Indexing Metadata/Description

› **Title/condition:** CHARGE Syndrome: Communication Impairments

› **Synonyms:** (Communication impairments) speech impairment; speech disorder; speech delay; speech deficit; language impairment; language disorders; language delay; language deficit; communication delay; communication disorder; communication deficit

› **Anatomical location/body part affected:** Multiple structures used for communication can be affected in a person with CHARGE syndrome (auditory structures, oral structures, pharynx, larynx, respiratory system, cognitive ability)

› **Area(s) of specialty:** Pediatric Genetic and/or Neurological Disorders, Craniofacial and/or Oral Motor Abnormalities

› **Description:** CHARGE syndrome is a genetic condition marked by a specific pattern of major and minor birth defects. It is the leading genetic cause of deaf-blindness at birth in the United States.\(^2,37\) The features of CHARGE syndrome considered critical in diagnosing and defining the syndrome include coloboma of the eye (hole in the lower part of the iris, retina, and/or optic nerve that creates a void in the visual field), congenital heart malformation, choanalatresia (blockage of the breathing passages), delayed growth and development, dysfunction of cranial nerves (I, VII, VIII, IX, X), genital anomalies, and external ear and middle ear anomalies (pinnae are soft and floppy and appear stretched and cupped, ossicular malformations, absence or dysplasia of lateral semicircular canals).\(^1,2,38,39\) The acronym CHARGE is based upon the cardinal features (Coloboma, Heart malformation, Atresia, Retardation of growth, Genital anomalies and Ear anomalies).\(^3\) Minor features are those that occur less frequently and/or are also characteristic of other syndromes and are not unique to CHARGE: cleft lip/palate, hypotonia, renal anomalies, tracheoesophageal fistula or atresia, esophageal atresia, and short stature.\(^1,2,40\)

› **ICD-9 codes**
  • 759.89 CHARGE association/syndrome
  • Hearing loss
    – 389.06 conductive hearing loss, bilateral
    – 389.1 sensorineural hearing loss
    – 389.22 mixed bilateral conductive and sensorineural hearing loss
    – 389.9 unspecified hearing loss
    – 315.34 speech and language developmental delay due to hearing loss
  • Blindness
    – 369.0 profound impairment, both eyes
    – 369.1 moderate or severe impairment, better eye, profound impairment lesser eye
    – 369.2 moderate or severe impairment, both eyes
    – 369.3 unqualified visual loss, both eyes
    – 369.4 legal blindness, as defined in United States

› **ICD-10 codes**
  • H54 blindness
  • H90.3 sensorineural hearing loss bilateral
  • H90.0 conductive hearing loss, bilateral
G-Codes

• Swallowing G-code set
  – G8996, Swallowing functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals
  – G8997, Swallowing functional limitation, projected goal status, at initial therapy treatment/outset and at discharge from therapy
  – G8998, Swallowing functional limitation, discharge status at discharge from therapy/end of reporting on limitation

• Motor Speech G-code set
  – G8999, Motor speech functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals
  – G9186, Motor speech functional limitation, projected goal status, at initial therapy treatment/outset and at discharge from therapy
  – G9158, Motor speech functional limitation, discharge status at discharge from therapy/end of reporting on limitation

• Spoken Language Comprehension G-code set
  – G9159, Spoken language comprehension functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals
  – G9160, Spoken language comprehension functional limitation, projected goal status, at initial therapy treatment/outset and at discharge from therapy
  – G9161, Spoken language comprehension functional limitation, discharge status at discharge from therapy/end of reporting on limitation

• Spoken Language Expressive G-code set
  – G9162, Spoken language expression functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals
  – G9163, Spoken language expression functional limitation, projected goal status, at initial therapy treatment/outset and at discharge from therapy
  – G9164, Spoken language expression functional limitation, discharge status, at discharge from therapy/end of reporting on limitation

• Attention G-code set
  – G9165, Attentional functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals
  – G9166, Attentional functional limitation, projected goal status, at initial therapy treatment/outset and at discharge from therapy
  – G9167, Attentional functional limitation, discharge status at discharge from therapy/end of reporting on limitation

• Memory G-code set
  – G9168, Memory functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals
  – G9169, Memory functional limitation, projected goal status, at initial therapy treatment/outset and at discharge from therapy
  – G9170, Memory functional limitation, discharge status at discharge from therapy/end of reporting on limitation

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<thead>
<tr>
<th>G-code Modifier</th>
<th>Impairment Limitation Restriction</th>
</tr>
</thead>
<tbody>
<tr>
<td>CH</td>
<td>0 percent impaired, limited or restricted</td>
</tr>
<tr>
<td>CI</td>
<td>At least 1 percent but less than 20 percent impaired, limited or restricted</td>
</tr>
<tr>
<td>CJ</td>
<td>At least 20 percent but less than 40 percent impaired, limited or restricted</td>
</tr>
<tr>
<td>CK</td>
<td>At least 40 percent but less than 60 percent impaired, limited or restricted</td>
</tr>
<tr>
<td>CL</td>
<td>At least 60 percent but less than 80 percent impaired, limited or restricted</td>
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</tbody>
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Reimbursement: Reimbursement for therapy will depend on insurance contract coverage; no specific issues or information regarding reimbursement have been identified.

