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/ataresia
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: _____/_____/______ 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: _____/_____/______
2.4 Paternal date of birth: _____/_____/______
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 – ENDOCRINE

10.1 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): __________________________________________________

13.11 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.