THE EYES IN CHARGE: FOR THE OPHTHALMOLOGIST

Roberta A. Pagon, M.D.
Division of Medical Genetics, CH-25, Children's Hospital /Medical Center, Box C5371, Seattle, WA 98105-0371
bpagon@u.washington.edu  (206) 526-2056

OPHTHALMOLOGIC ANOMALIES SEEN IN CHARGE
Coloboma (80-90%)
  Iris
  Retina
  Optic nerve
  Microphthalmia
Facial nerve palsy (40%, unilateral >> bilateral)
Visual acuity abnormalities (90%)
Strabismus or amblyopia (frequent)
Ptosis
Cataracts
Retinal detachment
Photophobia (frequent)

DIAGNOSTIC TESTS
  Dilated funduscopic examination

MEDICAL CONSEQUENCES

- Colobomas of the iris typically do not affect visual acuity or visual field
- Colobomas of the retina cause visual field defects in the upper visual field. They also predispose the patient to retinal detachment.
- Colobomas of the macula and/or optic disk usually affect visual acuity significantly.
- Facial palsy can result in lack of blinking and resultant dry cornea which can lead to corneal scarring.

MEDICAL MANAGEMENT WITH CAVEATS

- Accurate description of visual acuity and visual field are of paramount importance for educational and communication purposes, particularly since most children with CHARGE have mild to profound hearing loss as well.
- Glasses (spectacles) to correct refractive error
- Tinted glasses for photophobia
- Occlusive patching for treatment of amblyopia
- Surgery for strabismus, cataracts, retinal detachment, as appropriate
- Artificial tears or gel to treat corneal exposure associated with facial palsy
- Regular (yearly) ophthalmologic evaluations to assess changes in visual acuity, refractive error, and potential for retinal detachment. Parents should be informed of the risk of retinal detachment and the importance of immediate medical assessment if there is any change in the vision status of the child.
NON-MEDICAL MANAGEMENT ISSUES

- A diagram of the visual fields should be given to families and vision teachers or therapists so that communication programming will be optimized. The visual field may be a crescent-shaped area of the lower visual field. Some children will tilt their heads back in order to compensate to see. Accurate assessment of the most comfortable head position for viewing objects is important.

- Low vision aids such as magnifying bars, televisions and binoculars may be helpful.

- Many children with CHARGE are sensitive to bright lights. Sunglasses can be very helpful in making the child more comfortable.

- Often the child appears to see better than would be predicted based on results of formal acuity and visual field testing. Many children who are legally blind function quite well visually. The parents and teachers usually can provide an excellent description of what the child can see.

- Demonstrate for parents what the vision is with best correction to help them understand what the child can and cannot see. For instance, parents frequently misunderstand that children with high myopia can see object moving at a distance when lighting and contrast are adequate, but cannot see detail clearly. In this situation, parents often have the impression that the child is not significantly visually impaired, when that is not the case.

- Most children with CHARGE have multiple anomalies, especially hearing loss. Significant vision problems combined with facial palsy, deafness, and inability to communicate may result in autistic-like behavior. However, once vision and hearing have been accurately assessed and an appropriate communication pattern established, such behaviors are often extinguished. Evaluation by a deafblind specialist (not simply a low-vision specialist) is essential.

REFERENCES
THE EYES IN CHARGE: PARENT INFORMATION

Robert A. Pagon, M.D.
Division of Medical Genetics, CH-25, Children's Hospital /Medical Center, Box 5371, Seattle, WA 98105-0371
bpagon@u.washington.edu  (206) 526-2056
and
Meg Hefner, M.S.
Assistant Professor of Pediatrics, Division of Medical Genetics, St. Louis University School
Hefnerma@slu.edu or Meg@chargesyndrome.org (314)768-8730

NORMAL STRUCTURE AND FUNCTION OF THE EYE

Structure: Parts of the eye

Function
Transmission of light
cornea
aqueous humor
lens
vitreous humor
Conversion of light to electricity: retina
Transmission of electrical signals to the brain:
optic nerve (cranial nerve II)
optic tract
Interpretation of electrical signals: occipital cortex and surrounding tissues (brain)
Problem List: eye problems seen in CHARGE

