

CHARGE Syndrome Canada is a National organization which promotes the awareness and recognition of persons of CHARGE syndrome in Canada, and provides information to these individuals, their families, professionals and the Canadian public. We hope to:

- Increase public awareness of CHARGE syndrome
- Act as a resource for individuals with CHARGE syndrome, their families and professionals
- Support the philosophy of using individualized approaches and techniques for persons with CHARGE syndrome
- Work with both government and non-government agencies and organizations to foster networking and information exchange
- Support related research

CHARGE Syndrome Canada helps to network professionals, individuals with CHARGE syndrome, families, and those with an interest in CHARGE syndrome.

Contact us for information. Be added to the mailing list and become a member of the organization. Make a donation, volunteer, or share a story.

Registered charity #888-365-707-RR0001

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CHARGE SYNDROME CANADA



WHAT IS CHARGE SYNDROME?

CHARGE syndrome is a multi-clefting, missing or undeveloped tissue disorder which is thought to occur around the third week of fetal development. Infants experience many early critical medical procedures and long hospital stays, which could cause a delay in development.

Sensory loss also affects infant development. Vision and/or hearing may be affected, as well as the vestibular sense and the sense of smell. Unique sensory processing is common, and sensory integration therapy is often useful.

Many children can be smaller than average in their growth. This could also be due to hormone deficiency and/or feeding and nutrition problems.

Not all individuals with CHARGE will have the same clinical features. All features can vary in their degree of severity.



REVISED CRITERIA 2003

Definite CHARGE: 3 Major characteristics or 2 Major and 3 Minor

Probable/Possible CHARGE:

1 Major characteristic and several minor or other characteristics

USUAL MAJOR FEATURES:

Coloboma An incomplete congenital development of various structures of the eye(s) or visual system resulting in varying degrees of vision loss and/or interference.

Choanal atresia or stenosis passages at the back of the nose to the throat which allow for breathing are blocked (atresia) or narrow (stenosis) often requiring surgery.

cranial nerve dysfunction or anomaly

Common problems can include facial palsy (one or both sides), problems with swallowing, feeding, and movement of food down the gastrointestinal tract.

Characteristic CHARGE ear and Absent, missing or malformed **semi-circular canals**



MINOR FEATURES

Characteristic CHARGE face
Characteristic CHARGE hand
Urinary tract and/or renal anomalies
Orofacial
Congenital heart defects
Genital (hypogonadotropic hypogonadism)
Postnatal growth deficiency
Hypotonia (weakness)
Anomalies of trachea and/or esophagus
Chronic otitis and sinusitis
Sloping shoulders
Limb/skeletal
CNS anomalies
Thymic or parathyroid hypoplasia
Nipple anomalies
Omphalocele
Developmental delay

**Evaluation for a diagnosis should be made by a geneticist who is familiar with CHARGE syndrome.*

OUTCOMES

Early diagnosis of all symptoms is extremely important for an individual with CHARGE syndrome. Cardiac, renal, Ophthalmological, audiological, and gastrointestinal evaluations should be done on any individual suspected of having CHARGE.

Medical and surgical interventions will usually take place in the first couple of years, when the focus will shift more towards development and educational needs.

A multi-disciplinary team, managed by a pediatrician or geneticist is often helpful and necessary for a child with CHARGE.

With early diagnosis, treatments and follow up, individuals with CHARGE can achieve their best potential. These children have unique learning challenges but often catch up on developmental milestones with therapy, assistive devices, technology, and specific approaches.

CAUSE of CHARGE

August, 2004, the first major CHARGE gene was reported. CHD7 is a gene on chromosome 8q12. Mutations in this gene were identified in about 65% of children tested. This confirms CHARGE as an autosomal dominant condition, with most cases representing new mutations. The cause of this gene change in the fetus is unknown. Prevalence is under reported but thought to be at least 1:12 000 births.

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