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Case Report

Bilateral Retinoblastoma

Hussein Aldayyeni[1*], Riyam Al-Dulaimi[2]

[1]Nottingham University Hospital NHS Trust, the Queen 'Medical Centre, Nottingham, United Kingdom

[2]Neonatology clinical fellow in the Nottingham University Hospital NHS Trust, United Kingdom

***Corresponding author:** Hussein Aldayyeni, Nottingham University Hospital NHS Trust, the Queen 'Medical Centre, Nottingham, United Kingdom

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Summary

Bilateral retinoblastoma is one of the congenital retinal lesions that is found in the early life and also recognized as one of the reasons of loss of vision. It is an autosomal dominant condition that is mean if any of the biological parents has the condition should do the ophthalmological screen to the new-born baby in his few days of his life. It needs genetic screen. If the disease has been confirmed by either the screen or by the ophthalmology assessment, it needs an operation (surgical removal).

Background

Retinoblastoma is a rare form of cancer that rapidly develops from the immature cells of a retina [1], the light-detecting tissue of the eye [2]. It is the most common primary malignant intraocular cancer in children, and it is almost exclusively found in young children [3].

Though most children in high income countries survive this cancer, they may lose their vision in the affected eye(s) [4] or need to have the eye removed.

Almost half of children with retinoblastoma have a hereditary genetic defect associated with retinoblastoma. In other cases, it is caused by a congenital mutation in the chromosome 13 gene 13q14 (retinoblastoma protein). The most common and obvious sign of retinoblastoma is an abnormal appearance of the retina as viewed through the pupil, the medical term for which is leukocoria, also known as amaurotic cat's eye reflex [3]. Other signs and symptoms include deterioration of vision, a red and irritated eye with glaucoma, and faltering growth or delayed development. Some children with retinoblastoma can develop a squint [5], commonly referred to as "cross-eyed" or "wall-eyed" (strabismus). Retinoblastoma presents with advanced disease in developing countries and eye enlargement is a common finding [6].

Depending on the position of the tumors, they may be visible during a simple eye examination using an ophthalmoscope to look through the pupil. A positive diagnosis is usually made only with an examination under anesthetic (EUA). A white eye reflection is not always a positive indication of retinoblastoma and can be caused by light being reflected badly [7] or by other conditions such as Coats' disease [8].

Case Presentation

This is a case of a term neonatal who was born by an elective caesarean section at 36+5 weeks due to being small for gestational age. He was born in good condition, requiring no resuscitation but had a mild respiratory distress from few minutes of age and was transferred to the neonatal unit for observation. During his admission, the patient was treated for mild respiratory distress (TTN), hypoglycaemia and jaundice. On day 2 of his life, he was seen by the ophthalmology team due to his mother having a history of retinoblastoma and positive for RB1 gene.

Investigations

1. Bilateral ophthalmoscope assessment by the ophthalmology team showed bilateral retinal lesion (Figure 1).
2. Genetic screen was done to the new-born for RB1 gen.

