



Genetic Education Materials for School Success

CHARGE Syndrome

Table of Contents

| | |
|--|----|
| At a Glance | 2 |
| Medical/Dietary Considerations | 3 |
| Education Supports | 5 |
| Behavior and Sensory Support | 8 |
| Physical Activity, Trips, Events | 10 |
| School Absences and Fatigue..... | 11 |
| Emergency Planning..... | 11 |
| Resources | 12 |
| Info for School Nurse and Primary Care Staff..... | 13 |
| Meet a Young Woman with CHARGE Syndrome : <i>Tricia!</i> | 14 |

Acknowledgment:

Genetic Education Materials for School Success (GEMSS) was created and maintained by an interdisciplinary team of genetic counselors, physicians, families, and students working with New England Regional Genetics Network (NERGN) and the Institute on Disability at the University of NH. Thank you to these amazing partners. This work was supported by HRSA grant UH7MC30778.

Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

CHARGE Syndrome

At a Glance

CHARGE syndrome is a rare genetic condition that affects many parts of the body. CHARGE stands for **C**oloboma, **H**ear defects, **C**hoanal **A**tresia, **R**etardation, **G**enital abnormalities, and **E**ar anomalies.

Most children will have hearing loss, vision loss, and balance problems. These individuals often have other physical and behavioral differences, but the effects of CHARGE syndrome are different for each person.

CHARGE syndrome usually occurs as new mutations meaning there is no family history. However, in rare cases, an affected person inherits the gene from an affected parent.



Meet Tricia on page 14

Common Features of CHARGE Syndrome:

- Small or underdeveloped eyes
- Coloboma – missing piece in the eye structure which may impair vision
- Narrow or blocked nasal passages (*choanal atresia*) which may impair breathing
- Cleft lip/palate
- Unusually-shaped ears
- Cranial nerve abnormalities
 - Swallowing problems
 - Paralysis of face
 - Diminished sense of smell
 - Hearing loss
- Heart defects
- Slow growth
- Delayed motor skills
- Delayed puberty
- Wide range of cognitive function – from normal intelligence through major learning disabilities

In-Depth

Medical/Dietary Considerations

What you need to know

The severity of any one of the possible medical conditions varies widely between individuals, therefore, it is important to ask the parents about their child's medical issues. Children often have problems with breathing, swallowing, eating, drinking, and temperature control. School-age children with CHARGE will have multiple doctor and specialist visits to monitor medical conditions.

Medical Considerations

- Heart defects – may require more than one surgery
- Neurologic Issues
 - Hydrocephalus – fluid buildup in brain
 - Seizures
 - Cranial nerve abnormalities may lead to
 - missing or decreased sense of smell
 - swallowing difficulties and possibly aspiration
 - facial palsy (paralysis) on one side or both
- Eye abnormalities
 - Vision impairment varies
 - Coloboma (missing pieces of tissue in structures that form the eye)
 - More severe forms include microphthalmos (small eyes) or anophthalmos (missing eyes)
- Ear abnormalities
 - May have chronic ear problems
 - Outer ear anomalies
 - Often outer ear is short and wide with small lobes
 - Inner ear folds are prominent, and outer folds appear “snipped off”
 - Middle ear anomalies
 - Bones of middle ear are malformed causing conductive hearing loss
 - Prevalence of severe to profound hearing loss is 50%
 - Inner ear anomalies
 - Malformed cochlea and/or small or absent semicircular canals
 - Can cause hearing loss and balance problems
- Narrowed or blocked passages between the nose and the throat (choanal atresia)
- Cleft lip - with or without cleft palate
- Abnormalities with trachea and esophagus – feeding and choking issues
 - Tracheoesophageal (T-E) fistula
 - Esophageal atresia (esophagus is not open)
- Kidney abnormalities - small, missing, or misplaced kidney

- Gastric reflux
- Hypotonia- low muscle tone –
- Genital abnormalities - delayed or lack of puberty without hormone intervention
- Growth deficiency - short stature
- Other Structural Findings
 - Sloping shoulders (common) due to underdeveloped shoulder muscles
 - Small or missing pectoral (high in chest) muscles
 - Short neck
 - Skeletal anomalies
 - Absent thumb
 - Extra fingers
 - Vertebral abnormalities
 - Scoliosis
 - Umbilical hernia
 - Extra, missing, or misplaced nipples

