CONGENITAL MYOTONIC DYSTROPHY (CMD)

CASE DEFINITION FOR CONGENITAL MYOTONIC DYSTROPHY (CMD)
Report any children up to the age of three years with a new diagnosis of CMD. A diagnosis of CMD will be included if children have both of the following clinical and genetic criteria:

- Symptoms of myotonic dystrophy in the newborn period (≤ 30 days), such as hypotonia, feeding or respiratory difficulties, requiring hospitalization to a ward or to the neonatal intensive care unit for greater than 72 hours.*
- CMD genetic tests confirming an expanded trinucleotide CTG repeat in the DMPK gene in the child or the mother. An expanded CTG repeat size is >200 repeats or E1-4 classification (E1: 200-500; E2: 500-1,000; E3: 1,000-1,500; E4: >1,500).

* Infant does not necessarily need to be born during the surveillance period, as a diagnosis confirmed later may clearly show neonatal complications requiring admissions.

SECTION 1 – DEMOGRAPHIC INFORMATION

1.1 Date of birth: ______/_____/______
1.2 Sex: Male ___ Female ___
1.3 Province/Territory of residence: ____________________

SECTION 2 – MATERNAL HISTORY OF MYOTONIC DYSTROPHY

2.1 Clinical information:
   2.1.1 Diagnosis prior to pregnancy ___ ___ ___
   2.1.2 Age of mother at diagnosis: ______ years ___
   2.1.3 Number of trinucleotide repeats: _________ ___
   2.1.4 Previous pregnancy ___ ___ ___
       If yes, specify number: ______
   2.1.5 Previous interrupted pregnancies ___ ___ ___
   2.1.6 Genetic counseling prior to this pregnancy ___ ___ ___
   2.1.7 Prenatal genetic testing with this pregnancy ___ ___ ___
       If yes, please specify: chorionic villous sampling ___ amniocentesis ___ number of repeats ___

SECTION 3 – PREGNANCY AND BIRTH HISTORY

Mother
3.1 Age: ______ years Parity: G ____ P ____ A ____
SECTION 3 – PREGNANCY AND BIRTH HISTORY (CONT’D)

3.2 Clinical information:  
3.2.1 Polyhydramnios  
3.2.2 Decreased fetal movements  
3.2.3 Length of labour: _______ hours  
3.2.4 Prolonged rupture of membranes  
3.2.5 Evidence of fetal distress  
3.2.6 Type of delivery:  vaginal ____  forceps utilization ____  vacuum extraction ____  caesarian ____  unknown ____  

Newborn  
3.3 Gestational age: _______ weeks  
3.4 Apgar score:  1 min. ___  5 min. ___  10 min. ___  20 min. ___  Unknown ___  
3.5 Cord gas:  pH:______  base excess: _____________  Unknown ___  
3.6 Next gas:  pH:______  base excess: _____________  Unknown ___  
3.7 Resuscitation interventions:  None ___  Oxygen only ___  c-PAP only ___  Bag and mask ventilation ___  Intubation and ventilation ___  Cardiac compression ___  Unknown ___  
3.8 Resuscitation medications:  No ___  Yes ___  Unknown ___  

SECTION 4 – PATIENT DIAGNOSIS  
4.1 Child is the first diagnosis in the family:  No ___  Yes ___  Unknown ___  
4.2 EMG:  Not done ___  Done (results): ______________________________  
4.3 Biopsy:  Not done ___  Done (results): ______________________________  
4.4 Genetic testing:  Number of CTG repeats: _______  or  classification E1 ___  E2 ___  E3 ___  E4 ___  

SECTION 5 – INITIAL DIAGNOSES AND MANAGEMENT  
5.1 Respiratory diagnosis (check all that apply):  
Pulmonary hypoplasia ____  Pneumothorax ____  Pneumonia ____  Raised hemidiaphragm ____  
Bronchopulmonary dysplasia ____  Other, please specify: ______________________________  
5.2 Respiratory therapy:  None ___  
If multiple therapies, please specify temporal order:  
<table>
<thead>
<tr>
<th>Type</th>
<th>Approximate duration</th>
<th>Order</th>
<th>Current</th>
</tr>
</thead>
<tbody>
<tr>
<td>Oxygen</td>
<td>_____________________</td>
<td>______</td>
<td>______</td>
</tr>
<tr>
<td>c-PAP</td>
<td>_____________________</td>
<td>______</td>
<td>______</td>
</tr>
<tr>
<td>Assisted ventilation (any type, for any portion of day)</td>
<td>_____________________</td>
<td>______</td>
<td>______</td>
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<tr>
<td>Respiratory stimulants (type:____________________)</td>
<td>_____________________</td>
<td>______</td>
<td>______</td>
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<tr>
<td>Other medications (surfactant ___  steroids ___  nitrous oxide ___ )</td>
<td>_____________________</td>
<td>______</td>
<td>______</td>
</tr>
<tr>
<td>Surgery: Tracheostomy ___  Other ___ (type:____________________)</td>
<td>_____________________</td>
<td>______</td>
<td>______</td>
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</tbody>
</table>
SECTION 5 – INITIAL DIAGNOSES AND MANAGEMENT (cont’d)

5.3 Feeding diagnosis (check all that apply):
- Delayed gastric emptying ____
- Poor intestinal transport ____
- Constipation ____
- Gastro-esophageal reflux ____
- Necrotizing enterocolitis ____
- Unknown ____
- Other, please specify: _____________________________________________________________________________

5.4 Feeding therapy: Normal oral feeding ____
If multiple therapies, please specify temporal order:

<table>
<thead>
<tr>
<th>Type</th>
<th>Approximate duration</th>
<th>Order</th>
<th>Current</th>
</tr>
</thead>
<tbody>
<tr>
<td>Naso-gastric tube feeding</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Gastrostomy/jejunostomy tube feeding</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Total parenteral nutrition</td>
<td></td>
<td></td>
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<tr>
<td>Prokinetic agent: (type:____________)</td>
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</tbody>
</table>
| Surgery: Fundoplication ____ Other ____ (type:_________________________________________________________)

5.5 Other complications
- CNS: Seizures ____  Apnea ____  Intraventricular hemorrhage ____  Ventriculomegaly ____
- MRI patterns of hypoxic ischemic encephalopathy ____
- Cardiac: Structural abnormality ____  Dysrhythmia ____  Hypotension ____
- Infections: Sepsis ____  Meningitis ____  Urinary tract infection ____
- Other, please specify: _____________________________________________________________________________

SECTION 6 – OUTCOME

6.1 Hospital stay: Total number of days: _______  Days in intensive care unit: _______
6.2 Still in hospital ____  Home ____  Transfer to another facility, please specify: _____________________________
6.3 Age at last follow-up: _______ months / years
6.4 Medical complications as of last follow-up: ___________________________________________________________
________________________________________________________________________________________________
________________________________________________________________________________________________

6.5 Deceased: ____
If yes: Age at time of death _______ (weeks / months)
- Duration of hospitalization prior to death: _______ (weeks / months / years)
- Cause of death: ________________________________________________________________
- Withdrawal of life support: No ___  Yes ____
If yes, please specify reason: ________________________________________________________________

SECTION 7 – REPORTING PHYSICIAN

First name_________________________ Surname_________________________
Address____________________________________________________________________
City_____________________________ Province________________________ Postal code________
Telephone number__________________ Fax number_______________________
E-mail___________________________ Date completed____________________

Thank you for completing this form.