Answer to Ophthaproblem *continued from page 155*

5. All of the above

Leukocoria, meaning "white pupil" in Greek, is the term used to describe a white pupillary reflex upon clinical examination.[1] A number of intraocular and systemic diseases can manifest as leukocoria at any age, although the differential diagnosis is more extensive—and worrisome—in children. The detection of leukocoria in a pediatric patient is therefore considered an ocular emergency that requires urgent evaluation by an ophthalmologist.[2]

Usually it is the parents who are the first to notice the strange appearance of their child's eye. They might discern a whitish discolouration of the child's pupil in dim lighting conditions or from certain angles. They might also report seeing a white eye instead of a red eye in flash photographs of their child, a phenomenon known as photoleukocoria.[3] Usually, flash photography causes the eye to appear red—as the pupil does not have time to contract, the camera captures a red reflection from the underlying retina. Any condition that blocks the light of a camera's flash from reaching the retina will produce photoleukocoria.[4] Photoleukocoria does not always signify underlying pathology. Marshall and Gole[5] reported an observational case series of 3 children who presented with unilateral leukocoria but had normal ocular examinations. In each case the child was fixating approximately 15° off axis, which resulted in a white pupillary reflex, as the normal optic nerve head was illuminated by the camera flash. This is known as pseudoleukocoria and it is normal. Nevertheless, it is essential that children with any suggestion of photoleukocoria be urgently examined by an ophthalmologist to rule out any life- or sight-threatening conditions.[6]

Differential diagnosis

In children, the differential diagnosis for leukocoria includes a wide spectrum of both ocular and systemic conditions. The main conditions that cause leukocoria are congenital cataract (60%), retinoblastoma (18%), retinal detachment (4.2%), persistent fetal vasculature (4.2%), and Coats disease (4.2%).[7] Many of these conditions are first suspected with routine screening (ie, red reflex examinations in all newborns and retinopathy of prematurity screening in premature babies) and require prompt referral to an ophthalmologist to optimize visual prognosis.[4]

The most important condition to rule out in a child presenting with leukocoria is retinoblastoma. Retinoblastoma is the most common intraocular malignancy in children, and is triggered by genetic mutations in 1 or more cells of the retina.[3] The incidence of retinoblastoma is 1 in 15 000 live births, with about 23 Canadian children being diagnosed each year.[8] Large reviews have shown that leukocoria is the most common presenting sign (>50% of cases).[2,8,9] As the disease can progress rapidly, retinoblastoma is considered an ocular emergency. Any delay in treatment can result in local and hematogenous metastases, which substantially increase mortality rates. If left untreated, retinoblastoma is universally fatal. Fortunately, with early diagnosis and treatment, more than 98% of children with retinoblastoma can be cured,[10-12] many with useful vision.[13] All children with leukocoria should immediately see an ophthalmologist for a dilated eye examination to rule out retinoblastoma.[4]

Management

Patients with leukocoria require urgent referral to an ophthalmologist for assessment, including a complete clinical and family history and a dilated ophthalmoscopic examination. If retinoblastoma is confirmed, the child requires a metastatic work-up, including neuroimaging, multidisciplinary management by a team of experts, and long-term follow-up. Treatment depends on the stage and extension of the tumour. Enucleation remains the treatment of choice for large unilateral tumours that threaten to extend outside the eye. Chemotherapy, cryotherapy, and laser treatment are used to salvage vision in patients with intraocular disease. Children with metastatic disease require extensive treatment, including high-dose chemotherapy with stem cell rescue.

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Competing interests
None declared

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