

CHARGE Syndrome *At a Glance*

CHARGE syndrome is a pattern of differences seen at birth. CHARGE stands for **C**oloboma, **H**ear defects, **C**hoanal **A**tresia, **R**etarded growth and development, **G**enital abnormalities, and **E**ar anomalies.

The effects of CHARGE syndrome are different for each person, and most often, there is no family history of CHARGE. At birth, some babies diagnosed with CHARGE have life-threatening issues. Most children will have hearing loss, vision loss, and balance problems. Individuals often have multiple physical, sensory, and behavioral differences.

About 1 in every 9-10,000 babies is born with CHARGE syndrome.



Meet Tricia on page 17.

Things to think about:

1. Medical/Dietary Needs

What you need to know

Medical Needs

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 doctors and specialist visits to monitor medical conditions.

Regular neurodevelopmental and developmental/behavioral evaluations may be helpful.

Dietary Needs

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- Feeding and swallowing difficulties
 - The causes and severity vary greatly
 - Swallowing mechanism may be affected
 - May be due to abnormalities of the cranial nerve
 - May be secondary to weakness and motor planning disorders
- Sensory difficulties can contribute to feeding and swallowing challenges
 - Tactile defensiveness
 - Oral hyposensitivity

Physical characteristics and/or symptoms:

Not all people with CHARGE have all of these characteristics.

Common Characteristics

Eye

- Coloboma (missing pieces of tissue in structures that form the eye) (80-90%)
 - Appear as notches or gaps in one of several parts of the eye, including
 - **Iris** (colored part of the eye) iris
 - **Retina** (specialized light-sensitive tissue that lines the back of the eye)
 - **Choroid** (the blood vessel layer under the retina)
 - **Optic nerves** (carry information from the eyes to the brain)
 - More severe forms include microphthalmos (small eye) or anophthalmos (missing eye)

Heart defects (75-85%)

- Many are complex heart defects requiring more than one surgery

Narrowed or blocked passages between the nose and the throat

- Choanal atresia (narrowed or blocked passages between the nose and the throat) (50-60%)
 - Can be on one side or both side

Cranial nerve abnormality

- Cranial nerve I (One) (90-100%)
 - Leads to missing or decreased sense of smell
- Cranial nerve IX/X (Nine or Ten) (70-90%)
 - Leads to swallowing difficulties and possibly aspiration
- Cranial nerve VII (Seven) (40%)
 - Leads to facial palsy (one side or both)

Ear abnormalities

- Outer ear anomalies (>50%)
 - Often a typical "CHARGE" ear that is short and wide with small lobes
 - Inner ear folds are prominent and the outer folds appear "snipped off" (for an illustration, go to this link: <http://www.ncbi.nlm.nih.gov/books/NBK1117/figure/charge.F1/?report=objectonly>)
- Middle ear anomalies (common)
 - Bones of the middle ear are malformed
 - Causes conductive hearing loss
 - Prevalence of severe to profound hearing loss is 50%
- Inner ear anomalies (90%)
 - Malformed cochlea and/or small or absent semicircular canals
 - Can cause hearing loss and balance problems

Low muscle tone

- Hypotonia (low muscle tone) (90%)

Less Common Characteristics

Cleft lip

- Cleft lip with or without cleft palate (20%)

Opening between trachea and esophagus

- Tracheo-esophageal fistula (abnormal opening between trachea and esophagus) (15-20%)
 - Esophageal atresia (esophagus is not open)

Kidney abnormalities (40%)

- Small kidney
- Missing kidney
- Misplaced kidney
- Reflux

Genital abnormalities

- Males (50%)
- Females (25%) Both may have lack of puberty without hormone intervention (90%)

Growth

- Growth deficiency (15%)

Short stature (70%)

