

CHARGE ASSOCIATION/SYNDROME (CAS)

CANADIAN PAEDIATRIC SURVEILLANCE PROGRAM

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REPORTING INFORMATION

(To be completed by the CPSP Senior Coordinator)

Report number:

Month of reporting:

Province:

Today's date:

**Please complete the following sections for the case identified above.
Confidentiality of information will be assured.**

CASE DEFINITION FOR CHARGE ASSOCIATION/SYNDROME

- Infant/child/adult with **all four major criteria**.
- Infant/child/adult with **three major and three minor criteria**.
- **Previously diagnosed** child with CAS that does not fit major or minor criteria, but has a combination of the above plus some occasional findings; renal, hand, spine/limb, abdominal (hernia) anomalies.

Major inclusion criteria

1. Coloboma – of iris, retina, choroid, disc; microphthalmia
2. Choanal atresia – unilateral/bilateral, membranous/bony, stenosis/atresia
3. Characteristic ear abnormalities – external ear (lop or cup-shaped), middle ear (ossicular malformations, chronic serous otitis), mixed deafness, cochlear defects
4. Cranial nerve dysfunction – facial palsy (unilateral or bilateral), sensorineural deafness and/or swallowing problems

Minor inclusion criteria

1. Genital hypoplasia – males: micropenis, cryptorchidism; females: hypoplastic labia; both males and females: delayed, incomplete pubertal development
2. Developmental delay – delayed motor milestones, language delay, mental retardation
3. Cardiovascular malformations – all types, especially conotruncal defects (e.g., tetralogy of Fallot), AV canal defects, and aortic arch anomalies
4. Growth deficiencies – short stature, growth hormone deficiency
5. Orofacial cleft – cleft lip and/or palate
6. Tracheoesophageal-fistula – tracheoesophageal defects of all types
7. Characteristic face – sloping forehead, flattened tip of nose

Exclusion criteria

Exclude other conditions such as velocardiofacial syndrome (VCS) and DiGeorge Sequence (DGS) using FISH test (Fluorescent In Situ Hybridisation) to exclude 22q 11 deletion.

SECTION 1 – DEMOGRAPHIC INFORMATION

1.1 Date of birth: / /
 DD MM YYYY

1.2 Sex: Male Female

SECTION 2 – DIAGNOSTIC INVESTIGATION RESULTS

2.1 Karyotype: Normal Abnormal Unknown

2.2 Fluorescent in situ hybridisation (FISH): Normal (neg) Abnormal (22q 11 deletion) Unknown

2.3 Maternal date of birth: / /
 DD MM YYYY

2.4 Paternal date of birth: / /
 DD MM YYYY

SECTION 3 – BIRTH PARAMETERS

- 3.1 Gestational age: ____ weeks
- 3.2 Birth weight: ____ kg NA ____
- 3.3 Birth length: ____ cm NA ____
- 3.4 Head circumference: ____ cm NA ____
- 3.5 Asphyxia at birth: Yes ____ No ____ NA ____
If yes, describe: _____
- 3.6 Apgar (5 min.): _____ NA ____
- 3.7 Date of diagnosis of CHARGE child: ____ / ____ / ____
DD MM YYYY
- 3.8 Date of death of CHARGE child: ____ / ____ / ____ NA ____ (please provide autopsy report if available)
DD MM YYYY

SECTION 4 – PHYSICAL FINDINGS (MAJOR CRITERIA)

- 4.1 **Coloboma (or microphthalmos):** Yes ____ No ____ NA ____

(if “no” or not available [NA], proceed to question 4.2)

- 4.1.1 Iris: Right eye: Yes ____ No ____ NA ____ Left eye: Yes ____ No ____ NA ____
- 4.1.2 Retina/choroid/disc: Right eye: Yes ____ No ____ NA ____ Left eye: Yes ____ No ____ NA ____
- 4.1.3 Microphthalmos: Right eye: Yes ____ No ____ NA ____ Left eye: Yes ____ No ____ NA ____
- 4.1.4 Visual impairment (VI): Normal ____ Mild ____ Moderate ____ Severe ____ Profound ____ NA ____

(**Mild** = VA 6/6 – 6/18 corrected in better eye; severe or profound problem with one eye only, other eye normal. **Moderate** = VA 6/24 – 6/36 in better eye; able to read print with simple aids and/or education assistance; defect of at least half visual field (hemianopia); VA may be normal. **Severe** = VA 6/60 – 3/60 in better eye; unable to read large print without intensive educational assistance or sophisticated aids; severe visual field defect with impaired visual acuity. **Profound** = VA <3/60, i.e., counting fingers, hand movements, light perception or less; very little useful visions.

