CHARGE SYNDROME REPORTING FORM

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REPORTING INFORMATION

(To be completed by Dr. Kim Blake)
Unique identifier:
Month of reporting:
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 CHARGE that does not fit major or minor criteria, but has a combination of the above plus some occasional findings; renal, hand, spine/limb, abdominal 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 Unique identifier: ____________________________ 1.2 Date of birth: ____ /_____ /_______
DD MM YYYY

1.3 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 ___

2.2.1 Gene testing for CHARGE (i.e., CHD7) Yes ___ No ___ Describe results______________________________

2.2.2 Subtelomeric FISH studies done: Yes ___ No ___ NA ___ Describe results _________________________

2.3 Maternal date of birth: ____ /_____ /_______ 2.4 Paternal date of birth: ____ / ____ / ________
DD MM YYYY DD MM YYYY

2.5 Mother's place of birth: ____________________ 2.6 Father's place of birth: ______________________

SECTION 3 – BIRTH PARAMETERS (NOTE: NA = NOT AVAILABLE)

3.0 Place of birth (Province, Country): ____

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 CHARGE diagnosis: ____ /_____ /_______
DD MM YYYY

3.8 Date of death of individual with CHARGE: ____ /____ /_____ 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 (R. eye): Normal ___ Mild ___ Moderate ___ Severe ___ Profound ___ NA ___

4.1.5 Visual impairment (L. eye): Normal ___ Mild ___ Moderate ___ Severe ___ Profound ___ NA ___

(**Mild** = visual acuity 6/6 (20/20) – 6/18 (20/60); **Moderate** = visual acuity 6/24 (20/80) – 6/36 (20/120); **Severe** = visual acuity 6/60 (20/200) – 3/60 (20/400); **Profound** = visual acuity <3/60 (20/400))

4.1.6 Visual field defect (Right eye): Yes ___ No ___ NA ___

4.1.7 Visual field defect (Left eye): Yes ___ No ___ NA ___

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/osseous or membranous): ___________________________________

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 (small bones) 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 ___

4.3.6 Hearing impairment (R. ear): Normal ___ Mild ___ Moderate ___ Severe ___ Profound ___ NA ___

4.3.7 Hearing impairment (L. ear): Normal ___ Mild ___ Moderate ___ Severe ___ Profound ___ NA ___

(**Mild** = hearing loss 20-40 dB; **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 section 5)

4.4.1 Weak chewing/sucking: Yes ___ No ___ NA ___

4.4.1.1 Problems with sense of smell 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) (NOTE: NA = NOT AVAILABLE)

Male

5.1 Micro penis: Yes___ No___ Not applicable___ 5.2 Cryptorchidism: Yes___ No___ Not applicable___

Female

5.3 Hypoplastic labia: Yes ___ No ___ Not applicable ___

Heart Anomalies

5.4 Minor cardiovascular malformations (e.g., PDA, PFO, small ASD or small VSD (no repair needed))

5.4.1 Patent Ductus Arteriosus (PDA) Yes ___ No ___ NA ___
5.4.2 Patent Foramen Ovale (PFO) Yes ___ No ___ NA ___
5.4.3 small ASD Yes (primum defect) ___ Yes (secundum defect) ___ No ___ NA ___
5.4.4 small VSD Yes (membranous) ___ Yes (muscular) ___ No ___ NA ___
5.4.5 other minor (e.g., vascular ring) describe:
____

5.5 Major cardiovascular malformations: (e.g., Tetralogy of Fallot, AV canal, aortic arch anomaly, large repaired ASD, large repaired VSD)

5.5.1 Tetralogy of Fallot Yes ___ No ___ NA ___
5.5.2 AV canal Yes ___ No ___ NA ___
5.5.3 Aortic arch anomaly Yes ___ No ___ NA ___
5.5.4 ASD – large repaired Yes (primum defect) ___ Yes (secundum defect) ___ No ___ NA ___
5.5.5 VSD – large repaired Yes (membranous) ___ Yes (muscular) ___ No ___ NA ___
5.5.6 other major (describe):
____

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/foot anomalies (e.g., polydactyly, thumb hypoplasia): Yes ___ No ___ NA ___

Describe: ____

6.3 Spine anomalies (e.g., hemivertebrae): Yes ___ No ___ NA ___

6.31 Scoliosis Yes ___ No ___ NA ___ Describe: ____

6.32 Osteoporosis Yes ___ No ___ NA ___

6.33 Describe DEXA scan results and treatment for osteoporosis:
____

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 6A – IMAGING FINDINGS (E.G., XRAY, CT, MRI)

6A.1 Temporal bone abnormality Yes ___ No ___ NA ___ Describe: ______________________________________

6A.2 Semicircular canals anomaly: Yes ___ No ___ NA ___ Describe: ____________________________________

6A.3 Mondini defect (i.e., decreased cochlear turns): Yes ___ No ___ NA ___ Describe:______________________

6A.4 Olfactory bulb anomaly: Yes ___ No ___ NA ___ Describe: ____

6A.5 Other MRI findings (e.g., atrophy, brainstem anomalies, etc.) Yes ___ No ___ NA ___
Describe: ____

SECTION 7 – FAMILY HISTORY

7.1 Similarly affected relatives (with any features of CHARGE, even if mild): Yes ___ No ___ NA ___
Describe: ____

7.2 Other significant family history (e.g., genetic disorders, hearing impairments, developmental disability/learning, etc.)

7.2.1 Maternal: Yes ___ No ___ NA ___ Describe: ____
____

7.2.2 Paternal: 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 (applicable for age 3 yrs or older): Yes ___ No ___ NA ___

9.2 Major sleep problems: Yes ___ No ___ NA ___

9.3 Repetitive/obsessive/compulsive (talk or movement; applicable for age 3 yrs or older): 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 ___

11.3 Migraine: Yes ___ No ___ NA ___

11.4 Brain 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, amount, 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): _____________________________________

13.11 Contact with dry-cleaning substances: Yes____ No____ NA____

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.

(CHARGE Blake/July 2005)