

## Ten Important Diagnoses during Pediatric Eye Check up

As a pediatric ophthalmologist at the University of Minnesota, I spend most of my day caring for children with eye diseases and teaching about and researching ocular conditions in children. Whether a cataract in a 7-day-old infant, glaucoma in a toddler, or nystagmus in a preschooler, these cases are often referred to me as the result of serial vision screening done by primary care physicians. Primary care physicians play a critical role in the early diagnosis and care of children with ocular disorders and, therefore, need to stay knowledgeable about the most common eye diseases and new developments in their management. This article reviews critical eye conditions that primary care providers may encounter in children and highlights recent advances in their treatment.

### **1. Refractive Errors**

Screening for refractive errors starts in infancy with serial red reflex exams and culminates with vision testing in school-aged children. When a child fails a vision screening, he or she often needs glasses. The most common scenario is an older child whose visual blur at a distance is detected through acuity testing.

There are, however, a variety of other, more serious refractive conditions that an astute primary care physician might pick up during a screening. A complete medical history will often uncover risk factors for other refractive problems in the first years of life. These risk factors include significant prematurity, diagnoses such as Marfan or Stickler syndrome, and a family history of wearing prescription eye wear.

In addition, primary care physicians should look carefully at the red reflex in children of all ages. Any variance in the pupil's color and intensity from top to bottom or right to left, or asymmetry in brightness compared with the other eye likely signals a refractive problem. This may be anisometropia, an unequal refractive error between the child's eyes, or a bilaterally high prescription, which puts the child at risk for amblyopia.

### **2. Amblyopia**

Amblyopia, often called "lazy eye," is a disorder characterized by poor or indistinct vision in an eye that is otherwise physically normal. It is a failure of normal vision development in a child's brain because of another ocular abnormality. In other words, the brain favors one eye because of a problem such as cataract, strabismus, or a major difference in refractive error between the eyes. Even when the inciting abnormality is addressed, the overlying amblyopia in the central nervous system still needs treatment. Amblyopia treatment typically involves having the patient wear a patch over one eye or administering atropine drops to blur the vision in the better eye.

Management varies depending on the patient's age, visual acuity, and related eye problems. Therapy typically begins within months of detection of a visual abnormality and may continue until vision matures.

Pediatric ophthalmologists at the University of Minnesota and other locations have participated in extensive research on the treatment of amblyopia associated with strabismus and far-sightedness through the Pediatric Eye Disease Investigator Group (PEDIG), a collaborative

network dedicated to multicenter clinical research into eye disorders that affect children. Notable findings include the following:

- Understanding that glasses play a critical role in rehabilitating vision,
- Defining atropine's beneficial role in treating certain types of amblyopia,
- Determining the amount of eye patching that is typically successful for various conditions, and
- Discovering that children as old as 18 years may benefit from amblyopia care.

Many pediatric ophthalmologists have incorporated the PEDIG findings into their practices. It should be noted that these discoveries may not be applicable to other causes of amblyopia including myopia, cataracts, glaucoma, ptosis, and retinal abnormalities. Ongoing PEDIG research is exploring treatment for amblyopia resistant to first-line management and the potential benefits of levodopa in such treatments.

### **3. Strabismus**

With the exception of brief episodes of eye misalignment seen in babies younger than 4 months of age, any constant ocular deviation (strabismus) should be referred to an ophthalmologist with pediatric expertise for evaluation (Figure 1). Strabismus is a common etiology for amblyopia and may be a harbinger of a cataract, retinal disease, or a neurologic abnormality. Screening for strabismus starts with simple observation immediately after birth and progresses to cross-cover testing during the preschool years.

Notable advances in strabismus surgery include the use of adjustable sutures, hidden conjunctival wounds, and earlier surgery for maximal stereopsis development. One result of the University of Minnesota's collaborative research on esotropia, more commonly referred to as crossed eyes, is that more pediatric ophthalmologists are recommending surgery for the condition at or before 6 months of age. Researchers are also studying the potential for Botox and other toxin injections for treating certain pediatric strabismus cases. Despite excellent initial motor alignment following surgery, long-term follow-up is often necessary to treat amblyopia and refractive errors. Because a patient's brain may let the eyes slip out of alignment, additional restorative surgery is needed in approximately 25% of cases.

### **4. Childhood Glaucoma**

Glaucoma is an optic nerve disease associated with high intraocular pressure and is a frequent etiology for blindness in children. Unfortunately, glaucoma often becomes quite advanced before significant signs and symptoms develop. Children who are sensitive to light, rub their eyes more than usual, and have larger-than-normal eyes should be referred to providers who specialize in pediatric glaucoma. Many cases of childhood glaucoma are associated with ocular disorders and systemic diseases such as Sturge-Weber syndrome and neurofibromatosis.

Surgical intervention is the standard treatment for infantile glaucoma. This is frequently performed soon after diagnosis, often during the first months of life. Recent advances, including fiberoptic instrumentation, allow for more safe and complete treatment of a child's intraocular aqueous drainage system. Through angle surgery, tube shunts, and chronic use of topical

medications, most childhood glaucoma can be successfully managed.

## **5. Pediatric Cataracts**

Because the first months of life are a critical period in vision development, a cataract that blurs vision can permanently stunt a child's ability to see clearly (Figure 2). Cataracts can be detected at any age on a careful red reflex examination. They are more common among children with a history of intrauterine infection, metabolic disease, and trauma, or those with a genetic syndrome or from families with a history of pediatric cataracts.