- 70% have short stature

Typical CHARGE facial features

- Square face
- Broad prominent forehead
- Arched eyebrows
- Large eyes
- Occasional droopy lids
- Prominent nasal bridge and thick nostrils
- Prominent nasal columella (space between nostrils)
- Flat midface
- Small mouth
- Small chin
- Facial asymmetry with or without facial palsy

Typical behavior (more than 50%)

- Perseverative behavior in younger children (repetitive or stuck on one thing)
- OCD in older children and adults

Other Findings

- Chronic ear problems (85%)
- Sloping shoulders (common)
 - Underdeveloped shoulder muscles
- Small or missing pectoral (high in chest) muscles
- Short neck
- Limb/skeletal anomalies
 - Absent thumb
 - Extra fingers
 - Vertebral abnormality
- Central Nervous system (CNS) abnormalities:
 - Hydrocephalus
 - Seizures
- Umbilical hernia (15%)
- Nipple anomalies (occasional)
 - Extra, missing, or misplaced nipples
- Scoliosis (common)

What you can do

- A yearly check-up and studies as needed should occur in the child's Medical Home.
- 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.
- Work with parents to ensure the child is fed in a way that is best for the child.
- If there are seizures, make sure you have a seizure care plan.

2. Education Supports

What You Need to Know

Individuals with CHARGE syndrome often have multi-sensory challenges. They often have difficulties with vision, hearing, and the senses that perceive balance, touch, temperature, pain, pressure, and smell. Intelligence may be underestimated because of their vision, hearing, 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. Members of this team may include:

- Physical therapy
- Occupational therapy
- Speech therapy
- Sensory integration program specialist
- Deaf or Deafblind specialist
 - Most states have access to a deaf blind specialist through the state deaf-blind project. Please see <https://nationaldb.org/members/list?type=State+Project>.

Communication

- Individuals with CHARGE may have challenges in developing clear speech and/or the ability to sign well.
 - 60% acquire symbolic language and communicate with spoken language, signs, and or visual symbols.
 - Speech may be affected by craniofacial and breathing problems.
 - Language delays are caused by multiple sensory and motor challenges, and delays in finding an appropriate communication system.

- Bilateral facial palsy and central vision loss can lead to lack of facial expression and this can add to individual's communication problems.
- Most individuals benefit from a total communication approach.
 - Total communication approach means incorporating anything that works including:
 - Gestures
 - Simple signs
 - Braille/print
 - Facial expressions
 - Symbols (PECS or picture exchange communication system)
 - Speech therapy
 - Sign language
- Balance challenges can affect communication
 - Development of memory
 - Effective use of vision
 - Processing of auditory input
 - All of these can have cumulative effect on speech and language development.
- Challenges to speech include:
 - Hearing impairment
 - Vision impairment
 - Facial palsy
 - Low muscle tone
 - Poor tactile sense
 - Oral facial cleft
 - Enlarged tongue
 - Poor tongue movement
 - Small lower jaw
 - Larynx and pharynx anomalies
 - Breathing difficulties
 - Swallowing difficulties
 - Dental abnormalities
 - Delayed/immature eating skills
- Challenges to signing:
 - Under-functioning tactile and proprioceptive sense
 - Low muscle tone
 - Severe balance problems
 - Dyspraxia

- Initiating speech/signs may be due to specific brain anomalies

Hearing

- An audiological evaluation can be challenging because
 - Very young children don't speak or sign
 - Visual problems interfere with sound field testing
 - Tactile defensiveness is common
 - Hearing loss may be great and asymmetrical
 - Resistance and risks to sedation may exist
 - Ear anomalies may make it more difficult to fit ear molds and hearing aids
 - Narrow ear canals can block from fluid accumulation. This, combined with congested breathing, leads to noisy breathing. This makes it harder to hear.
 - Conductive hearing loss
 - Middle ear malformations
 - Sensorineural loss because of malformation of cochlea

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
- Hearing impairments and vestibular (balance) abnormalities affect the amount and quality of information received from the environment
- Equilibrium triad (vision sense, balance sense, tactile/ proprioceptive senses) may be missing or impaired.