Presentation/signs and symptoms: Communication development in individuals with CHARGE syndrome can be affected by multiple factors, including hearing and vision loss, cognitive disorders, developmental delay, severe neurologic involvement, craniofacial anomalies (e.g., cleft palate, vocal fold paralysis, facial palsy), physical anomalies (e.g., low muscle tone), and prolonged hospitalization. Children with CHARGE syndrome who have tracheostomy tubes have difficulty producing audible and intelligible speech. A large proportion of individuals with CHARGE syndrome do not acquire symbolic language, the ability to understand and use abstract representations of concepts, and rely on pre-symbolic communication (e.g., gestures, behaviors, picture boards). Most children with CHARGE syndrome also have swallowing and feeding disorders (for detailed information about dysphagia in individuals with CHARGE syndrome, see Clinical Review...Feeding Disorders: CHARGE Syndrome; CINAHL Topic ID Number: T708992).

Causes, Pathogenesis, & Risk Factors

Causes
- CHARGE syndrome has an established genetic basis, arising from mutations in a member of the homeodomain gene family, the CHD7 gene.

Pathogenesis
- Most children with CHARGE syndrome have communication difficulties from infancy. The severity and type of communication difficulties will vary with the individual.

Risk factors (for reoccurrence CHARGE syndrome)
- If one parent has CHARGE syndrome, the reoccurrence risk is 50%.
- If a parent has had a child with CHARGE syndrome, the reoccurrence risk is usually the same as in the general population (1 in 10,000, or .01%).
- If a parent has had a child with CHARGE syndrome and has a genetic condition called “germline mosaicism” (the existence of 2 or more populations of cells with different genotypes in a person who has developed from a single fertilized egg), the reoccurrence rate is about 6%.

Risk factors (for severe communication difficulties associated with CHARGE syndrome): Characteristics that adversely affect communication in children with CHARGE syndrome include physical disorders, intellectual disability, vision loss, and hearing loss. A large proportion of individuals with CHARGE syndrome do not acquire symbolic language, the ability to understand and use abstract representations of concepts. In a study of 28 children with CHARGE syndrome conducted in the United States, researchers investigated factors related to the development of symbolic communication. Factors that adversely affected the development of symbolic language included lack of success with hearing amplification (e.g., uncorrected hearing loss, active middle ear disease, late fitting for hearing aids), lack of early intervention (before 3 years of age), and inability to walk independently.

Overall Contraindications/Precautions

Infant mortality is a serious concern in early development due to heart conditions. Parents are often focused on infant survival and surgical procedures during the first few months of life rather than with other aspects of development.

During assessment and treatment, the speech-language pathologist (SLP) must consider the space in which a speaker or signer can be seen or heard optimally by the individual with CHARGE syndrome (“communication bubble”). Lighting and environmental noise must be controlled.

Symptoms associated with CHARGE syndrome vary, and precautions will differ according to the individual patient and the severity of accompanying language, cognitive, vision, hearing, and/or motor impairments.
Examination

Contraindications/precautions to examination

- Development of an individualized family services plan (IFSP; ages 0-3) or individualized education plan (IEP) in collaboration with the parents/caregivers and school personnel for school-aged individuals with CHARGE syndrome is essential to address communication and/or swallowing problems in the school environment.
- Assessment times may be longer with children with multiple sensory deficits than other children because it is critical to promote a positive interaction through nonvisual and nonauditory channels before beginning the evaluation. The following are strategies to facilitate interaction before and during the evaluation:
  - To identify oneself as an interaction begins, touch a nonthreatening part of the child’s body (hand, top of the wrist). Ask the child’s teacher/family how the patient is typically approached.
  - Show the child what will happen by placing an object under the patient’s hand (e.g., toy from an assessment).
  - Parents can supply information about preferred interactions for their child. Use activities that the child prefers for initial interaction (e.g., rocking together in a chair). When possible, observe child interacting with parent.
  - Never grab or restrain the child.

History

- History of present illness/injury
  - Course of treatment
    - Medical management: Medical management will vary according to the patient’s presenting symptoms.
    - Medications for current illness/injury: Determine what medications the physician has prescribed; are they being taken?
    - Diagnostic tests completed: Diagnosis of CHARGE syndrome is made by a geneticist or physician familiar with CHARGE and is based upon one of the following criteria:
      - Clinical criteria: The presence of three major features of the syndrome OR the presence of two major features and three minor features of the syndrome.
      - Genetic criteria: Identification of a mutation of the CHD7 gene.
    - To assess communication in individuals with CHARGE syndrome, the SLP can use:
      - Direct observation of the individual in a natural environment and in an interaction with a parent (e.g., home).
      - Parent questionnaires and interviews.
      - Interviews with educational staff.
      - Adapted tests.
  - Home remedies/alternative therapies: Document any use of home remedies or alternative therapies and whether or not they help.
  - Previous therapy: Document whether patient has had occupational, speech, or physical therapy for this or other conditions and what specific treatments were helpful or not helpful.
  - Aggravating/easing factors
    - What is the nature of the individual’s sensory deficits? Patients with CHARGE syndrome can have deficits in hearing, vision, touch, smell, balance, and pain.
    - Does the patient or the patient’s family speak another language?
    - What modality does the patient use to communicate?
    - Is the patient currently being fed by mouth, or is the child tube fed (orogastric, nasogastric, gastronomy, jejunostomy, or a combination)?
  - Body chart: Use body chart to document location and nature of symptoms.
  - Nature of symptoms: Speech and language development are affected by the multisensory impairment in patients with CHARGE, and children with CHARGE show a range of communicative abilities, from pre-symbolic communication to developed conversational skills.
  - Rating of symptoms: Use a visual analog scale (VAS) or 0-10 scale to assess symptoms at their best, at their worst, and at the moment.
  - Pattern of symptoms: Document changes in symptoms throughout the day and night, if any (AM, mid-day, PM, night); also document changes in symptoms due to other external variables such as communication partner, and noise in the environment.
– **Sleep disturbance**: Document number of wakings/night. Individuals with CHARGE have been reported to have apnea(1)
  - In a questionnaire-based study that surveyed 44 families in the United Kingdom with children who had CHARGE syndrome, researchers found that 30% of the children in this study required medications to treat sleep disorders(33)