<table>
<thead>
<tr>
<th>Problem</th>
<th>Test(s)</th>
<th>Specialist</th>
</tr>
</thead>
<tbody>
<tr>
<td>Coloboma of the iris (keyhole pupil)</td>
<td>External examination</td>
<td>Ophthalmologist</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Pediatric/Family physician</td>
</tr>
<tr>
<td>Coloboma of retina, optic nerve</td>
<td>Dilated eye examination</td>
<td>Pediatric ophthalmologist</td>
</tr>
<tr>
<td>Visual acuity (blurriness)</td>
<td>Eye charts or cards</td>
<td>Pediatric ophthalmologist</td>
</tr>
<tr>
<td>Visual field defects (blind spots)</td>
<td>Dilated eye examination</td>
<td>Pediatric ophthalmologist</td>
</tr>
<tr>
<td></td>
<td>Visual field testing</td>
<td></td>
</tr>
<tr>
<td>Retinal detachment</td>
<td>Dilated eye examination</td>
<td>Ophthalmologist - retinal specialist</td>
</tr>
<tr>
<td>Corneal exposure secondary to facial palsy</td>
<td>External examination</td>
<td>Ophthalmologist</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Pediatric</td>
</tr>
<tr>
<td>Cataracts</td>
<td>External examination</td>
<td>Ophthalmologist</td>
</tr>
<tr>
<td>Potosis (droopy lids)</td>
<td>External examination</td>
<td>Ophthalmologist</td>
</tr>
<tr>
<td>Stabismus or amblyopia (weak eye)</td>
<td>External examination</td>
<td>Ophthalmologist</td>
</tr>
</tbody>
</table>

EFFECT OF PROBLEM ON CHILD

Iris coloboma:
This does not affect vision, but may make the child more sensitive to light (photophobia).

Retinal coloboma:
This will result in large blind spots, usually in the upper field of vision, (as if the child were wearing a baseball cap). Many children with retinal colobomas prefer to be upside down and to bottom-shuffle, in part because that way they can best make use of their available visual field.

Coloboma of the macula or optic nerve:
This often results in blurry vision as well as large blind spots. Children with extensive colobomas are often legally blind (20/200 acuity or worse). They may not look directly at objects or establish eye contact because of poor central vision.

Microphthalmia (small eye)
This can be associated with large colobomas of the retina.

Any coloboma of the retina or disk puts a child at increased risk of retinal detachment.
Any sudden change in vision should be treated as a medical emergency.

Strabismus
Loss of vision can result if not corrected.
DEVELOPMENTAL EFFECTS OF VISION LOSS (also see Development sections)

Infants with decreased vision will have delayed motor milestones. This is especially true for children with CHARGE, who often also have hearing loss, vestibular (balance) abnormalities and serious medical problems requiring multiple hospitalizations and surgeries.

Communication can be complicated by vision problems in children with CHARGE. Because of the hearing loss, sign language, speech reading and other visual communication is often used. Decreased visual acuity can make this more of a challenge.

MEDICAL TREATMENT OPTIONS

There is no medical or surgical treatment for coloboma

Retinal detachment can often be treated surgically if it is correctly diagnosed shortly after it occurs. Suspected retinal detachment should be regarded as a medical emergency.

Amblyopia may be treated with patching; strabismus may be treated with surgery.

Refractive errors that reduce visual acuity can often be helped with glasses.

NONMEDICAL MANAGEMENT

Photophobia can be helped by using tinted glasses, indoors as well as outdoors.

It is important to know the extent of your child’s visual field. If there is very little vision in the upper portion of the visual field, sign language and objects the child may want to see must be placed in the lower half of the visual field (in the lap). Many children compensate for small visual fields by adopting a certain head position or body position.

Education
If your child has both hearing loss and vision loss (even if she or he is not “deaf” or “blind”), the educational team for your child should include a specialist in deaf-blind children. Such specialists exist in every state in the U.S. Input from such an expert is important even if the hearing loss or vision loss is not “complete.”