Balance

- Malfunctioning or absent semicircular canals (the receptors of balance sense) in the inner ear canals.
 - This plays a crucial role in organizing sensory inputs in all other sensory channels
 - This may affect walking and standing
- Postural security and good sense of equilibrium depends on the development of vision sense, balance sense, tactile, and proprioceptive senses

- 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
- Balance and vision
 - 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
 - May be reluctant to go outside because these “markers” don’t exist outside
- Balance and Hearing
 - There is a link between balance sense and ability to process sound and to develop spoken language

Sensory (see also Behavior and Sensory Supports)

Some children may experience sensory difficulties.

- Sensory impairment is caused by impaired and poorly modulated sensory systems
- Sensory breaks can help children cope with sensory overload
- Sensory integration dysfunction may be evident in many areas for some children
 - Rejection of textures in mouth
 - Inability to chew and bite into foods but may grind teeth
 - Extreme postural insecurity
 - Enjoy swinging and bouncing
 - High pain threshold
 - Delayed bowel and bladder movements
 - Disturbed and inconsistent sleep patterns
 - Self-stimulation behaviors
 - Self-biting
 - Scratching
 - Skin picking
 - Spinning
 - Rocking
 - Hand flapping

Motor delays

- Could occur due to vestibular dysfunction and/or prolonged hospitalization

- May also have
 - Low muscle tone in trunk/core
 - Balance problems
 - Reduced sensory input
 - Can lead to reduced perceptual awareness
 - Can have lack of motivation to move
 - May be resistant to exercise
 - Low tone with poor tactile senses may cause a child to use excessive force in movement
 - May be perceived as aggressive, rough, or clumsy
 - Flexible joints
 - Poor vision
 - Severe balance problems
 - Breathing difficulties
 - Reduced perceptual awareness

What you can do

The CHARGE Syndrome Foundation produced a Professional Packet that you may find helpful, in addition to the ideas below (<http://chargesyndrome.org/resources-professionalpacket.asp>).

Vision

- Consult Deafblind specialists
- Make accommodations for safety
 - i.e., visual enhancement, like bright duct tape on a step
- Field loss or monocular vision
 - May have difficulty to follow a line(s) on paper
 - Makes reading difficult
 - Use large bold print
 - Use bold lines
 - Use underlining or high-lighter
- Photophobia (intolerance to light)
 - Tinted glasses
 - Sun visor
 - Low classroom lighting
 - Rest breaks in a dark room

General strategies

- 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
 - Break assignments into small tasks
- Establish 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
- Sensory break
 - Allow pause time during and between activities
- Curriculum:
 - Child centered curriculum
 - Expanding environment
 - Start small and expand as child is comfortable
- Social skills:
 - Individuals may need help to
 - Learn how to be a part of a group
 - How to negotiate
 - How to take turns and share
 - How to help
- Cognitive skills: Help them to:
 - Be organized

- Anticipate activities
- Cope with behaviors
- Make choices

Sensory Strategies

- Deep tissue massage
- Brushing techniques
- Weighted clothing and blanket
- Joint compression
- Bean bag chair
- Cushion on floor
- Magazines and books in a comfortable space
- Quiet room with low lightening
- Swings designed for sensory issues
- Need a flexible schedule for breaks
- Break tasks into smaller steps
- Give choices as much as possible
- Clear expectations and firm limits
- Adapted furniture
- Consistent routines

Social

- Foster peer-to-peer interactions
- Help with effective communications
- Model how to be part of a group, how to take turns in games and conversation

3. Behavior and Sensory Support

What You Need to Know

(See also Education supports for more information on sensory issues.)

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, smell, breathing and swallowing, eating, drinking, digestion, and temperature control maybe impaired. All of these senses play a role in organizing how one takes 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.

Behavioral problems may also be caused by a child's frustration of not being able to effectively communicate. 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. It is also important to be aware that changes in behavior are often due to pain, which can be difficult to identify in children with poor communication skills.

