

## THE EYES IN CHARGE: FOR THE OPHTHALMOLOGIST

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### 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 fundoscopic 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.

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## THE EYES IN CHARGE: PARENT INFORMATION

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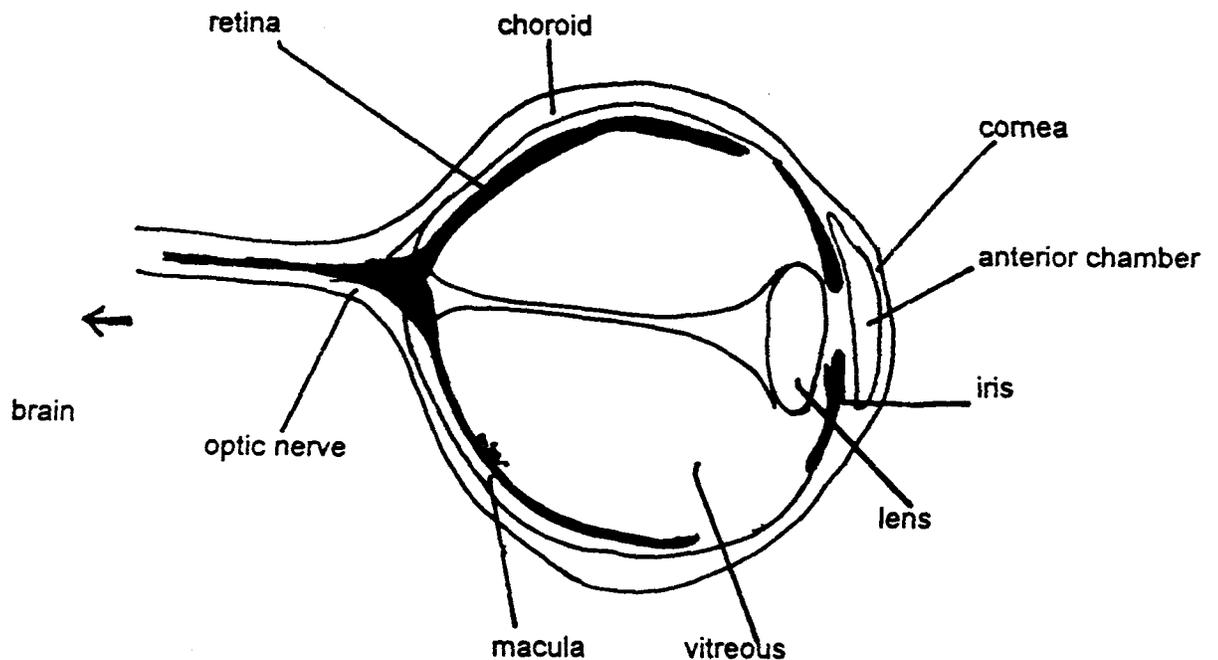
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### NORMAL STRUCTURE AND FUNCTION OF THE EYE

#### 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

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

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."