– **Other symptoms**: Document other symptoms patient is experiencing that could exacerbate the condition and/or symptoms that could be indicative of a need to refer to physician, psychiatrist, or social worker. Many children with CHARGE syndrome have swallowing and feeding disorders. Other common symptoms include bowel/bladder dysfunction, urinary tract infections, abdominal pain, anxiety disorder, self-abuse, and balance/equilibrium impairment(2)

– **Respiratory status**: Note history of respiratory status and current respiratory status for any indication of aspiration. Note any history of respiratory disorder

– **Psychosocial status**: Document the patient’s psychosocial status; obtain information from the patient’s parents and/or caregivers
  - Many patients with congenital deaf-blindness have a positive outlook on life with moderate to high levels of social engagement.(8) Given the significant and unique challenges that exist for patients with congenital deaf-blindness, these patients are at increased risk for social disengagement.(8) (For detailed information on congenital deaf-blindness, see [Clinical Review...Deaf-Blindness, Congenital; CINAHL Topic ID Number: T708768])
  - Behavior in individuals with CHARGE syndrome is greatly influenced by sensory deficits and communication ability. Behavior can be viewed as a form of communication when the individual has presymbolic communication and/or is unable to communicate with gesture. Common behaviors include repetitive behaviors, self-stimulatory behaviors, maladaptive routine behaviors, OCD, attention deficit disorder (ADD), tactile defensiveness, and autistic-like behaviors (e.g., self-stimulation, self-injurious behaviors, withdrawal, perseveration)(1)
  - A CHARGE “behavioral phenotype” has been proposed to describe behaviors that are reliably associated with CHARGE syndrome:
    - Low normal cognitive functioning(34)
    - Goal-directed and persistent behavior(34)
    - Possessing a sense of humor(34)
    - Interested in interacting with others but socially immature(34)
    - Stress-induced repetitive behaviors(34)
    - High levels of sensation-seeking behaviors(34)
    - Difficulty with self-regulation during times of stress or sensory overload(34)
    - Difficulty shifting attention and transitioning to new activities(34)
  - If there are concerns regarding behavioral disturbance or psychosocial status, refer the patient/family to a mental health professional or behavioral specialist

– **Hearing**: Hearing loss is present to some degree (often severe to profound) in almost all individuals with CHARGE syndrome and can be conductive, sensorineural, or mixed.(1,36,38,39) In rare cases, normal hearing abilities have been reported.(36) Approximately 80-90% of individuals with CHARGE syndrome experience both hearing and vision loss.(37)
  - Note the results of audiological reports and otologic treatment and refer to an audiologist if information is incomplete or outdated or if the child shows a change in hearing (e.g., as the result of a middle ear infection). High variability has been noted between and among (different loss in different ears) individuals with CHARGE syndrome. Typically, hearing loss is mixed and in the moderately severe to severe range. Cochlear implants can improve both audiological and speech outcomes for some patients with CHARGE syndrome who present with a sensorineural hearing loss.(9,10,11)
  - Patients with CHARGE syndrome often have structural auditory anomalies that include:(1)
    - **External ear**: Pinnas are floppy, asymmetrical, and malformed. External auditory canals may be narrowed or absent, making it difficult to fit a hearing aid ear mold in the inner ear(1)
    - **Middle ear**: Ossicles might be malformed (e.g., stapes) and cause significant conductive hearing loss (typically greatest in the low frequencies). Chronic Eustachian tube dysfunction can cause chronic otitis media, resulting in perforated ear drums and drainage(1)
    - **Cochlea**: Sensorineural hearing loss can result from malformed cochlea and damaged or missing hair cells(1)
- Researchers in Germany investigated hearing and speech perception test results of 163 children with various underlying diagnoses who received cochlear implants between 1996 and 2008. The researchers found that children with CHARGE syndrome showed a wide range in performance on hearing and speech perception tests, and they attributed the extent of additional disabilities to the variation in performance. All of the participants with CHARGE syndrome performed poorly on speech perception tests even after 4 years of rehabilitation.

- Researchers in South Korea evaluated the audiological perception and speech intelligibility of 6 children with CHARGE syndrome who had received cochlear implants between 2002 and 2012. The mean age of the children was 4.9 years. Three children had typical intellectual ability and 3 others had mild to moderate intellectual disability. Researchers found that the hearing ability and speech intelligibility of 5 of the 6 children showed continuous improvement over time on all measures. They concluded that children with CHARGE syndrome, including patients with intellectual disability, can benefit from early intervention and cochlear implantation.

- Researchers in Australia evaluated the benefit of cochlear implants in 10 children with CHARGE syndrome. Seven of the children were prelingual profoundly deaf and had hypoplastic or absent auditory nerves bilaterally while 3 of the children had progressive sensorineural hearing loss and a normal auditory nerve in at least one ear. The children who were profoundly deaf were implanted between 8 months and 4 years of age. The children with progressive hearing loss were implanted between the ages of 13 and 16 years. Researchers found that the children with prelingual deafness continued to benefit from sign and oral language while the children with progressive hearing losses were able to continue to communicate using only spoken language. Further, researchers noted that due to the anomalies of the middle ear, preoperative considerations need to be made and some children would benefit from CT-guided implantation.