Describe acuity: _____

Describe adaptation: _____

- 4.2 **Choanal atresia/stenosis:** Yes ____ No ____ NA ____

(if “no” or not available [NA], proceed to question 4.3)

- 4.2.1 Bilateral choanal atresia: Yes ____ No ____ NA ____
- 4.2.2 Choanal atresia: Right: Yes ____ No ____ NA ____ Left: Yes ____ No ____ NA ____
- 4.2.3 Choanal stenosis: Right: Yes ____ No ____ NA ____ Left: Yes ____ No ____ NA ____

Describe choanal atresia (bony or osseous): _____

- 4.3 **Characteristic ear abnormalities (either/or external/middle/internal):** Yes ____ No ____ NA ____

(if “no” or not available [NA], proceed to question 4.4)

- 4.3.1 External ear (lop or cup shape): Yes ____ No ____ NA ____
- 4.3.2 Middle ear (ossicular malformation): Yes ____ No ____ NA ____
- 4.3.3 Middle ear (chronic serous otitis media, often needing T tubes): Yes ____ No ____ NA ____
- 4.3.4 Inner ear – deafness: Right: Yes ____ No ____ NA ____ Left: Yes ____ No ____ NA ____
- 4.3.5 Temporal bone CT scan: Yes ____ No ____ NA ____
- 4.3.6 Hearing impairment (HI): Normal ____ Mild ____ Moderate ____ Severe ____ Profound ____ NA ____

(**Mild** = hearing loss 20-40 dB; severe or profound loss in one ear only, other ear normal. **Moderate** = hearing loss 41-70 dB. **Severe** = hearing loss 71-95 dB. **Profound** = hearing loss >95 dB)

Describe loss in decibels: _____

Describe adaptation(s), i.e., aids, cochlear implants: _____

- 4.4 **Cranial nerve anomalies:** Yes ___ No ___ NA ___
 (if “no” or not available [NA], proceed to question 4.5)
- 4.4.1 Weak chewing/sucking: Yes ___ No ___ NA ___
- 4.4.2 Facial palsy: Right: Yes ___ No ___ NA ___ Left: Yes ___ No ___ NA ___
- 4.4.3 Sensory neuro deafness: Yes ___ No ___ NA ___
- 4.4.4 Balance/vestibular problems: Yes ___ No ___ NA ___
- 4.4.5 Swallowing problems: Yes ___ No ___ NA ___

SECTION 5 – PHYSICAL FINDINGS (MINOR CRITERIA)

Male

- 5.1 Micro penis: Yes ___ No ___ NA ___ 5.2 Cryptorchidism: Yes ___ No ___ NA ___

Female

- 5.3 Hypoplastic labia: Yes ___ No ___ NA ___

Male/Female

- 5.4 Cardiovascular malformations (minor): Yes ___ No ___ NA ___
 (minor = PDA, small ASD/VSD, no repair)
- 5.5 Cardiovascular malformations (major): Yes ___ No ___ NA ___
 (major = Tetralogy of Fallot AV canal, aortic arch)
- 5.6 Describe heart disease: _____
- 5.7 Cleft lip: Yes ___ No ___ NA ___
- 5.8 Cleft palate: Yes ___ No ___ NA ___
- 5.9 Tracheoesophageal fistula: Yes ___ No ___ NA ___
- 5.10 Distinctive face of CHARGE: Yes ___ No ___ NA ___

SECTION 6 – PHYSICAL FINDINGS (OCCASIONAL)

- 6.1 Renal anomalies: Yes ___ No ___ NA ___
 Describe: _____
- 6.2 Hand anomalies (e.g., polydactyly, thumb hypoplasia): Yes ___ No ___ NA ___
- 6.3 Spine anomalies (e.g., hemivertebrae): Yes ___ No ___ NA ___
- 6.4 Abdominal defects (e.g., hernia): Yes ___ No ___ NA ___
- 6.5 Neck anomalies (sloping shoulders/webbing/short): Yes ___ No ___ NA ___
- 6.6 Teeth anomalies: Yes ___ No ___ NA ___
- 6.7 Immune function anomalies: Yes ___ No ___ NA ___ If yes, specify: _____
- 6.8 Other findings not listed above: _____

