

CHARGE SYNDROME

ABOUT THE DISORDER

CHARGE syndrome is a complex genetic condition seen in about 1 in 10,000 births. The name “CHARGE” is an acronym stemming from some of the features seen in many children with CHARGE: **C**oloboma, **H**eart, **A**tresia of the choanae, **R**etardation of growth and development, **G**enito-urinary, **E**ar anomalies and hearing loss.

CHARGE syndrome is no longer diagnosed using the acronym features. A diagnosis of CHARGE syndrome is made using **Major** and **Minor** clinical features or by finding a mutation in the CHD7 gene. CHARGE syndrome is a genetic condition most often caused by a new change (mutation) in the CHD7 gene on chromosome #8. It is usually sporadic, with no other affected family members. It is found in every ethnic group all over the world.

Major features of CHARGE syndrome are:

- **Coloboma:** Coloboma is a cleft of the eyeball. It can result in a keyhole-shaped pupil (iris coloboma) and/or abnormalities in the retina or optic nerve. Colobomas of the retina or optic nerve can cause vision loss or blindness.
- **Choanal atresia or stenosis:** a blockage or narrowing of the airway from the back of the nose to the throat. Many individuals with CHARGE also have apnea or other breathing problems.
- **Cranial nerve abnormalities.** Most individuals with CHARGE syndrome have abnormalities of the cranial nerves (nerves for the head and neck), especially facial palsy (partial paralysis of the facial muscles), swallowing difficulties, and decreased or absent sense of smell. The swallowing problems can be severe and require tube feeding for many years to get adequate nutrition and avoid aspiration pneumonia.
- **Characteristic CHARGE ears:** Most individuals with CHARGE syndrome have unusually formed ears. They are often short, wide (cupped) ears with little or no earlobe. Sometimes the outer fold is missing. They are often very floppy. Most people with CHARGE syndrome have *hearing loss*, ranging from mild to profound and inner ear problems leading to *poor balance*.

Minor features of CHARGE syndrome are:

- Heart defects (often complex and difficult to correct)
- Cleft lip or palate
- Tracheo-esophageal (TE) fistula or esophageal atresia
- Genital abnormalities
- Kidney problems
- Characteristic face
- Hockey-stick hand crease.

Other features of CHARGE syndrome may not be as helpful in making a diagnosis, but they can be very important for management. These include growth deficiency, poor upper body strength, sleep difficulties, and typical behaviors (perseverative behavior, obsessive-compulsive disorder).

SYMPTOMS or BEHAVIORS

Children with CHARGE syndrome have developmental delays. Some will have mental retardation, but many have normal intelligence. Because of all of the medical issues, many children spend months in the hospital and have multiple surgeries, especially in the first few years of life. Most are followed by six or more medical specialists. Even so, referrals to educational resources as early as possible are very important. Children with both hearing loss and vision loss are classified as “*deafblind*.” These children need to be followed by specialists in deafblind. ***Early diagnosis and intervention for hearing loss and vision loss are critical to development of communication.***

A comprehensive educational program should include information on balance, muscle tone, breathing, feeding, and other specific needs, including vision, hearing and communication.

CHARGE is medically and developmentally one of the most complex conditions known. Children with CHARGE syndrome are also likely to be amongst the most truly 'multi sensory impaired' people you will ever meet, having difficulties not just with vision and hearing but also with the senses that perceive *balance, touch, temperature, pain, pressure, and smell*.

Sensory Integration Difficulties

- Rejecting of textures in the mouth apart from pureed food, but mouthing of all kinds of non-food items, for example, stones, wood, cloth, soil
- Absence of chewing and biting on solid foods, but excessive chewing and biting on non-food items, often with persistent teeth grinding
- Rejecting certain tactile inputs as if they are painful, but apparent non-awareness of certain other tactile inputs (which for others might be painful!)
- In the early years, extreme postural insecurity when placed in a sitting or standing position by an adult, or when moved unpredictably, but pleasurable responses to strong rhythmic movement experiences (e.g., rocking, bouncing, swinging)
- Severe problems with regulating arousal levels, often described as periods of frantic over-activity and over-excitement and stress, but also sudden periods of apparent “burn-out”
- Abnormally high pain thresholds
- Inconsistent or inappropriate use of pressure when touching or grasping with the hands, often described as the child being very “rough” or “clumsy” or “aggressive,” and generally poorly graded movements
- Very delayed awareness of bowel and bladder movements
- Disturbed and inconsistent sleep patterns
- Behaviors that seek and provide very strong sensory inputs like self-biting or scratching, skin picking, spinning, rocking, bouncing, shoulder shrugging, leg swinging, hyperventilating, hand flapping, self-slapping, as a way of getting the body reorganized

Physical Characteristics of Children with CHARGE Syndrome

Children with CHARGE syndrome have physical characteristics which affect their gross motor performance. These characteristics interact with each other to affect the child’s balance and ability to interact with their environment. The following areas are common characteristics:

Low muscle tone

Visual Impairments

Skeletal Anomalies

Vestibular System Impairments

Balance

EDUCATIONAL IMPLICATIONS

COMMUNICATION

Most children with CHARGE have both vision loss and hearing loss (dual sensory impairment: Deaf-blindness). Most benefit from a **total communication** approach. Total communication means incorporating anything and everything: gestures, simple signs, print/Braille, facial expression, symbols, and PECS (Picture Exchange Communication System) in addition to speech and sign language. The modes for each child are highly individualized and the entire team (including the family) needs to participate in developing and consistently instituting the plans.

