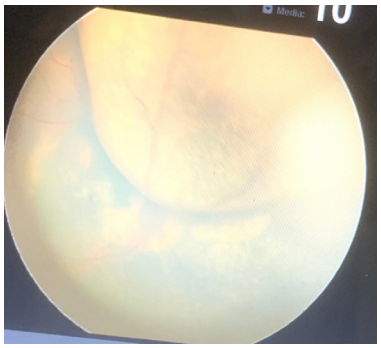


Figure 3: Widefield imaging showed superonasal retinal detachment with diffuse exudation and telangiectasia © Dr. Weheliye Abdinoor

Patient 4

An 18-month-old boy was referred to our pediatric ophthalmology clinic with a diagnosis of right eye retinoblastoma for examination under anesthesia (EUA) and further management. He presented with a 3-month history of a white reflex in the right eye. There was no reported discharge, proptosis, or ocular trauma. His birth and past medical history were unremarkable.

Ophthalmic Examination

- Visual Acuity:
  - Right eye: Not fixating or following light.
  - Left eye: Able to pick 100s/1000s at 33cm
- Extraocular Motility: Free in both eyes.
- Fundoscopy (Right Eye):
  - A mass filling the vitreous cavity.

Imaging and Investigations

- B-Scan Ultrasound (Right Eye):
  - Hyperechoic mass filling the vitreous, attached to the retina with moderate to high spikes
  - Findings were highly suspicious of retinoblastoma

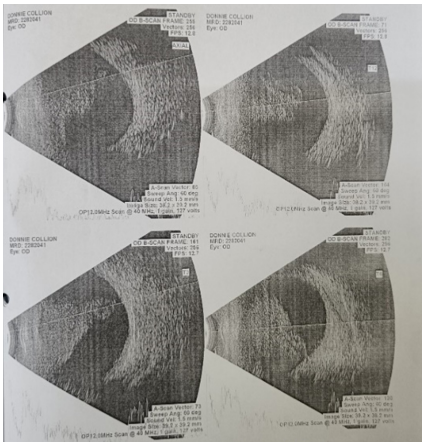
Diagnosis and Management

The child was admitted with a provisional diagnosis of right eye retinoblastoma. Examination under anesthesia (EUA) was performed for both eyes.

- EUA Findings (Right Eye):
  - Funnel-shaped total retinal detachment with

diffuse telangiectasia.
- No retinoblastoma mass was identified.

A final diagnosis of right eye Coats' disease was made. The child was discharged home and scheduled for follow-up in the eye clinic.



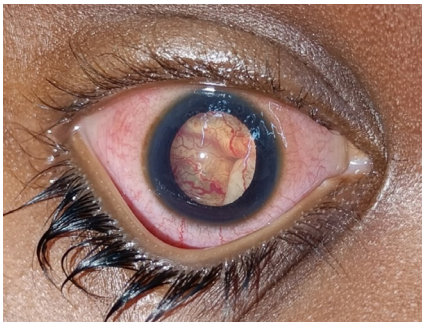


Figure 4: Right Eye EUA showed a funnel-shaped total retinal detachment with diffuse telangiectasia. B-Scan showed, hyperechoic mass filling the vitreous, attached to the retina with moderate to high spikes © Dr. Pranav Chadha

Patient 5

An 8-month-old male was referred to the pediatric eye clinic from a peripheral facility with a diagnosis of right eye retinoblastoma. The child had a history of a white reflex in the right eye, first noted at 5 months of age. He also had an outward deviation of the right eye. His past ocular and systemic history were unremarkable, as were his birth and developmental history.

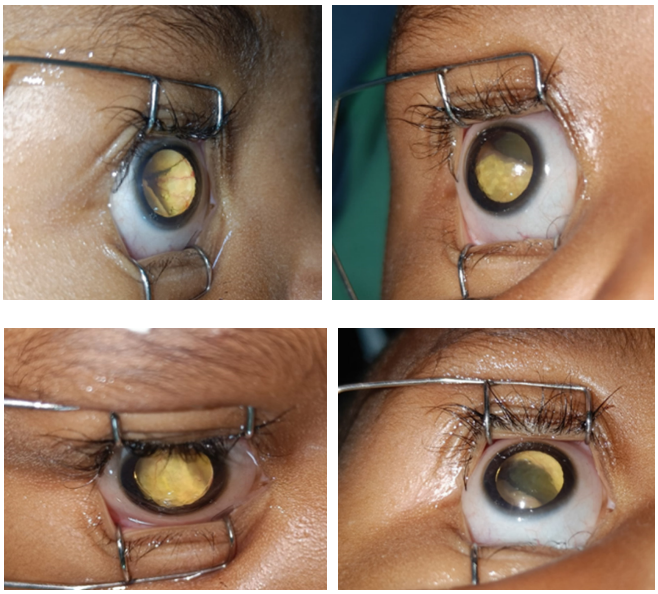
Ophthalmic Examination

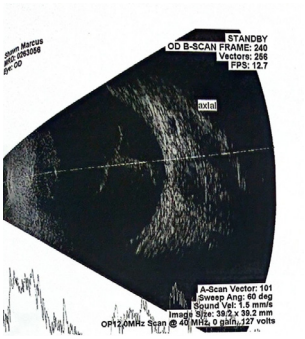
- Visual Acuity:
  - Right eye: Defensive reaction.
  - Left eye: Fixing and following small objects of 3 cm.
- Intraocular Pressure: Normal in both eyes.
- Ocular Alignment: Right eye exotropia.
- Anterior Segment Examination:
  - Relative afferent pupillary defect (RAPD) in the right eye.
  - Leukocoria present.
- Fundoscopy (Right Eye):
  - Total funnel-shaped retinal detachment.
  - Overlying telangiectatic vessels and retinal macrocysts.
- Ocular Ultrasound:
  - Confirmed funnel-shaped retinal detachment.
  - Presence of subretinal fluid.

