Pediatric Clinical Support: CHARGE Syndrome

First described in 1979 and then used as an acronym in 1981 (Pagon, Graham, Zozana, &Yong, 1981), CHARGE syndrome is a genetic disorder that is linked with a non-random pattern of unusual congenital features. Six features were initially used to describe the syndrome, however, now more than 20 features are included in this list (http://www.asha.org/aud/articles/CHARGESyndrome.htm). Prevalence of this rare condition is approximately 1 in 10,000.

Characterized by:

- **C**oloboma of the eye
- **H**eart defects
- **A**tresia of the nasal choanae
- **R**etardation of growth and/or development
- **G**enital and/or urinary abnormalities
- **E**ar abnormalities and hearing loss.*

**Structural abnormalities can occur with almost all portions of the auditory system, including: very soft/floppy pinnae that may not provide enough support for a BTE, ossicular malformations, Mondini dysplasia, incomplete or missing semicircular canals, retrocochlear involvement and neural abnormalities.*

Not all individuals will be diagnosed with all characteristics, but the different combinations of characteristics can create a unique set of both physical and sensory deficits. CHARGE syndrome is the leading cause of congenital deaf-blindness.

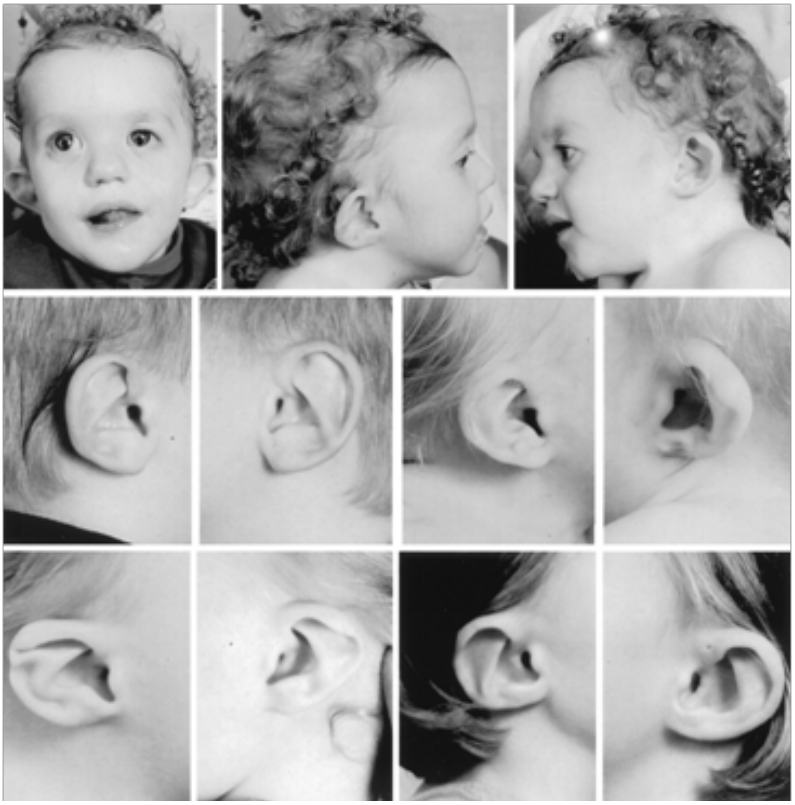

Figure 1. Examples of eye and ear anomalies related to CHARGE syndrome. Note the coloboma of the eye (top left) and bowl-shaped and concave ears.
From: http://pediatrics.aappublications.org/content/108/2/e32/F1.large.jpg

Manifestation: Can include abnormal appearance of ears, developmental delay, hypospadias, undescended testicles, and other urinary abnormalities. Typical delays are seen with motor skills and intellect.

Gene Expression: CHARGE has been connected with mutations on the gene CHD7, which plays a role in gene transcription regulation by chromatin remodeling. Several studies reported approximately 60% or more of the participants had a mutation of the CHD7 gene.

Differential Diagnosis: Severe developmental delay, autism spectrum disorders, auditory neuropathy/dyssynchrony, velo-cardio-facial syndrome, cat eye syndrome, VATER association, Wolf-Hirschhorn syndrome, trisomy 13.

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Behavior, Communication, and Advocacy: Behaviors can be challenging and include defensiveness, autistic-like behaviors, and even self-mutilation. Many individuals with CHARGE do not have symbolic communication, so it is helpful to have a caregiver or parent present to aid in dialogue with others. Parents must learn to advocate for their child in terms of services for communication, development, and learning.

Audiological Findings: Approximately half of those diagnosed with CHARGE syndrome have severe to profound hearing loss, although lesser degrees of sensorineural losses and conductive losses have been noted. There is no "typical" configuration noted for hearing loss related to CHARGE syndrome.

Treatment and Rehabilitation: Hearing evaluations to determine severity, which can determine appropriate amplification or cochlear implants for profound losses. Hearing assistive technology can be used in addition or in conjunction with amplification and/or cochlear implants. American Sign Language can be used as a communication method. Orientation and mobility training will aid in balance issues.

Educational Considerations: CHARGE should be indicated and addressed in the child's IEP, as hearing, vision, and other medical conditions may impact their classroom abilities. The amount of support required by the child should be established and recorded in the child's IEP. This includes allowing access to information, including strategies to systematically develop the use of sensory information with remaining functional vision and hearing, and effective strategies and approaches that teach environmental information that the child may be missing. Integration in social environments could include methods to orient the child to other students and locations, and improve the number and quality of interactions and relationships that the child has with others.

Professional Considerations: Early intervention should include occupational therapy and physical therapy to promote mobility, improve static posture and ambulation, and to teach self-care. Due to the potential combinations of systems within the body being affected, quality of life considerations should be addressed, including the implications of dual sensory loss. A large support team may be involved with the child diagnosed with CHARGE syndrome, including pediatrician, oral/maxillofacial surgeon, ENT, ophthamologist, cardiologist, urologist, developmental specialist, radiologist, low vision or mobility instructor, child social worker or psychologist, speech pathologist, and audiologist.

Support and Other Online Sources:

http://www.asha.org/aud/articles/CHARGESyndrome.htm

http://www.chargesyndrome.org/

http://emedicine.medscape.com/article/942350-overview

http://ghr.nlm.nih.gov/condition/charge-syndrome

http://www.ncbi.nlm.nih.gov/books/NBK1117/

http://www.ojrd.com/content/1/1/34

Other References:

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Shprintzen, RJ (2001). Syndrome Identification for Audiology: an Illustrated Pocket Guide. Singular, Thomson Learning: San Diego.

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Vissers LE, van Ravenswaaij CM, Admiraal R, Hurst JA, de Vries BB, Janssen IM, et al (2004). Mutations in a new member of the chromodomain gene family cause CHARGE syndrome. Nature Genetics, 36(9): 955-957.

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