Motor Delays

- May result from
 - Low muscle tone in trunk/core
 - Balance problems/inner ear dysfunction
 - Reduced sensory input
 - Poor vision, positional awareness, tactile senses
 - Can lead to reduced perceptual awareness
 - Breathing difficulties
 - Lack of motivation to move or resistance to exercise
 - Prolonged hospitalization
- Note –Low tone may cause a child to use excessive force in movement. This along with poor coordination of movements may be perceived as aggressive, rough, or clumsy

Dietary Considerations

- Feeding and swallowing difficulties
 - Causes and severity vary greatly
 - Swallowing mechanics may be affected by
 - abnormalities of the cranial nerve
 - weakness and motor planning disorders
 - May lead to dehydration/malnutrition
 - Food aspiration, choking
- Sensory difficulties can contribute to feeding and swallowing challenges
 - Tactile defensiveness
 - Oral hyposensitivity

What you can do

- Encourage a yearly check-up and studies as needed in the child's Medical Home. Obtain updated recommendations as available

- Follow any medical recommendations as provided
- Consider PT/OT
- Work with parents to ensure the child is fed in a way that is best for the child
- Be aware of any changes in behavior or mood that seem out of line with the situation and notify the parents.
- It is important to be aware of any academic changes. Contact parents when any differences are noticed.
- If there are seizures, make sure you have a seizure care plan.

Education Supports

It is important to have high learning expectations for children who have CHARGE, utilizing their strengths and interests as a foundation. Encourage use of the core educational curriculum and modify it to meet the child's individual needs.

What you need to know

Individuals with CHARGE syndrome often have multi-sensory challenges. They often have difficulties with vision, hearing, smell, and the senses that perceive balance, touch, temperature, pain, and pressure. Intelligence may be underestimated because of their sensory, learning, motor, and/or speech disabilities. It is important for teachers and caretakers to take time to develop a relationship with the children and their families. A team that is knowledgeable in sensory challenges is an important part of the child's education team. Regular neurodevelopmental and developmental / behavioral evaluations may be helpful.

Communication/Speech

- Individuals with CHARGE may have challenges in developing clear speech and/or the ability to sign well.
 - Language delays are caused by multiple sensory and motor challenges, and by delays in finding an appropriate communication system.
 - Speech may be affected by structural anomalies, craniofacial nerve issues, and breathing problems.
 - Bilateral facial palsy and central vision loss can lead to lack of facial expression and this can add to individual's communication problems.
 - Challenges to signing:
 - Under-functioning tactile and proprioceptive sense
 - Low muscle tone
 - Severe balance problems
 - Dyspraxia – problem coordinating movement
 - Initiating speech/signs may be due to specific brain anomalies
- Most individuals benefit from a total communication approach, i.e., incorporating anything that works

Hearing

- Hearing deficits are common and have multiple, overlapping causes

- Deficits can be significant and impact learning and classroom participation
- An audiological evaluation is vital but can be challenging
- Hearing impairments and vestibular (balance) abnormalities affect the amount and quality of information received from the environment

Vision

- Children with CHARGE often have vision challenges due to colobomas
- Colobomas can cause
 - Visual field loss
 - Blind spots
 - Acuity problems
 - Light sensitivity
 - Monocular vision
 - Lack of depth perception
- It is important to understand the extent of their vision loss

Balance

- Postural security and good sense of equilibrium depends on vision, balance, tactile, and proprioceptive senses
- Malfunctioning or absent semicircular canals (the receptors of balance sense) in the inner ear can impact balance.
 - This may affect walking and standing
- There is a link between balance sense and ability to process sound and to develop spoken language
- Problems with balance may affect the ability to maintain a stable visual field, follow moving objects, and differentiate when one thing is moving
 - May compensate for lack of visual sense by using the walls, door and window lines as markers
 - May be reluctant to go outside because these “markers” don’t exist outside
- Problems with postural control, sitting, and standing may cause fatigue

