Greetings

Dear Colleagues:

This edition of the newsletter features a brief article on congenital corneal opacities by Dr. Danielle Trief, our pediatric cornea specialist. Our goal is always to present clinically relevant information that will help primary care practitioners on the front lines provide sound, informed management for their pediatric patients with eye issues. We also provide an EyeQ test to challenge your ophthalmic knowledge, as well as updates on the many things that are going on in the Division of Pediatric Ophthalmology at Columbia University Medical Center. As always, copies of this and prior newsletters, as well as detailed information about our outstanding physicians, services, and facilities can be found on our webpages at www.columbiaeye.org. We want to be an important resource for both you and your patients.

Congenital Corneal Opacities

The cornea should be transparent at birth and remain so throughout life. An infant born with a cloudy cornea is at very high risk of visual impairment and necessitates prompt evaluation by a pediatric ophthalmologist or cornea specialist.

If the cornea remains opaque, severe amblyopia ("lazy eye") is likely. Although some cases may clear with time or medical intervention alone, others may require surgery to restore clarity, a situation that will often require frequent, long-term follow-up and topical medication. There are several congenital conditions characterized by clouding of the cornea.

Peters anomaly (Figure 1) is a condition in which central corneal opacification is accompanied by adhesions between the iris and cornea (Peters type I) or between the cornea and lens (Peters type II). It may be either unilateral (40%) or bilateral (60%). Peters anomaly may be associated with genetic mutations in the FOXC1, PAX6, PITX2 or CYP1B1, genes, resulting in abnormal neural crest cell migration to the posterior cornea. Some children with Peters anomaly also have associated systemic abnormalities. (continued on page 2)
EyeQ Test:

1. A child with combined deafness and vision loss is most likely affected by:
   a. Usher Syndrome
   b. CHARGE Syndrome
   c. Congenital syphilis
   d. A. or B.

2. The most appropriate initial treatment for a stye of the eyelid is:
   a. Oral Keflex
   b. Topical steroids
   c. Warm compresses
   d. Surgical drainage

3. Gene therapy is currently FDA-approved for:
   a. Aniridia
   b. Infantile forms of retinitis pigmentosa
   c. Juvenile X-linked retinoschisis
   d. Congenital cataract

4. Avastin, an inhibitor of VEGF, is used to treat:
   a. Retinopathy of prematurity (ROP)
   b. Uveitis
   c. Retinal detachment
   d. Congenital glaucoma

5. Children are susceptible to amblyopia from
   a. Birth to 3 years
   b. 3 years to 6 years
   c. Birth to 6 years
   d. 2 years to 8 years


Congenital Corneal Opacities (continued from page 1)

The added presence of systemic features including short stature, developmental delay, dysmorphic facies, cardiac, genitourinary, and central nervous system malformations is known as Peters Plus Syndrome.

Congenital hereditary endothelial dystrophy (CHED) is another cause of congenital corneal opacification. In CHED the corneal endothelium is defective, resulting in congenital swelling, or edema, of the corneal stroma and epithelium (Figure 2). CHED is bilateral and involves the entire cornea. Unlike Peters anomaly, CHED can be treated with a lamellar corneal transplant consisting only of the endothelial layer and its basement membrane, a procedure called DSAEK (pronounced dee-sek). CHED is an autosomal recessive condition associated with mutations in the SLC4A11 gene, which codes for a channel protein involved in sodium and boron ion transport. In Harboyan syndrome, patients have CHED as well as progressive sensorineural deafness.

Other causes of congenital corneal opacities include birth trauma, infection, glaucoma, and corneal dermoids. Although many cases are sight threatening, prompt evaluation by a pediatric cornea specialist or pediatric ophthalmologist may provide an opportunity to save or restore vision. An awareness of the urgency for referral, as well as the potential need for genetic and systemic evaluation, will help ensure the best possible outcome for the patient.