SECTION 7 – FAMILY HISTORY

- 7.1 Similarly affected relatives (with any features of CHARGE): Yes ___ No ___ NA ___
 Describe: _____
- 7.2 Other significant family history (hearing impairments, developmental disability or learning):
 Yes ___ No ___ NA ___ Describe: _____
- 7.3 Ethnicity (mother): Caucasian ___ Asian ___ Afro-Canadian ___ South Asian ___ Native Canadian ___
 Other _____
- 7.4 Ethnicity (father): Caucasian ___ Asian ___ Afro-Canadian ___ South Asian ___ Native Canadian ___
 Other _____

SECTION 8 – GASTROINTESTINAL

8.1 Gastroesophageal reflux: Yes ___ No ___ NA ___

8.2 Feeding problems: Yes ___ No ___ NA ___

Describe: _____

8.3 Required a G or J tube for feeding? Yes ___ No ___ NA ___

Describe (how long): _____

Describe current method(s) of feeding: _____

SECTION 9 – BEHAVIOURAL/PSYCHOLOGICAL

9.1 Hyperactivity/inattention: Yes ___ No ___ NA ___

9.2 Major sleep problems: Yes ___ No ___ NA ___

9.3 Repetitive/obsessive/compulsive (talk or movement): Yes ___ No ___ NA ___

9.4 Medications for behaviour: Yes ___ No ___ NA ___

Describe: _____

SECTION 10 – ENDOCRINE10.1 Short stature (< 5th centile): Yes ___ No ___ NA ___

10.2 Growth hormone deficiency: Yes ___ No ___ NA ___

10.3 Delayed puberty: Yes ___ No ___ NA ___

10.4 Medication/HRT for endocrine disorder: Yes ___ No ___ NA ___

Describe: _____

SECTION 11 – NEUROLOGY

11.1 Seizures: Yes ___ No ___ NA ___

11.2 Scoliosis: Yes ___ No ___ NA ___

11.3 Migraine: Yes ___ No ___ NA ___

11.4 CT/MRI scan abnormal: Yes ___ No ___ NA ___

If abnormal, describe: _____

SECTION 12 – SURGERY/ANAESTHESIA

12.1 Tracheostomy: Yes ___ No ___ NA ___

12.2 T tube insertion number: 1-2 ___ 3-4 ___ 5-6 ___ 7+ ___ Zero ___

12.3 Surgical procedures: 1-3 ___ 4-6 ___ 7-10 ___ 11-14 ___ 15+ ___

12.4 Anaesthesia: 1-3 ___ 4-6 ___ 7-10 ___ 11-14 ___ 15+ ___

12.5 Anaesthetic complications: Yes ___ No ___ NA ___

Describe: _____

12.6 Sedation complications: Yes ___ No ___ NA ___

Describe: _____

SECTION 13 – PATIENT INFORMATION

13.1 Father's occupation (before pregnancy): _____

During pregnancy

13.2 Mother's occupation: _____

13.3 Use of alcohol: Yes ___ No ___ NA ___

Describe (when and how much): _____

13.4 Smoking: Yes ___ No ___ NA ___

Describe (when and how much): _____

13.5 Medications used: Yes ___ No ___ NA ___

Describe (types of medications, when, quantity and reason): _____

13.6 Fever/Infection: Yes ___ No ___ NA ___

Describe (when, what, and duration): _____

13.7 Bleeding: Yes ___ No ___ NA ___

Describe (when, how much, duration): _____

13.8 X-rays: Yes ___ No ___ NA ___

Describe (when, type and why): _____

13.9 Use of hair treatments: Yes ___ No ___ NA ___

Describe (when and what type): _____

13.10 Contact with pesticides: Yes ___ No ___ NA ___

Describe (when, duration, type): _____

Describe (when and frequency): _____

13.12 Any other concerns about exposures: Yes ___ No ___ NA ___

Describe (when and what type): _____

SECTION 14 – REPORTING PHYSICIAN

First name _____ Surname _____

Address _____

City _____ Province _____ Postal code _____

Telephone number _____ Fax number _____

E-mail _____ Date completed _____

Thank you for completing this form.