- **Auditory nervous system:** The auditory nerve is sometimes reduced in function or absent. Other anomalies include brain stem and/or auditory cortex (structures involved in auditory processing).

- **Barriers to learning**
  - Are there any barriers to learning? Yes ____ No ____
  - If Yes, describe __________________________

- **Medical history**
  - **Past medical history:** A complete medical and developmental history should be obtained as part of a comprehensive communication evaluation. Document any past as well as future planned surgical interventions. Information should also be gathered on fine motor development and speech and language development.

  - **Previous history of same/similar diagnosis:** Document any previous communication assessment and treatment.

  - **Comorbid diagnoses:** Ask patient or caregiver about other diagnoses or minor features of the disorder that can affect health, behavior, and communication: feeding difficulties, obsessive compulsive disorder (OCD), maladaptive behaviors, heart conditions, cleft lip/palate, hypotonia, tracheoesophageal fistula or atresia and/or esophageal atresia, and middle ear infections.

  - **Medications previously prescribed:** Obtain a comprehensive list of medications prescribed and/or being taken (including over-the-counter drugs).

  - **Other symptoms:** Ask patient or patient’s family about other symptoms he/she is experiencing.

- **Social/occupational history**
  - **Patient’s goals:** Document what the patient and important people in the patient’s life (i.e., family, school staff) hope to accomplish with therapy and in general.

  - **Vocation/avocation and associated repetitive behaviors, if any:** Things to consider include: Does the patient currently receive intervention services? If so, are they home, school, or clinic based? Does the patient attend daycare, school, or work? If the patient is in school, what support services are in place?

  - **Functional limitations/assistance with ADLs/adaptive equipment:** Obtain information on adaptive equipment the patient is using, such as assistive/alternativemunication devices, hearing aids, glasses, or adaptive eating equipment.

  - **Living environment:** With whom patient lives (e.g., caregivers, siblings). Identify if there are barriers to independence or communication in the home; any modifications necessary?

- **Relevant tests and measures:** (While tests and measures are listed in alphabetical order, sequencing should be appropriate to patient medical condition, functional status, and setting)

  - **Arousal, attention, cognition (including memory, problem solving):** The evaluating or treating SLP should note the results of any neurological (MRI, CT), neuropsychological, or psychological/cognitive tests that have been completed. Cognitive abilities of children with CHARGE syndrome are often underestimated due to hearing, vision, and communication difficulties. The Callier-AzusaScale allows for observation of the child in multiple areas of development.
using a checklist format.\(^{12}\) Other developmental scales such as the Bayley Scales of Infant and Toddler Development-3rd Edition can be adapted for use.\(^{13}\) Standardized scores from tests that have been normed on children without hearing and vision deficits are not valid for individuals with CHARGE. Parent interview combined with observations of the child within a natural environment and in interactions with the parent often yield the most valid information about cognitive abilities.\(^{1}\)

**Assistive and adaptive devices:** Document use of any adaptive devices for hearing (e.g., hearing aids, cochlear implants), vision, communication, feeding, body positioning, or mobility. Individuals with CHARGE syndrome sometimes have difficulty stabilizing behind-the-ear hearing aids on external pinnas due to “CHARGE ear.”\(^{4}\)

**Ergonomics/body mechanics:** Note any difficulties with positioning and body mechanics that may interfere with feeding. Collaborate with physical therapist (PT) and/or occupational therapist (OT) as necessary. Individuals with CHARGE often have hypotonia and have difficulty sitting upright for a long period of time (e.g., school).\(^{1}\) In addition, they may have poor vestibular function and difficulties with balance.\(^{1}\)

**Motor function (motor control/tone/learning):** Individuals with CHARGE are often severely delayed in motor development but have been found to achieve more advanced motor skills later in development.\(^{1}\) Mobility and symbolic language development has been found to be related in individuals with CHARGE syndrome.\(^{1}\) One reason might be that a patient with mobility has the advantage of self-positioning to receive maximum benefit during communicative interactions. Orientation and mobility (O&M) training should be integrated with communication as soon as possible. Collaborate with the PT and/or OT as necessary.

**Oral mechanism exam and related tests:** Complete an oral mechanism examination including examination of the lips, tongue, jaw, teeth, and palate (hard and soft). Abnormalities of the oral mechanism common in individuals with CHARGE syndrome include dentition problems (e.g., hypodontia [small teeth], periodontal disease, poor oral hygiene), facial palsy, micrognathia (small jaw), overall weakness of oral musculature, and cleft lip and/or palate.\(^{41,42}\) Additionally, assess oral sensation as sensory deficits are associated with CHARGE syndrome and can affect feeding abilities (e.g., oral hyposensitivity, oral hypersensitivity, tactile defensiveness).\(^{35}\)

**Posture:** Collaborate with PT and/or OT as necessary to assist child in attaining efficient posture for communication. Individuals with CHARGE syndrome often have hypotonia and/or scoliosis, which can affect posture.\(^{1}\)

**Vision:** Vision loss is caused by colobomas of the eye (unilateral or bilateral) that do not cause complete blindness but reduce the visual field, reduce visual acuity, and cause difficulties in certain lighting situations.\(^{1,2}\)