What you can do

A team that is knowledgeable in sensory challenges is an important part of the child’s education team. Members of this team may include:

- Physical therapy
- Occupational therapy
- Speech therapy
- Sensory integration program specialist
- Deaf or Deafblind specialist

General Strategies

- Encourage organizational skills -help child to work in organized manner
- Negotiation - allow child to feel in control
- Sharing -foster peer to peer interactions

- Motivation- select skills that are interesting to student
- Functional use - ask if activity or skill is useful or appropriate to student
- Set child up for success
 - Model steps in the activity
 - Have clear expectations and firm limits
 - Break assignments into small tasks
 - Give choices as much as possible
- Establish consistent routines
- Sensory techniques
 - Awareness of hands/touch
 - Allowing others to touch for signaling/tapping
 - Signing
- Signals
 - Gesturing to gain attention
 - Voice or sound cue to gain attention
 - Use adult's hands as guide
 - Help with effective communication
- Sensory break
 - Allow pause time during and between activities
 - Provide flexible schedule for breaks
- Curriculum
 - Child-centered curriculum
 - Expanding environment
 - Start small and expand as child is comfortable
- Social skills- encourage how to
 - Be a part of a group
 - Negotiate
 - Take turns and share
 - Help others
- Cognitive skills- help them to
 - Be organized
 - Anticipate activities
 - Cope with behaviors
 - Make choices

Communication/Speech

- Most individuals benefit from a total communication approach, i.e., incorporating anything that works including
 - Gestures
 - Simple signs
 - Braille/print
 - Facial expressions
 - Symbols (PECS or picture exchange communication system)
 - Speech therapy

- Sign language

Hearing

- Consult deafblind specialists if needed
- Obtain audiological evaluation
 - Address specific problems identified (suggest hearing aid, minimize background noises, etc.)

Vision

- Consult deafblind specialists if needed
- Make accommodations for safety
 - e.g., visual enhancement, like bright duct tape on a step
- Address field loss or monocular vision
 - For reading difficulty use
 - large bold print
 - bold lines
 - underlining or highlighting
- Address photophobia (intolerance to light) by using
 - Tinted glasses
 - Sun visor
 - Low classroom lighting
 - Rest breaks in a dark room

Balance

- Coordinate with PT and OT
- Problems with postural control, sitting, and standing may cause fatigue
 - Young children may benefit from adaptive chair with arms and footboards
 - Children may need to get into horizontal position to “relax” after sitting position
- May need supports for difficulty with sitting or standing
- May sit at a desk but need a break in a horizontal position to relax

Behavior and Sensory Support

What you need to know

Children with CHARGE often have challenges in several sensory systems. Sensory impairments are caused not just by vision and hearing loss, but also because the senses that perceive balance, touch, temperature, pain, pressure, and temperature control may be impaired. All these senses play a role in organizing how we take in information both from outside and inside one’s own body. Individuals may also have difficulty regulating their sleep/wake cycle, hunger cycle, their ability to control emotions, and their ability to plan motor activities. Sensory issues can cause stress leading to unexpected outbursts or passivity in students.

Behavioral problems may also be caused by a child’s frustration of not being able to effectively

communicate. It is also important to be aware that changes in behavior can often be caused by pain. Parents and teachers need to work together to determine an effective means of communication for home and school and determine what triggers may be affecting their behavior. Sensory triggers of behavioral issues can include

- Sensory processing issues
- Sensory deprivation
- Sensory overload (too much noise, light, activity, etc.)
- Physical discomfort, e.g. uncomfortable chair or body position
- Problems with glasses or hearing aids
- Fatigue

Possible Physical Outcomes of Sensory Challenges

- Sensory integration dysfunction may be evident in many areas for some children
 - Inability to chew and bite into foods
 - Extreme postural insecurity
 - Enjoy swinging and bouncing
 - High pain threshold
 - Delayed bowel and bladder movements
 - Disturbed and inconsistent sleep patterns
- Problems with balance will inhibit the development of
 - Postural control
 - Equilibrium
 - Muscle tone and motor coordination
 - May have tactile defensiveness
- Fatigue

Behavior

Some individuals will have challenging behaviors that will vary from mild to more intense

