



## Factsheet 15

# Early intervention for children with CHARGE syndrome

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Early intervention for children with CHARGE syndrome presents some highly specialised challenges to all of those involved. Early intervention is often delayed due to pressing medical concerns, with medical issues and medically-based therapy taking precedence over educational and less medically based therapy. It is important that intervention begins as early as possible.

### Parents

The most effective interventions are thought to be a coordinated service from a multidisciplinary or a transdisciplinary team, who value family-centred practice (Dunst, 2002) and the Strength-Based approach – which emphasises people’s ability to be their own agents of change by creating conditions that enable them to control and direct the process of change (McCashen, 2005).

It is considered that when parents are involved in their children’s early intervention better outcomes are realised.

### Some of the questions, which may arise for families

- What is early intervention?
- When does this start?
- Who will coordinate these services?
- Who are the service providers in early intervention?
- What do they do and how can they help?

The professionals involved in early intervention will need to address these questions with families.

They may include:

- Educators
- Speech and Language Therapists
- Physiotherapists
- Occupational Therapists



- Educational Psychologists
- Social Workers
- Dieticians
- Sign Language Tutors

### The roles of professionals likely to be involved in early intervention

#### **SPECIALIST TEACHERS IN EARLY CHILDHOOD DEVELOPMENT**

- Teacher of children who are deaf or hearing impaired
- Teacher of children who are visually impaired or blind
- Teacher of children who are Multi-Sensory Impaired (MSI)/Deafblind
- Portage/Early Years Teachers

Children with CHARGE have characteristic outer ear, middle ear and inner ear conditions, which include ossicular malformations, mondini defect of the cochlear and absent or malformed semi circular canals, seen in



80–90% of affected individuals (Hartshorne *et al.* 2011), resulting in a sensorineural and conductive hearing loss.

In assigning a **teacher of children who are deaf or hearing impaired** to a family who has a child with CHARGE syndrome, consideration should be given to someone who:

- is fluent in sign language, knowledgeable on augmentative communication systems and the development of speech and communication
- has a thorough knowledge of the technology available, including hearing aids, cochlear implants, bone anchored hearing aids (BAHA) and FM systems and the agencies and companies who provide these.

Knowledge of the organisations available to people who are deaf and deafblind/MSI is also helpful and the resource of a **Sign Language Tutor** may be sought.

It is important for these teachers to be aware of the ways in which children with CHARGE syndrome develop a meaningful communication system. This is often not typical of other children who are deaf. For example, some children with CHARGE acquire a visual language initially, which may be sign language, picture symbols, object symbols and then at a later stage develop speech.

The critical issue is that a communication system needs to be explored, developed and provided as early as possible. The child should be immersed in these systems, always taking into consideration their hearing, vision, medical circumstances and the complex issues surrounding each individual.

The **teacher of children who are visually impaired or blind** needs to:

- be familiar with the assessment and development of vision
- provide the appropriate play environment and educational materials for a child with vision impairment
- have knowledge of the impact of coloboma on the use of vision, which is present in 80–90% of affected individuals with CHARGE (Hartshorne *et al.* 2011).

Coloboma is an eye defect where parts of the eye do not grow fully when developing in the womb. If a part of the eye does not grow then this is known as a coloboma. Coloboma means ‘curtailed’ or ‘unfinished’. The parts that sometimes do not finish growing fully include the iris, lens, retina or optic nerve.

The effect is that the ‘unfinished section’ allows too much light to enter the eye so that glare and bright light are an issue. Creating a suitable visual environment for the child, where light is controlled and glare reduced, is essential – with sunglasses and hats being worn outside.

A **teacher of children who are MSI/deafblind** is skilled in supporting the **overall** development of these children. This will include all the areas already mentioned and particular skills in the development of an individualised communication programme. They recognise and respond to the child’s different sensory impairments and provide programmes accordingly.

Other specialists will be **audiologists** for hearing and **orthoptists** for vision.

Due to the ongoing assessment of the child’s hearing and vision, it’s useful to seek permission from the family to accompany them to these assessments. It provides an opportunity to share knowledge and build useful professional relationships.

## SPEECH AND LANGUAGE THERAPISTS

In children with CHARGE, we see characteristic ear abnormalities involving the external ear, middle ear malformations, mixed deafness and cochlear defects. Also cranial nerve dysfunction involving anosmia, facial palsy and vestibular (balance) problems and/or eating and drinking difficulties.

