

# Leukocoria and Retinoblastoma in an Infant: A Case Presentation

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## 1. Introduction

It is imperative that pediatric health care providers preach and practice the importance of a thorough newborn (NB) assessment, whether within the first 24 hours of life, during the very first outpatient NB examination, or at any outpatient visits with the provider. These NB assessments identify abnormalities associated with anomalies, birth injuries, jaundice, or cardiopulmonary issues. The American Academy of Pediatrics (AAP, 2024) recommends that Preventative Pediatric Health Care include NBs being seen for their initial or interval history between days three to five of life.

The following case demonstrates the importance of checking for red-light reflex in pediatric patients during their examinations.

A NB female, born at 38 1/7 weeks gestation via normal spontaneous vaginal delivery (NSVD) with a birthweight of 6 lbs 2.8 oz (2.8 kg), was seen and examined at the hospital post-delivery by the primary care provider at fewer than 24-hours of life. The examination revealed a healthy NB female without any abnormalities or anomalies noted. Red-light reflex was noted in both eyes with this examination.

The patient's mother had good prenatal care, with blood work negative for Hepatitis B and HIV and positive for rubella immunity. There were no perinatal complications. The parents were both fewer than 30 years of age at the time of the patient's birth. The NB's APGAR scores were within the normal range for a full-term NB. The patient was her parents' third child.

The NB was seen at a private practice outpatient clinic at day four of life. The NB subsequently had one more follow-up appointment at day 10 of life, where documentation revealed a healthy and growing NB female. She had her wellchild checks at 1 month and 2 months of age where, again, everything was noted to be normal, including her red-light reflex. For outpatient pediatric primary care providers, wellchild checks/examinations correlate with the vaccine schedules, according to the "Recommendations for Preventative Pediatric Health Care, Bright Futures/American Academy of Pediatrics" (2024) at 3 – 5 days of life, by 1 month, 2 months, 4 months, 6 months, 9 months, 12 months of life, etc.

At her 4-month wellchild examination, the practitioner documented that the red-light reflex in her right eye was "absent" and "very little of whitish" was noted in her left eye. All other findings were normal. At this point, a stat referral was made to a local pediatric ophthalmologist. The patient was later referred to a pediatric ocular oncologist at the state's University of California hospital for more specialized care. She was seen within 2 days of the referral. The patient underwent testing that included a brain MRI with and without contrast where she was diagnosed with bilateral retinoblastoma 4 days after she turned 4 months old. Unfortunately, the right eye could not be saved and was removed. The infant underwent enucleation of her left eye a few times. She has partial vision in her left eye and wears a prosthetic right eye.

## 2. Case Discussion/Conclusion

Retinoblastoma typically presents as leukocoria in children under 3 years of age and accounts for 10% to 15% of cancers occurring within the first year of life. It is the most common primary intraocular malignancy of

childhood and can occur in heritable and nonheritable forms (Berry, 2024). As demonstrated by this case presentation, retinoblastoma can be devastating if not treated in a timely manner. With advances in the treatments, overall survival is now greater than 95% when accompanied with a prompt referral to an ocular oncologist for appropriate care and management. Aside from presenting as leukocoria, retinoblastoma can also commonly present as nystagmus and a red inflamed eye (2024). Both conditions warrant a prompt referral to an ophthalmologist.

The World Health Organization (WHO, 2024) recommends that red-light reflex screenings occur prior to discharge after a health facility birth or at the first postnatal care contact in an outpatient setting after a home birth. Documentation of red-light reflex, whether absent or present, should be recorded in the patient file.

Any dark spots, white reflex, marked diminished red-light reflex, and/or asymmetry of the light reflex warrant an urgent ophthalmology referral. Leukocoria, or white pupil, can signify abnormalities of the lens, such as cataract or retinoblastoma. An important question posed to the author was the length of time providers should continue to check for red-light reflexes in children. There is no definitive answer. However, it is highly recommended that providers check for red-light reflex at every encounter with children.

The process of examining for red-light reflex examination is not time consuming and can save “eyes and eyesight” if cancer is present and treated early. Have the parent hold the infant in their lap. It is crucial that the exam occurs in a darkened room. Begin the ophthalmoscopic examination of the eyes at a distance of about 46 cm or 18 inches away from the infant’s eyes. Project a beam of the light, at the largest diameter, onto the upper facial area of the infant. Adjust the lens to bring the skin around the child’s eyes into focus. The fundi should be visualized one at a time and then simultaneously. This process is called the Bruckner test. Examine the retina from the side view (obliquely) in addition to from a straight-on view. This technique may improve the detection of retinoblastoma (Drutz & Benjamin, 2023). It is important to note that patients with more darkly pigmented skin may have orange or light yellowish light reflexes instead of red. These are normal color variants of red-light reflexes.

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