- Perseverative behavior in younger children (repetitive or stuck on one thing)
- Obsessive-compulsive disorder in older children and adults
- Higher level of anxiety or nervousness possible
- Attention problems
- Executive function disorder
- Attention deficit hyperactivity disorder
- Pervasive developmental disorders
- Autistic-like behaviors
- Tic disorder
- Tantrums or aggressive outbursts
- Inflexible behavior (changes to schedule or routine)
- Poor self-regulation
- Self-stimulatory behavior is often a way of getting the body reorganized
 - Self-biting
 - Scratching, skin picking
 - Spinning, rocking

- Hand flapping

Social Impacts of Sensory Challenges

- Difficulty sharing or understanding other's point of view
- Difficulty in expressing self can lead some children to give up trying or to have explosive behaviors
- Passivity or refusal to cooperate

What you can do

Individuals may need help to reduce their stress and learn strategies for adapting. Children with diagnosed mental health conditions may be prescribed medications if appropriate.

Self-Regulation Tips

- Encourage them to talk about fatigue, anger, boredom, and restlessness
- Give them words (signs) for what they are feeling so they can learn to express it
- Encourage them to alternate between active and sedentary activities
- Consider switching to easier, fun activities when frustrated
- Take a sensory break and plan rest time

Sensory Strategies

- Sensory breaks can help children cope with sensory overload
- Deep tissue massage
- Brushing techniques
- Weighted clothing and blanket
- Joint compression
- Adapted furniture and other mobility support
- Bean bag chair
- Cushion on floor
- Magazines and books in a comfortable space
- Quiet room with low lightening
- Swings designed for sensory issues

Social Skills Strategies

- Ensure they have effective means of communicating
- Give them words (signs) for what they are feeling so they can learn to express it
- Foster peer-to peer interactions
- Model how to be part of a group, take turns, etc.

Physical Activity, Trips, Events

What you need to know

Physical activity

Exercise and physical education should be encouraged for strength building and obesity prevention. Certain sports may be difficult with vision/hearing limitations.

Field trips

- Any change in routine may produce anxiety, fears, and/or worry
- Crowds or loud noise may produce anxiety
- If a child has any sensory, hearing or vision issues, he/she may need preferred seating

What you can do

- Offer anticipatory guidance and preparation to prepare for a change in routine, such as a field trip.
- Create a picture story about the upcoming event. The child can rehearse it alone or with others.
 - Encourage use of their communication system to help them process concerns.
- If there is to be a lot of walking, it is important to consider that it will take more time and individuals will be tired.
 - Consider cutting down on walking when possible.
 - Use alternative forms of transportation if necessary.
- Offer supports as needed for vision and hearing issues.
- Make sure you have a plan to manage seizures or any other medical conditions if they are present.

School Absences and Fatigue

What you need to know

- There may be frequent absences due to illness and/or medical appointments.
- Sleep disorders are common.
 - Children with CHARGE may be tired and require rest opportunities or breaks in their day

What you can do

- Help to make transitions in and out of school as seamless as possible
- Be aware of fatigue and let parents know about fatigue issues affecting school

Emergency Planning

- Develop an emergency plan if necessary, depending on the needs of individual children.
- Include plans for managing seizures and emergency medications if they are part of the child's needs.

Resources

Medline Plus

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide a good medical overview of the condition without being too technical.

<https://medlineplus.gov/genetics/condition/charge-syndrome/>

CHARGE Syndrome Foundation

The CHARGE Syndrome Foundation is a nonprofit organization devoted to increasing awareness, resources, and research on the condition. Their website has lots of valuable information. <http://chargesyndrome.org/>. Educators may find this link particularly helpful

<https://www.chargesyndrome.org/for-professionals/education-professional-packet/>

Perkins School for the Blind

Perkins School for the Blind is committed to education and independence for those who are blind, visually impaired, and deafblind. They have several videos, articles and other resources for those with CHARGE syndrome. Go to their resources page and type CHARGE in the search bar. <https://www.perkins.org/resource-center/>

National Center on Deafblindness (NCDB)

This organization works to improve the quality of life for children who are deaf-blind and their families. They have helpful information for families and educators to meet the unique needs of these children. <https://www.nationaldb.org/>

Info for School Nurse and Primary Care Staff

CHARGE syndrome is a complex condition that will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student's team. Work with the parents to get signed releases to share information at the school nurse's office and the primary care office.