**Speech and language examination**

- **Communication modes:** Determine all communication modes used by observing/interviewing the patient or by parent/teacher report. Communication modes may include speech, physical manipulation, maladaptive behaviors, touch cues, body language, cued speech, lipreading, fingerspelling, gestures, sign language (e.g., American Sign Language, Signed Exact English), tactile sign language (the hand of the listener is placed over the communicator’s hand while signing), tangible symbols or picture symbols, communication boards, tactile calendars, and Tadoma (the hand of the listener is placed over the face and neck of the speaker to monitor actions of the face associated with speech).\(^{1,14}\)

- **Communicative function:** Assessment should include a description of the communicative functions (purpose for communicative) that the patient uses. Examples can include initiations for communication, responses, comments, requests for attention, and protests.\(^{1}\) Checklists can be used to document communicative function

- **Speech**
  - Speech analysis/segment level: If the child uses speech for communication, use a standardized test or an informal (nonstandardized) analysis to determine errors that have a high frequency of occurrence, are early developing, and are functionally important for the child and the child’s family. For specific sounds, the number of omissions, substitutions, distortions, and additions can be completed.

- **Language:** Some standardized tests of expressive and receptive language may be adapted for children with deaf-blindness (e.g., sign language, use of an interpreter), but normative data must not be formally used. Standardized tests are generally not appropriate for children who are deaf-blind and have intellectual disability.\(^{45}\) Systematic observation of play can yield information about receptive and expressive language.\(^{45}\)

- **Voice:** Assess vocal quality and other appropriate voice parameters (such as intensity, pitch, range); neurological impairment can affect vocal quality. Refer to otolaryngologist for complete workup to rule out laryngeal pathologies prior to completing a full evaluation.
For detailed information on voice disorders, see the series of Clinical Reviews on this topic.

- **Fluency:** Rule out or identify atypical speech disfluencies (stuttering)
- For detailed information on disorders of fluency, see the series of Clinical Reviews on this topic

- **Swallow examination:** For detailed information on assessment and treatment of swallowing disorders in patients with CHARGE syndrome, see Clinical Review...Feeding Disorders: CHARGE Syndrome, referenced above
- **Tracheostomy examination:** If present, assess tracheostomy tube and document date of placement, current respiratory status, and use of speaking valve. For detailed information on assessment of a tracheostomy tube and use of a speaking valve, see Clinical Review...Passy-Muir Tracheostomy & Ventilator Swallowing and Speaking Valve; CINAHL Topic ID Number: T708919

- **Special tests specific to diagnosis:** The communication abilities of children with CHARGE syndrome are almost always delayed and are affected by the combination of hearing loss, vision loss, cognitive delay, and the presence of facial nerve palsy due to dysfunction of the facial nerve.\(^1\) Individuals with facial nerve palsy have restricted nonverbal communication because they are unable to demonstrate facial expression (e.g., smiling).\(^1\) The SLP must adapt the evaluation protocol to accommodate the child’s hearing and vision abilities, as well as the child’s level of cognition and attention span. Other issues which can affect communication include cleft lip and palate, vocal fold paralysis, and the presence of a tracheostomy tube.

  - **Gesture/nonverbal interactions:** If the patient is using pre-symbolic communication, a description of gesture and nonverbal interactions can be compiled through observation or videotaped analysis (e.g., classroom, clinic, home) and parent/caregiver or teacher report. Examples of gestures used by children who are deaf-blind may include pushing an object, pushing a person’s hand, turning away, tapping an object, pointing with contact, pointing with distal approximation, “more” gestures, shaking or nodding head, and reaching.\(^16\)

  - **Tests and questionnaires to assess communication:** The following assessments are designed to use with individuals with early developing levels of communication
  - Communication Matrix: This assessment tool was designed to document the communication skills of children who have severe or multiple disabilities and is also applicable for adults who are functioning at early stages of communication.\(^17\)
    It can be used to determine how the individual is communicating and to outline a framework for communication goals.
  - Communication and Symbolic Behavior Scales Developmental Profile: First Normed Edition: This assessment was developed to evaluate communication and symbolic abilities of children with functional communication between the ages of 6 to 24 months of age. It consists of an infant-toddler checklist, a caregiver questionnaire, and a 30-minute behavior sample of the child.\(^18\)

- **Tests and questionnaires developed specifically for children who are deaf-blind**
  - HomeTalk: This assessment tool was developed for parents and care providers of children who are deaf-blind.
    HomeTalk provides a description of the child’s skills (e.g., vision, hearing, problem solving, communication), interests, and personality.\(^19\)

### Assessment/Plan of Care

- **Contraindications/precautions**
  - Only those contraindications/precautions applicable to this diagnosis are mentioned below, including with regard to modalities. Rehabilitation professionals should always use their professional judgment.
  - It is important that SLPs do not underestimate the communication abilities of patients with CHARGE syndrome. Even small gains in communication can significantly improve the patient’s quality of life.\(^1\)
  - Children with CHARGE syndrome have been found to achieve developmental milestones later in life, years after development was thought to have stopped.\(^1\) Treatment considerations should include the possibility for later improvement in communication skills.
  - It is crucial for the SLP to work with other professionals and the family to provide a system of communication for the child as early as possible to minimize isolation of the child, allow the child to communicate needs effectively, and prevent maladaptive behaviors associated with CHARGE syndrome.\(^1\)

- In a 5-case study, researchers in Sweden explored the “health care consumption” of babies with CHARGE syndrome in the first year of life.\(^32\) During the first year of life, the 5 subjects were hospitalized 3-9 times. The total time in the first year spent in inpatient care ranged from 26 days to 230 days. The children saw 8-11 specialists in their first year of life.
The researchers concluded that it is imperative for multiple healthcare providers to be included in the plan of care for a child with CHARGE syndrome and that care should be coordinated and individually adapted to meet each patient’s needs. 