- Sensory issues can cause stress leading to unexpected outbursts or passivity in students.
- Sensory problems may be caused by
 - Too much noise
 - Glare or too much light
 - Too much movement or distraction in the environment
 - Problems with glasses or hearing aides
 - Fatigue
 - Seating issues
 - Uncomfortable chair or body position
 - Sensory deprivation
 - Sensory overload
 - Sensory processing

Sensory challenges

- Semicircular canals play a role in organizing sensory perception through all sensory channels.
- Problems with balance will inhibit the development of effective body language.
 - Postural control
 - Equilibrium
 - Muscle tone and motor coordination
 - May have tactile defensiveness
- Difficulty in expressing self can lead some children to give up trying or to have explosive behaviors
- Later childhood and adolescence problems:
 - Fatigue
 - Difficulty maintaining postural control

Behavior

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

- Individuals may need help to reduce their stress and to be instructed in strategies for adapting
- Behavior problems could occur. Some examples may include:
 - Attention problems
 - Self-stimulatory or self-abusive behavior is often a way of getting the body reorganized
 - Self-biting
 - Scratching
 - Skin picking
 - Spinning
 - Rocking
 - Bouncing
 - Hand flapping
 - Obsessive-compulsive behavior
 - Tantrums or aggressive outbursts
 - Passivity or refusal to cooperate Give them words (signs) for what they are feeling so they can learn to express it.

Social skills problems

- Difficulty sharing or understanding other's point of view
- Problems with the sleep cycle are common

Behaviors seen in some children who have CHARGE

- Higher level of anxiety or nervousness possible
- Repetitive questions about same topics
- Inflexible behavior
- Upset with change in schedule or routine
- Impulsive
- Obsessive
- Self-stimulation
- Poor self-regulation
- Executive function disorder
- Attention deficit hyperactivity disorder
- Pervasive developmental disorders
- Obsessive-compulsive disorder
- Autistic like behaviors

- Tic disorder

What you can do

Self-regulation

- Talking about fatigue, anger, boredom, and restlessness
- Give them words (signs) for what they are feeling so they can learn to express it.
- Alternate active with sedentary activities
- Challenging with easier, fun activities.
- The child may not be a good judge of fatigue
- Impose sensory breaks and schedule rest time

May require an alternative space

- May have difficulty sitting or standing without supports
- Adaptive chairs may help
- Some can sit at desk but may need a break in a horizontal position to relax
- Bean bag chairs
- Soft cushion on floor
- Quiet space
- Low lighting
- Weighted blanket or vest

4. 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.
- If a child has any sensory, hearing or vision issues, he/she may need preferred seating.
- Crowds or loud noise may produce anxiety

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 planned for seizures or any other medical conditions if they are present.

5. School Absences and Fatigue

What You Need to Know

- Children with CHARGE may be absent due to illness and/or medical appointments.
- Children with CHARGE may have sleep disorders
 - Their sleep may be disturbed and inconsistent
 - 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

6. Emergency Planning:

What you need to know

- 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.

7. Resources:

The CHARGE Syndrome Foundation

Information and resources for parents and professionals

<http://chargesyndrome.org/>

<http://chargesyndrome.org/resources.asp>

National Consortium on Deafblindness (NCDB)

This organization works to improve the quality of life for children who are deaf-blind and their families.

<https://nationaldb.org/>

GeneReviews

Learn more about the genetics of CHARGE

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

Genetic Home Reference

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

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

Perkins School for the Blind

This site brings together the range of web-based content available through Perkins School for the Blind for teachers, families, and others interested in the education of children who are blind or visually impaired, including those with deaf blindness or additional disabilities.

<http://www.perkinselearning.org/webcast/charge-syndrome-impact-charge-communication-and-learning>

Classroom Accommodations for Students with Visual Issues

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

8. Meet 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 pay check 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 office. 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.