Gene Reviews is a peer-reviewed point-of-care reference accessible via the National Library of Medicine. The entry for CHARGE Syndrome provides a comprehensive summary of the condition and describes some ongoing surveillance and management issues. It is a great resource for medical providers. <https://www.ncbi.nlm.nih.gov/books/NBK1117/>

HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, remind the parents to sign a release of information form at the primary care office. The school nurse should keep a copy of that release in the student's file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

Example letter for physician to provide to school

"My patient _____ has been diagnosed with CHARGE syndrome, a rare genetic condition which affects multiple organ systems. This condition is characterized by visual, hearing, and other sensory deficits. Children with CHARGE may have difficulties with breathing, swallowing, and other motor tasks. Some children have medical complications like seizures or heart defects. It is likely this child may have increased absences for medical reasons. This condition necessitates an Individualized Education Program (IEP) or 504 plan.

Meet a Young Woman with CHARGE Syndrome : *Tricia!*



When you meet Tricia, you immediately tune into her warm smile, which is often followed by a contagious giggle! She is an energetic and lovely young woman, with a big sense of humor and matching spirit of fun. Tricia lives in a small New England town, and she, her family, and team have worked to create a good life for her, full of meaning, new experiences and ever-increasing independence.

Now 27, Tricia originally started her adult years involved in a program run by a vendor agency. She often participated in community outings, but her family didn't feel she was learning as much as she needed about living independently. Her family wondered, "What could we do differently to create a better, more stimulating and independent life for Tricia?" The answer was to design their own life plan with Tricia's input and run this "program" themselves. They all created goals that match Tricia's interests and needs, hired their own people to help ensure these goals could be realized, and have been working for the past three years to make this a strong, meaningful and ever-evolving life with Tricia leading the team.

Enter Martha, a former teacher. Martha is part of a team set up to support Tricia in developing life skills to further her independence, and she oversees all components of Tricia's new plan. They include: improving Tricia's verbal and written communication, staying organized, planning and cooking meals, and honing her social/soft skills. Tricia has become more proficient in writing, using her iPad, keeping track of time and money, and using a variety of technology to stay current and communicate effectively.

Then came Becky, Ashley, and Brianna (all are graduates of a local university in Sign Language Interpreting). Brianna currently supports Tricia in using her signing skills while working on a rigorous curriculum. By signing, Tricia can really "see" meanings and subtleties of language that aren't as easy to learn due to her hearing difficulties. Together with Brianna, Tricia assists ASL students in high school classes and attends a weekly deaf social hour where she enjoys interacting with young adults.

Tricia has been working at the local Walgreen's for 8 years, earning not only a paycheck but recognition as Employee of the Month. As another part of her program, Tricia is a volunteer at a local high school and elementary school. Along with her work in ASL classes, Tricia runs the snack cart and does specific tasks in the library, guidance, and athletic offices. This exposure to new activities and a wide range of people serves to increase Tricia's confidence, skills, independence and all-around enjoyment.

Donna, a third team member, facilitates Tricia's volunteer work for a care-giving organization in

her community, helping to deliver personal care to an elderly woman who lives at home alone. Tricia does this with another young woman, and they have developed a friendship outside of work. She and Tricia look forward to their weekly outings - going out to lunch, seeing a movie or going to the mall.

When enjoying some leisure time, Tricia rides her bike around the neighborhood, reads, watches closed captioned movies, and performs with a play group, putting on two variety shows a year. Tricia competes in Special Olympics, excelling at track and field, as well as golf (a talent which runs in the family), and recently had the honor of signing the National Anthem during the opening ceremonies of the State Games.

With this new life plan in place and working so well, Tricia has become a more confident woman, a stronger communicator, and a more self-sufficient and involved member of her community.

Thank you so much to Tricia and family for sharing their story! And thank you to the team at the Institute on Disability at UNH for assisting in collecting these stories.