



Sunday, August 4, 2019

Breakout Session E27 • 10:45-11:45 am • Grand Ballroom D/E

A Visual Understanding of the Ophthalmic Pathway in CHARGE

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Presenter Information

Dr. Eniolami O. Dosunmu is a Pediatric Ophthalmologist in the Abrahamson Pediatric Eye Institute, at Cincinnati Children's Hospital Medical Center (CCHMC), Cincinnati, OH. She completed her Ophthalmology residency at the Mayo Clinic, Rochester, MN, and her fellowship in Pediatric Ophthalmology at the Duke University Eye Center, Durham, NC. She currently serves as the Pediatric Ophthalmologist for the CHARGE syndrome center at CCHMC. She enjoys the privilege of caring for her CHARGE patients, and her research interests are in describing the ophthalmic findings in CHARGE syndrome, translating how those findings affect functional vision, and in optimizing the visual function of the CHARGE patient based on their ophthalmic findings.

Presentation Abstract

The eye, the visual pathway and visual function are very important in CHARGE syndrome. This session will aim to show you the eye, the way the ophthalmologist sees it, and to show you how the CHARGE child, or adult sees the world. Using visuals, one will "look" into the eye of a CHARGE patient, and then "look" out at the world like a CHARGE patient. Following which, we will discuss ways to optimize visual acuity and visual function for the CHARGE patient. The session will also address Cortical Visual Impairment in CHARGE syndrome.

Learning Objectives

- Understand the most common ophthalmic findings in CHARGE syndrome
- Understand how these findings affect visual function
- Understand methodologies to optimize visual function based on the ophthalmic findings

14th International CHARGE Syndrome Conference

THE CHARGE EYE:

A visual walk through (understanding) of the ophthalmic pathway in CHARGE

The EYE*

Potential Anatomic Change	Evaluation and Potential Effect on Function and Vision
Eyelids: <ul style="list-style-type: none"> - Incomplete closure <ul style="list-style-type: none"> o Lagophthalmos o Palsy - Asymmetry of the eyelids 	Eyelids: <ul style="list-style-type: none"> - Symmetry - Dryness of the ocular surface
Lacrimal system/Nasolacrimal system: <ul style="list-style-type: none"> - Epiphora/tearing - Exposure Keratopathy from eyelids - Puncta present and open? - Nasolacrimal Duct Obstruction 	Lacrimal system/Nasolacrimal system: <ul style="list-style-type: none"> - Epiphora/tearing <ul style="list-style-type: none"> o Blocked tear duct? o Foreign body o Other process? - Exposure Keratopathy (dryness) from eyelids <ul style="list-style-type: none"> o Risk of recurrent corneal erosions (scratches on the surface of the eye) o Risk of corneal infection o Effect on vision - Nasolacrimal Duct Obstruction <ul style="list-style-type: none"> o Tearing o Infections
Cornea: <ul style="list-style-type: none"> - Size? <ul style="list-style-type: none"> o Microcornea o Corneal opacities - Exposure keratopathy from the eyelids 	Cornea: <ul style="list-style-type: none"> - Microcornea <ul style="list-style-type: none"> o Is this limited to the cornea, or does it reflect the potential anatomic changes in the eye? - Corneal clarity - Exposure keratopathy - Corneal astigmatism
Iris: <ul style="list-style-type: none"> - Coloboma 	Iris: <ul style="list-style-type: none"> - Coloboma <ul style="list-style-type: none"> o Location o Size o Photophobia o Anisometropia and amblyopia - Other: APD[†]
Lens: <ul style="list-style-type: none"> - Colobomatous? <ul style="list-style-type: none"> o Induced astigmatism? o Typically located inferiorly 	Lens: <ul style="list-style-type: none"> - Size of coloboma - Photophobia - Refractive error that needs correction - Cataract:

<ul style="list-style-type: none"> ○ Degree determines clinical significance ○ Photophobia ○ Induced Astigmatism ○ Anisometropia - Cataract <ul style="list-style-type: none"> ○ Opacification, or clouding of the lens ○ Location? - In proper location? <ul style="list-style-type: none"> ○ Lens dislocation or subluxation 	<ul style="list-style-type: none"> ○ Depends on if the cataract is visually significant ○ Outside of the visual axis = no treatment ○ Manage risk factors ○ When indicated, surgical extraction
Optic Nerve: <ul style="list-style-type: none"> - Colobomatous - Any associated fluid - Other anatomic changes - Associated with chorioretinal coloboma 	Optic Nerve: <ul style="list-style-type: none"> - Retina and choroid
Retina and Choroid: <ul style="list-style-type: none"> - Colobomatous? <ul style="list-style-type: none"> ○ Location determines the visual implication ○ Visually significant if involving the macula ○ More so if it involves the fovea 	Retina and Choroid: <ul style="list-style-type: none"> - Location and anatomic change are the determining factor for care - Can result in refractive errors - Colobomas are associated with retinal detachments - Colobomas are associated with visual field defects

Cortical Visual Impairment:

- Visual processing/impairment secondary to a cerebral/cortical process
 - Often times can coexist with an ocular reason for visual impairment
- Difficulty with distance viewing
- Visual Reflex responses
- Difficulty with new visual input
- Visual motor delay

References:

- American Academy of Ophthalmology (AAO.org)
- American Association for Pediatric Ophthalmology and Strabismus (AAPOS.org)
- McDonald HR, Lewis H, Brown G, et al.: Vitreous surgery for retinal detachment associated with choroidal coloboma. *Arch Ophthalmol*.109:1399-1402 1991
- Lawrence A. Yannuzzi. The Retinal Atlas.1st edition.
- Creig S. Hoyt and David Taylor. [Pediatric Ophthalmology and Strabismus](#). 4th edition.

* Focusing on the more common findings

† Result of the optic nerve pathway