Diagnosis and Management

A diagnosis of right eye Coats' disease was made. The child was admitted for external drainage of subretinal fluid and cryotherapy. The procedure was successfully performed, and the subretinal fluid was drained.

However, there was no improvement in vision, and the child remains on follow-up in the eye clinic.





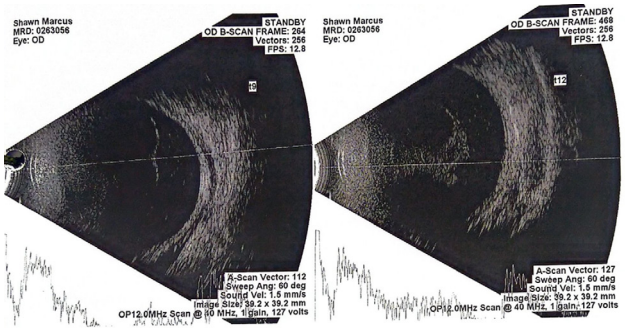


Figure 5: Right eye total exudative retinal detachment that is funnel shaped with retinal macrocysts © Dr. Pranav Chadha

Right eye ocular ultrasound showing funnel shaped retinal detachment, high reflectivity spikes. Other transverse cuts showing retinal detachment with subretinal fluid and some dot echoes (possibly lipid crystals)

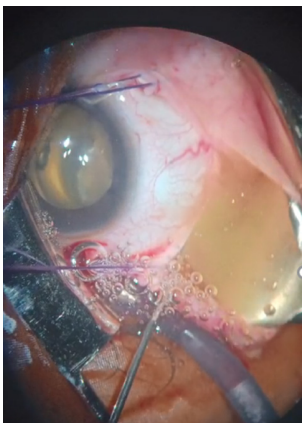


Figure 6: Intra operative external drainage of subretinal fluid from a scleral incision. The subretinal fluid is viscous, straw colored with glistening cholesterol crystals as typically seen in coats disease © Dr. Pranav Chadha

Ethical considerations

Informed consent was taken before the photos to be taken. The names of the patients remained anonymous, and their faces were concealed.

Discussion

Described by George Coats in 1908, Coats' disease is an idiopathic telangiectatic neovascular disease of the retina that is mostly unilateral. It is characterized by abnormal retinal vessel development, leading to leakage of fluid and exudation, ultimately causing retinal detachment. Coats' disease has been associated with certain syndromes, such as Turner syndrome. However, it is typically sporadic and does not have a clear genetic inheritance pattern.

Most studies on Coats' disease originate from the Western world, with very few major studies reported from the African continent. Only one case has been documented in Tanzania and one case series in Nigeria. The five cases presented in this study included four males and one female, all under three years of age. All patients exhibited profound visual loss with unilateral involvement (three in the right eye and two in the left eye). Notably, four of the patients were referred with an initial diagnosis of retinoblastoma, and one patient underwent enucleation based on the suspected malignancy before histological confirmation of Coats' disease.

Coats' disease typically presents in boys with a median age of 5 years, although cases range from neonates to adults. Severity is inversely related to the age of presentation, meaning younger patients tend to have more advanced disease and poorer visual prognosis. Unlike retinoblastoma, Coats' disease is non-heritable in more than 95% of cases and has no racial predilection.

Coats' disease is often misdiagnosed as retinoblastoma, primarily because both conditions can present with leukocoria (white pupillary reflex) and retinal detachment. According to a cohort study by Dalvin et al. (2), which included a large sample of patients with Coats' disease, found that younger age (three years or younger) at diagnosis was associated with more advanced disease, worse visual prognosis, and a higher likelihood of enucleation.

A case series study by Carol Shields et al, A large case series found that Coats' disease was the most common condition simulating retinoblastoma, emphasizing the need for proper diagnostic imaging to avoid unnecessary enucleation.

Takahashi et al., Japan (3) - Reported a high prevalence of Coats' disease in young boys with advanced exudation and retinal detachment, with fluorescein angiography aiding in diagnosis.

Ogun et al., Nigeria (4) - A case series highlighting the underdiagnosis of Coats' disease and its frequent misdiagnosis as retinoblastoma, leading to unwarranted enucleation.

MRI is useful for evaluating intraocular tumors like retinoblastoma. Unlike Coats' disease, retinoblastoma shows homogeneous or heterogeneous intraocular mass enhancement and often has optic nerve invasion or extraocular spread.

CT Scan helps in identifying calcifications within the mass, which are highly characteristic of retinoblastoma but absent in Coats' disease.

Conclusion

Coats' disease remains a rare but significant masquerader of retinoblastoma, particularly in young children presenting with leukocoria and retinal detachment. Sometimes, clinical diagnosis can be challenging, and enucleation may be necessary for definitive diagnosis as seen in one of our cases. A multimodal imaging approach, including B-scan ultrasound, fluorescein angiography, MRI, and CT scan, plays a crucial role in accurate diagnosis. Awareness and early detection are critical, as younger age at diagnosis is linked to more aggressive disease, poorer visual prognosis, and a higher likelihood of surgical intervention. Further studies are needed in African populations to better understand the epidemiology and clinical spectrum of Coats' disease.

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