SENSORY LOSSES AND THE IMPACT OF VISION AND HEARING LOSS

It is important for the team to take careful notice of the functional vision of the child as well as the hearing loss, the impact of hearing loss on communication and the total effects on day-to-day functioning. Determine the communication bubble! [The communication bubble includes how far out and how well a child sees and how far out and how well a child hears. It also needs to take into account things like lighting, ambient noise and other factors.] Many types of accommodations will likely be necessary in the educational setting and these should be written into the child's IEP. Consultation with vision and hearing specialists can be helpful in determining appropriate accommodations.

INSTRUCTIONAL STRATEGIES AND CLASSROOM ACCOMODATIONS

As far as possible, these ideas should be integrated into all educational settings and across all environments:

Strategies for structuring activities

- Organizational skills- work in an organized manner (left to right, top to bottom), using checklists
- Negotiation – allow the child to feel as if he/she is in control. Give a variety of choices as well as use first, then, strategies.
- Sharing – foster peer-to-peer interactions. Encourage turn taking as a precursor for conversational communication.
- Motivation- select activities that are interesting to the student. Use materials that are interesting and rewarding.
- People preferences – give choices of WHO should do tasks/lessons with the student whenever possible.
- Partial vs. Full Participation – have the child do as much of a task or activity as possible to feel successful. Avoid setting up for failure by having an activity that has too many steps or is too long.
- Functional Activity- in all activities, ask yourself, “Will this skill or activity be useful in the future?”
- Natural routines environment – teach in environments in which activities naturally occur.
- Variety of exciting content – although routines and structure are important, keeping the content dynamic is critical for the student with CHARGE!
- Task analysis – break tasks into smaller steps that allow the child to feel successful.

Sensory techniques

- Awareness of hands/touch-allowing others to touch for signaling, (tapping) tactile, signing, etc.
- Signals: Verbal/Auditory/Visual-gesturing to gain attention, using voice or sound cue to gain attention
- Prompt levels hand-under-hand – use an adults' hand as guides under the child's as a less invasive technique.
- Sensory breaks – allow pause time during activities and in between activities.

Timing

- Beginning-middle-end – make sure all activities have a clear beginning, middle, and end that are at the student’s level of understanding.
- Structure and routine – having predictable schedules and routines play to the strength of the child.
- Pause time for response – WAIT, WAIT, WAIT, allow the child with CHARGE to respond in a manner they are most comfortable.

Curriculum

- Child centered curriculum – the child should drive the curriculum, the curriculum should not drive the child.
- Expanding environments –some children with CHARGE need to start activities in “smaller” environments (less physical space or less cluttered). As they mature and grow environments can become bigger (physically) and more challenging.

What Every Student with CHARGE Needs to Know:

Social Skills (how to be an effective communicator)

- How to be a part of a group- as member that is accepted by the group
- How to Negotiate- how to get their point across and know when to surrender
- How to take turns & share- - to take the appropriate amount of turns in a conversations—BE a listener as well as a speaker signer.
- How to help out – be part of a family by doing chores. In school have school jobs.

Cognitive Skills

- How to be organized- work, think & execute in an organized manner
- How to anticipate activities (calendar system)- know exactly when the order things will happen
- How to cope with behaviors- know when & what to do when on “sensory” overload or stressed.
- How to make choices- know that there are choices & the choices may be from a “set” group. Know that all choices are not possible at all times.

Educational Professionals Who May Be Involved

General Education Teacher
 Special Education Teacher
 Teacher for the Visually Impaired
 Deaf/Hard of Hearing Teacher
 Speech Language Pathologist
 Adaptive Physical Education Teacher
 Occupational Therapist
 Physical Therapist
 School Counselor

RESOURCES

CHARGE Syndrome Foundation, Inc.
 Phone: (516) 684-4720 Fax: (516) 883-9060
 800-442-7604 (families)
 Email: info@chargesyndrome.org
<http://www.chargeysndrome.org>

Minnesota DeafBlind Technical Assistance Project
<http://www.dbproject.mn.org/education.html>

American Speech-Language Association (ASHA)
www.asha.org/Publications/leader/2006/.../f061017a.htm

Minnesota Low Incidence Projects
<http://www.mnlowincidenceprojects.org/pi.html>