- When working with the individual with CHARGE, caregivers, educators, and SLPs need to be aware of the individual’s “communication bubble,” the area in which the person can hear or see optimally. The size of the communication bubble will vary with respect to the child’s hearing and vision skills as well as environmental factors such as lighting and noise.

**Diagnosis/need for treatment:** Almost all children with CHARGE syndrome have significant speech and language delay and will require early intervention from infancy.

**Rule out:** Other syndromes or disorders should be ruled out by the physician. Features of CHARGE syndrome overlap with other conditions, including DiGeorge sequence, velo-cardio-facial syndrome, cat eye syndrome, and retinoic acid embryopathy.

**Prognosis:** Prognosis for individuals with CHARGE is variable and depends on the severity of sensory loss as well as physical and cognitive skills. In addition, the provision of early intervention services (before the age of 3 years) has been associated with the development of symbolic communication. The presence of a tracheotomy tube can affect speech development.

**Referral to other disciplines:** A team approach for a child with multiple medical and sensory issues is important. Orientation and mobility (O&M) training should be integrated with communication as soon as possible because of the strong association between communication and exploration. The SLP should collaborate with other members of the communication assessment team, including the parents and the deaf-blind specialists, to determine the best mode of communication for the child.

**Other considerations:** When possible, careful selection of equipment and therapy techniques may assist the child in working on speech/language goals and feeding goals simultaneously. Because of the low incidence of CHARGE syndrome, presented material is mainly based on expert opinion or on case studies of children with other causes of deaf-blindness.

**Treatment summary:** The SLP may be involved in direct therapy, parent training, and/or educational support.

Communication modes and methods for the individual with CHARGE syndrome might include spoken or gesture/signed language and/or augmentative and alternative communication (AAC). The communication mode must be consistent among all caregivers and educational specialists who work with the child. The following are types of treatments that have been used with individuals with CHARGE with early and developing communication skills:

- **Adapted prelinguistic milieu teaching (A-PMT):** The purpose of prelinguistic milieu teaching is to teach children to communicate using vocalizations and natural gestures. The approach incorporates the strategies of delayed prompting, modeling, and environmental arrangement in highly motivating teaching contexts. The adapted version can be modified for children with significant hearing and vision loss. Adaptations include minimizing activities that require vision and hearing, increasing vestibular and tactile activities, and use of physical prompts rather than verbal prompts (e.g., hand-under-hand and tactile prompts). Body orientation and searching behaviors can be interpreted as eye gaze and as indicative of attention shift. The effects of A-PMT were examined in a descriptive pilot study with 9 children with hearing and vision loss. Sessions were conducted at the participant’s school or at home. The intervention goal for each child was one initiated communication act per minute over three consecutive probe sessions. Intervention was terminated for each child when the goal was met, and participation in intervention ranged from 2.5 to 8 months. All 9 children demonstrated an increase in communication initiations throughout the study. The author of a case study reported on the use of A-PMT with a male patient with CHARGE syndrome who began treatment at the age of 5 and became increasingly communicative over the course of therapy. At the age of 13, the patient was demonstrating functional, self-initiated communication using manual signs and gestures, visual symbols, and a speech-generating device at home, school, and in the community.

- **Educator-oriented intervention:** Educators are trained to interpret and respond more appropriately to the child’s subtle interactive behaviors. In a multiple baseline study of 4 deaf-blind children (aged 6-9 years) and their 14 educators, researchers found 3 of the 4 children increased their appropriate interactive child behaviors while decreasing their inappropriate behaviors after educator training. Educators in this study were taught to react adequately to the child’s interactive behaviors (e.g., immediately repeating the child’s behavior or utterance) and adapt the interactional context to facilitate interactive behaviors (e.g., provide communicative aids, offer choices, remove distracting stimuli). In a second study examining the long-term effects of this intervention, researchers found that for 2 of the 4 child subjects, the positive effects of the intervention on both educator behaviors and target child behaviors were retained on follow-up (10 weeks post intervention).
• **Oral sensory motor therapy:** A high prevalence of oral sensory issues has been documented in individuals with CHARGE.\(^{(25)}\) There are no research studies on the effects of oral sensory motor therapy on speech and language abilities with children with CHARGE.

• **Positioning/postural adaptations:** Many children with CHARGE have abnormalities in posture, tone, and movement. In these cases, the SLP may collaborate with a PT and/or OT in providing a seating position that is most beneficial for communication and school.

• **Tangible symbols:** Tangible Symbols Systems is a method of communication that uses concrete, rather than abstract, symbols and was designed for people who are unable to understand abstract symbols.\(^{(26)}\) The symbols can include objects or pictures that stand for something (e.g., whole objects, parts of objects, associated objects, textures or shapes, line drawings, photographs). Information on constructing and using tangible symbols is available from Oregon Institute on Disability and Development.\(^{(26)}\)

• **AAC:** Patients with CHARGE syndrome are likely to benefit from AAC to increase functional communication. AAC systems for children with CHARGE syndrome should be individualized for the patient according to communication abilities, visual and hearing impairment, cognitive abilities, and the patient’s preferred method of communication.\(^{(27)}\) For detailed information about AAC, please see the series of Clinical Reviews on this topic.