Cataract extraction and the ensuing optical correction and amblyopia rehabilitation may start when the child is as young as 1 month old. The surgery is an outpatient procedure in which the opacified lens is removed. Although the procedure is similar to that performed on older people, the size of the eye, the nature of the tissue, and the optical goals make pediatric surgery complex. We have found that when cataract surgeries are done in the first weeks of life, patients have a higher risk of glaucoma as compared with patients whose surgeries were done later on in life.<sup>2</sup> Our team at the University of Minnesota was recently selected by the National Institutes of Health to study intraocular lens placement in children as young as 1 month of age (Figure 3).<sup>3</sup> This ongoing research will help guide how we treat and manage pediatric cataracts in the future. However, the key to a child's visual outcome is their compliance with patching.

## **6. Nystagmus**

Nystagmus is a rhythmical movement of the eyes that may be detected on simple observation. This movement abnormality represents a defect in the calibration of the ocular motor system and is seen as an isolated finding or with neurologic and eye disease. Children with nystagmus need to be promptly evaluated and potentially treated for a related or inciting condition such as albinism, septo-optic dysplasia, or strabismus.

Nystagmus can be treated with spectacles, contact lenses, prisms, and muscle surgery for vision or head posture improvements. Affected families may find it helpful to know that their child's vision will often improve over the years and that a patient's visual perception is not affected as the eyes appear to move. New approaches in nystagmus surgery are being studied to determine how to maximize vision in affected individuals.

## **7. Capillary Hemangioma**

These benign collections of capillaries can wreak havoc on ocular structures and vision. Ophthalmic indications for intervention include occlusion of the visual axis, severe proptosis, anisometropia, severe facial asymmetry, and permanent vision impairment because of amblyopia. These lesions of the face, eyelid, or orbit may start small and grow significantly during the first months of life.

When capillary hemangioma threatens a child's vision, treatment with local injections or systemic administration of steroids or chemotherapy is necessary. Other treatments including laser for the superficial skin component, glasses for astigmatism, and patching for amblyopia may also be needed. The use of propranolol has been recently reported to help elicit resolution.<sup>4</sup>

## **8. Nasolacrimal Duct Obstruction**

Tearing and mattery discharge are commonly reported during vision screening in infants and toddlers. Obstruction of the tear drainage pathway often spontaneously opens during the first months of life. Physicians should be sure to rule out tearing from glaucoma based on the patient's medical history and the exam findings.

When tearing fails to resolve or if the obstruction is associated with dacryocystocele or dacryocystitis, treatment is indicated. This typically involves probing or stenting the nasolacrimal duct. Success rates run approximately 80%. Current research may better define the rate of resolution in toddlers and the success of intervention performed in the office versus the operating room.

## **9. Retinoblastoma**

A physician who detects leukocoria on a red reflex examination in a newborn or during the first years of life may actually save the child's life. This intraocular cancer may present as an isolated lesion in one eye or a bilateral manifestation of a heritable trait. Retinoblastoma is typically associated with an abnormal white reflex, strabismus, or vision loss.

Usually, retinoblastoma patients are treated aggressively through a coordinated effort involving the ophthalmology, hematology/oncology, and pediatric services. Intervention is tailored to the child's condition and the potential for saving vision. Treatment includes various types of chemotherapy, laser therapy, cryotherapy, and radiation plaque therapy. Intralesional chemotherapy is being investigated as a novel approach to drug delivery. Secondary systemic tumors remain a long-term risk in children with an underlying germline mutation.

## **10. Retinopathy of Prematurity**

Infants who are born significantly premature are typically examined for retinopathy of prematurity (ROP) while they are in the NICU. A critical role for the primary care physician is ensuring compliance with ophthalmology follow up after discharge from the hospital, as these children are at risk for retinal detachment, blindness, and functional abnormalities.

Although laser surgery may be used to treat severe retinopathy in the postnatal period, children who are born with ROP remain at significant risk for ocular morbidity. Long-term care is necessary to screen for retinal complications and to treat nystagmus, strabismus, and high refractive errors associated with ROP.

### **Serial Vision Screening**

Serial vision screening of newborns, infants, and children is critical to promoting ocular health, good vision, and general well-being. A thorough medical history, general evaluation, and ocular exam are necessary to detect eye diseases that commonly affect children. Because no eye exam has perfect sensitivity and specificity and because different conditions occur at different ages, screenings need to take place serially. One of

the most critical tests at every age is the red reflex examination, which can detect pediatric glaucoma, infantile cataracts, retinoblastoma, amblyopia, refractive error, and other problems. No other component of screening is as likely to detect as many disorders. The American Academy of Pediatrics (AAP) recommends that the red reflex be checked during the neonatal period and during all subsequent routine health visits.<sup>1</sup>

The AAP recommends the following screening eye exam components:

### **Birth to 3 Years of Age**

- Ocular history
- Vision assessment
- External inspection of the eyes and lids
- Ocular motility assessment
- Pupil examination
- Red reflex examination

### **3 Years and Older**

All of the above, plus:

- Age-appropriate visual acuity measurement
- Attempt at ophthalmoscopy

Photoscreening is also proving to be a valuable tool in detecting amblyogenic risk factors in the preverbal age group. The recent Medicaid photoscreening code allocation is paving the way for reimbursement to help promote and support such screening. Children who fail a vision screening or who are found to have risk factors in their history or upon systemic evaluation should be referred for further evaluation.