A speech and language therapist is an essential member of the team involved with the child and the family. The aim is to develop customised communication systems, which may include speech, sign language, touch cues, object symbols and other augmentative strategies – such as pictographs and other technology applications currently available. Children with CHARGE may develop a visual language first, acquiring speech later.

It is also beneficial to have the involvement of a speech and language therapist who has a special interest in dysphagia (eating and drinking difficulties). Many children with CHARGE syndrome receive their sustenance from tube feeding, initially through naso-gastric feeding and perhaps progressing to gastrostomy feeding. Swallowing problems with aspiration occurs in 70–90% of children and reflux in 30–40% (Hartshorne *et al.* 2011).

A thoroughly planned programme needs to be in place to support the child and the family in transitioning from tube feeding to eating orally safely. The speech and language therapist and medical team will collaborate with the parents in the steps required to achieve this. This may also involve a dietician.

The achievement of transitioning from tube feeding to eating orally varies greatly from child to child – with some never managing to eat orally.



## PHYSIOTHERAPISTS

Physiotherapists are essential to assist in the development of movement in children with CHARGE syndrome.

Due to the absent or hypoplastic (underdeveloped) semicircular canals they do not develop movement in a typical fashion. This is seen in 80–90% of children with CHARGE (Hartshorne *et al.* 2011).

Other contributing factors are reduced vision and poor proprioception. Children with CHARGE are delayed in walking and tackle the process of learning to walk differently. Initially, they may lie on their back requiring the full support of the floor. Rolling and movement are delayed, with a back shuffle often used to start movement around the environment. They may attempt a five-point crawl using their head as the fifth point of contact to the floor. Eventually cruising around furniture commences, which may go on for a long time, always with poor balance and a wide stance.

Once independent walking begins, which may be from about three years on or later, poor balance and a wide stance continues. Being thrown off balance happens easily and vigilance is required to ensure safety. The miracle is that these children achieve walking at all.

## OCCUPATIONAL THERAPISTS

An occupational therapist's role is to help the child and the family in the development of play, fine motor skills, self-help skills, in identifying equipment needs and the way in which the child interacts with objects and people. The aim is to ensure the child is well supported in seating – be it a high chair, stroller or chair – and to adapt equipment to suit the child.

The occupational therapist also contributes to the child's physical development and balance issues.

It is also helpful for the occupational therapist to have knowledge of the importance of **Sensory Integration** and preferably possessing additional training in this area.

## EDUCATIONAL PSYCHOLOGISTS

Educational psychologists can provide assessments and information about the child's development and behaviour. They may also work with the family to help them to understand the implications of having a child who has CHARGE syndrome.

In addition, the services of **social workers** and **counsellors** may be sought to provide emotional and practical support. They may also refer families to community resources, which may best suit their family needs.

## Working in partnership with parents

It is widely acknowledged that parents are the most knowledgeable about their child and the practices that are most helpful to them. Discussions with parents reveal that they themselves become the coordinators of their child's services.

Parents require professionals to help them to gather and interpret information, answer questions, engage in discussions, provide support and help in decision-making and to share their professional expertise with the family.

They may benefit from a practitioner who can undertake a coordinating role and is readily available, reliable and knowledgeable so that appropriate support is provided through the complex, often extremely difficult, early years.

It is essential that parents are involved in all discussions and meetings surrounding their child and that they are given all information to enable them to make informed decisions.

Establishing educational and therapy plans from as early as possible, based on the family's aspirations and desires, should form the basis of the child's programme. Parents report that early predictions for their child's potential were often pessimistic and that their child's potential was in fact unclear in the early years. Many of these children are now teenagers and adults displaying a wide range of abilities and varied outcomes.

The importance of establishing a communication system/systems as early as possible cannot be stressed enough. It is seen as best practice for families, educators and therapists to work together to achieve this.

Regardless of the amount of help provided to parents by professionals, the ultimate goal is clear. Professionals must aim to transfer skills to parents, thereby ensuring that therapy and education is integrated naturally into all aspects of family life, resulting in the enhancement of parenting skills.

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## GLOSSARY

**Anosmia:** lack of the sense of smell

**Cranial nerve dysfunction:** an impairment of any or all of the cranial nerves – these being the nerves that emerge directly from the brain rather than the spinal cord.

**Proprioception:** the unconscious perception of movement and spatial orientation arising from the information received from the tendons, joints and ligaments.

**Strength-Based approach:** an approach that is child-led and focusses on the strengths of the child and the family.

**Transdisciplinary:** people from different professions working together to ensure a holistic approach.

## REFERENCES

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