<table>
<thead>
<tr>
<th>Problem</th>
<th>Goal</th>
<th>Intervention</th>
<th>Expected Progression</th>
<th>Home Program</th>
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</thead>
<tbody>
<tr>
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<tr>
<td>Patient has limited communicative interactions</td>
<td>Increase communicative interactions with the patient</td>
<td><strong>Methods to establish and increase interaction</strong></td>
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<tr>
<td>Some children with CHARGE have limited interactions with people in their environment. Some general principles to enhance interactions include:</td>
<td></td>
<td>Once established, progress from simple to complex communicative interactions as appropriate given the child’s abilities</td>
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<tr>
<td>• Let the child know what will happen before the interaction occurs (e.g., use touch or tangible symbols)</td>
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<tr>
<td>• Use touch and object cues for communication</td>
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<td>• Stay within the “communication bubble” (e.g., within the child’s visual field)</td>
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<td>• Use coactive and interactive signing (e.g., sign with the child)</td>
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<tr>
<td>• Use simple and direct messages</td>
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<tr>
<td>• Allow the child sufficient time to respond</td>
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<tr>
<td>• Acknowledge any attempt of the child to communicate</td>
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</tbody>
</table>

Methods to establish and increase the patient’s interactions should be used in all communicative contexts
| Patient does not have symbolic language | Increase the patient’s ability to communicate using symbols | **Tangible symbols**

Tangible Symbols Systems is a method of communication that uses concrete, rather than abstract, symbols and was designed for people who are unable to understand abstract symbols.\(^{26}\) The symbols can include objects or pictures that stand for something (e.g., whole objects, parts of objects, associated objects, textures or shapes, line drawings, photographs). Symbols are first used to request something that is desired (e.g., food, activity). Once the individual has learned to request, other communicative functions can be learned using the symbols (e.g., labeling, commenting). The SLP should begin with symbols that are at a level that the patient can understand and use them in a highly motivating context.

<p>| The individual will learn to respond by using the symbol (e.g., touching the symbol, handing the symbol to the teacher) | Once the patient learns to use specific symbols in a controlled context, they should be used in school and home |</p>
<table>
<thead>
<tr>
<th>Once the patient learns to use specific symbols in a controlled context, they should be used in school and home</th>
<th>The child will increase communicative acts</th>
<th>Adapted prelinguistic milieu teaching (A-PMT)</th>
<th>Increase vocalizations and natural gestures with delayed prompting, following the child's lead, modeling, social routines, and environmental arrangements</th>
</tr>
</thead>
<tbody>
<tr>
<td>Adapted prelinguistic milieu teaching (A-PMT)</td>
<td>The purpose of prelinguistic milieu teaching is to teach children to communicate using vocalizations and natural gestures. The approach incorporates the strategies of delayed prompting, following the child's lead, modeling, social routines (e.g., peekaboo, bathing, feeding) and environmental arrangement in highly motivating teaching contexts. (21,22) The adapted version can be modified for children with significant hearing and vision loss. Adaptations include minimizing activities that require vision and hearing, increasing vestibular and tactile activities, and use of physical prompts rather than verbal prompts (e.g., hand-under-hand and tactile prompts). Body orientation and searching behaviors can be interpreted as eye gaze and as indicative of attention shift.</td>
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</tr>
<tr>
<td>Use of inappropriate interactive behaviors</td>
<td>Reduce inappropriate interactive behaviors and increase appropriate behaviors</td>
<td>Educator-oriented intervention</td>
<td>Once identified, systematically respond to and either positively or negatively reinforce both appropriate and inappropriate behaviors</td>
</tr>
<tr>
<td>Educator-oriented intervention</td>
<td>Educators are trained to interpret and respond more appropriately to the child's subtle interactive behaviors (23,24)</td>
<td>No home-based program has been trialed but parents/caregivers should be taught these strategies to use at home</td>
<td>No home-based program has been trialed but parents/caregivers can also be taught these strategies to use at home</td>
</tr>
<tr>
<td>Lack of joint attention</td>
<td>Establish and increase joint attention between the child and communication partner</td>
<td><strong>Using preferred objects</strong></td>
<td>Once established, progress from simple to complex joint attention interactions as appropriate given the child’s abilities</td>
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<tr>
<td></td>
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<td>Joint attention can be first established on a variety of preferred objects. To scaffold a child’s exploration of objects, adults can demonstrate how to experience the object through multiple senses. Using hand under hand exploration, adults can indicate the visual, auditory, olfactory and tactile features of the object depending on the child’s abilities. It is important for the child to be free to leave and return to the adult’s hands for exploration so that the child can moderate the level of stimulation(^{27})</td>
<td></td>
</tr>
<tr>
<td>Lack of communicative initiations</td>
<td>Increase child’s communicative initiations using gesture and body language</td>
<td>Recognize and reinforce initiations</td>
<td>Systematic reinforcement of gestures and communicative initiations should be provided</td>
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<tr>
<td>Parents and teachers can be taught to recognize a child’s early initiations. Examples of such initiations may include reaching to locate an object or leaning toward others to indicate a desire to interact. Adults can reinforce such initiations by responding to their interpreted intent (e.g., giving the child an object). Gestures that spontaneously emerge in motivating situations are more likely to be recalled and reproduced. If a child uses “home signs” or idiosyncratic gestures, it is important that the adult first respond to the invented signs before shaping them into the conventional form.</td>
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</tbody>
</table>

(14, 27)
<table>
<thead>
<tr>
<th>Lack of vocalizations</th>
<th>Increase the child’s vocalizations</th>
<th><strong>Vocal training</strong></th>
<th>Systematic reinforcement of vocalizations should be provided</th>
<th>The child’s parent/caregiver can be trained to reinforce vocalizations</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td><strong>Reinforcement of vocalizations</strong></td>
<td>After maximizing the child’s access to auditory input through appropriate augmentation, adults can create opportunities for the child to vocalize by reinforcing babbling. Determine the functional intent of the vocalization and respond regularly. For example, if the caregiver determines the sound “ah” is to gain attention, then the caregiver should respond in a systematic way (e.g., touching the child’s hand) after hearing the vocalization</td>
<td></td>
</tr>
</tbody>
</table>