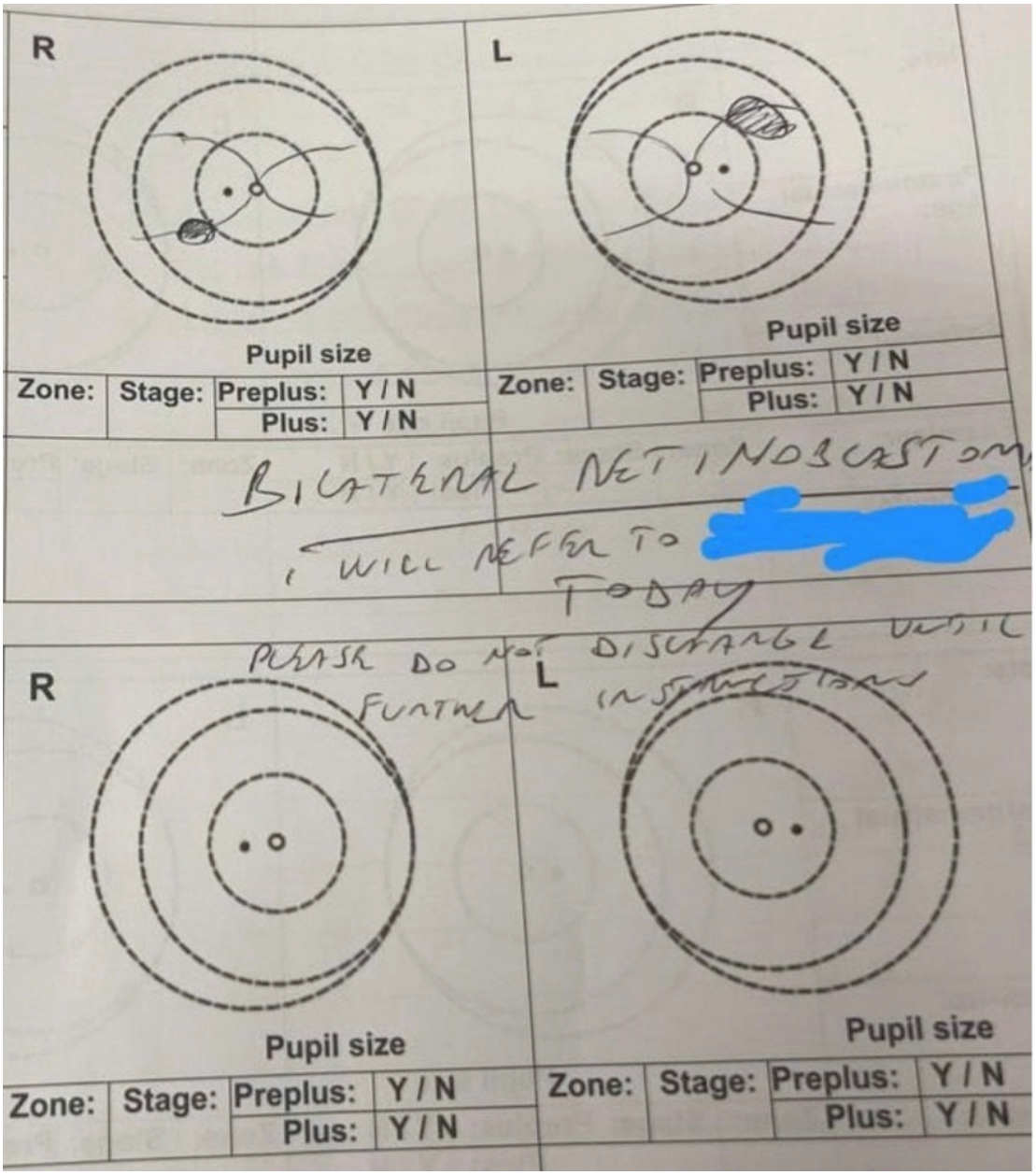

Figure 1: An ophthalmoscope assessment paper showing bilateral retinal lesion most likely retinoblastoma.

Treatment

The Ophthalmology team have done an ophthalmoscope examination and also found the patient having bilateral retinoblastoma. He was booked for operation for removal of the retinal lesion in few days' time and genetic screen for RB1 gene was sent.

Outcome and Follow-Up

The patient was booked for a surgical operation in few days' time and also he was awaiting the outcome for the genetic screen.

Discussion

Retinoblastoma is an autosomal dominant condition that is happen in the neo-born, children and early adulthood. If one of the parents having the disease, it means the child will carry a risk of 50% of having the disease in the future. Retinoblastoma is one of the reasons of blindness in the childhood and early adulthood. It needs a genetic screen for RB1 gene mutation. The management options will be surgical, chemotherapy or radiotherapy. It is the most common primary intra ocular malignancy in the children. For these reason, this new born was screened in the early days of his life to check if he has the condition. And after founding the retinal lesion, the patient was booked for an operation to remove it by the ophthalmology team. He will have a follow up appointments by the ophthalmology team to check his retina and check his visual acuity.

Learning Points/Take Home Messages

- Retinoblastoma is an autosomal dominant condition. It mean if one of the parents having the condition, the children having 50% of getting the condition in the future.
- If the parents having retinoblastoma, the new-born baby should be have a genetic screen for RB1 and should undergo an ophthalmoscopy assessment in the early few days of life.
- Surgical removal, chemotherapy and radiotherapy are the management options.

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