27
<table>
<thead>
<tr>
<th>Lack of variety in pragmatic function</th>
<th>Increase function of language</th>
<th><strong>Provide opportunities for the child to use a variety of communicative intents</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Lack of communication and engagement in the classroom</td>
<td>Increase opportunities for communication and engagement by restructuring the environment</td>
<td>Provide opportunities for communication in increasingly complex situations and with different communication partners</td>
</tr>
</tbody>
</table>

The following intents occur early in development but are often absent in the communication of a child with deaf-blindness: protesting, requesting objects, information and actions, calling for attention, greeting, showing objects, giving objects, answering, labeling, and commenting on objects and actions. Adults in the child's environment have to create opportunities for different functions (e.g., a situation that may elicit a protest, providing opportunities for choice-making at mealtimes)\(^{(14)}\)

<table>
<thead>
<tr>
<th>Lack of communication and engagement in the classroom</th>
<th>Increase opportunities for communication and engagement by restructuring the environment</th>
<th><strong>Environmental modifications</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Classrooms can be structured to increase opportunities for communication by 1) placing students so they face each other (U-shaped desks or round tables), 2) using structured routines, and 3) using calendars or communication shelves. Communication shelves are arranged in order of a routine and contain objects that signify a specific activity(^{(28)})</td>
<td>N/A</td>
<td>No home-based program has been trialed, but environmental modifications can also be applied in the home</td>
</tr>
</tbody>
</table>
Desired Outcomes/Outcome Measures

› Increased level of communicative interaction
  • Communication Matrix
  • Communication and Symbolic Behavior Scales Developmental Profile: First Normed Edition
› Increased variety of communicative functions/acts
  • Communication Matrix
  • Communication and Symbolic Behavior Scales Developmental Profile: First Normed Edition
› Increased ability to use a particular mode of communication (e.g., speech, sign language, gesture)
  • Communication Matrix
  • Communication and Symbolic Behavior Scales Developmental Profile: First Normed Edition
› Increased comprehension ability
  • Communication Matrix
  • Communication and Symbolic Behavior Scales Developmental Profile: First Normed Edition

Outcome statements outlined in the IFSP or IEP should indicate the context or environment in which the outcomes should occur
• For preschoolers, these outcomes might occur within the context of school in addition to the home setting

Maintenance or Prevention

› It is important to monitor any progressive hearing loss or vision loss. Refer to an ophthalmologist or audiologist if additional loss is suspected

Patient Education

› See Patient Information Series from the CHARGE Syndrome Foundation at http://www.chargesyndrome.org/
› See information on deaf-blindness from the National Center on Deaf-Blindness at https://nationaldb.org/
› CHARGE Family Support Group of the United Kingdom at http://www.chargesyndrome.org.uk/

Note

› Recent review of the literature has found no updated research evidence on this topic since previous publication on December 18, 2015

Coding Matrix

References are rated using the following codes, listed in order of strength:

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>M</td>
<td>Published meta-analysis</td>
</tr>
<tr>
<td>SR</td>
<td>Published systematic or integrative literature review</td>
</tr>
<tr>
<td>RCT</td>
<td>Published research (randomized controlled trial)</td>
</tr>
<tr>
<td>R</td>
<td>Published research (not randomized controlled trial)</td>
</tr>
<tr>
<td>C</td>
<td>Case histories, case studies</td>
</tr>
<tr>
<td>G</td>
<td>Published guidelines</td>
</tr>
<tr>
<td>RV</td>
<td>Published review of the literature</td>
</tr>
<tr>
<td>RU</td>
<td>Published research utilization report</td>
</tr>
<tr>
<td>QI</td>
<td>Published quality improvement report</td>
</tr>
<tr>
<td>L</td>
<td>Legislation</td>
</tr>
<tr>
<td>PGR</td>
<td>Published government report</td>
</tr>
<tr>
<td>PFR</td>
<td>Published funded report</td>
</tr>
<tr>
<td>X</td>
<td>Practice exemplars, stories, opinions</td>
</tr>
<tr>
<td>GI</td>
<td>General or background information/texts/reports</td>
</tr>
<tr>
<td>U</td>
<td>Unpublished research, reviews, poster presentations or other such materials</td>
</tr>
<tr>
<td>CP</td>
<td>Conference proceedings, abstracts, presentation</td>
</tr>
</tbody>
</table>

References

2. Blake KD, Prasad C. CHARGE syndrome. Orphanet J Rare Dis. 2006;1:34. (RV)
4. Thelin JW, Swanson LA. CHARGE syndrome: multiple congenital anomalies including disorders of all senses and speech, language, feeding, swallowing and behavior. ASHA Lead. 2006;11(14):6-7. (GI)
6. Holte L, Van Dyke DC, Lubrica P. Issues in the evaluation of infants and young children who are suspected of or who are deaf-blind. Infants Young Child. 2006;19(3):213-227. (GI)


28. Engleman MD, Griffin HC, Griffin LW, Maddox JI. A teacher's guide to communicating with students with deaf-blindness. Teach Except Child. 1999;31(5):64-70. (GI)


35. Rosenfeld-Johnson S. CHARGE syndrome: feeding and swallowing. ASHA Lead. 2006;11(14):40. (GI)


