CASE DEFINITION FOR MEDIUM CHAIN ACYL-COENZYME A DEHYDROGENASE DEFICIENCY (MCADD)

Report any patients newly diagnosed with MCAD deficiency following investigations initiated due to any of the following:
newborn screening, clinical symptoms, diagnosis in an affected family member or postmortem diagnosis.

A child will be considered to have a diagnosis of MCAD deficiency if at least ONE of the following biochemical/genetic diagnostic criteria are met:

1. Elevated plasma C6 to C10 acylcarnitines with predominance of C8 (octanoylcarnitine).
2. Elevated urinary organic acids: phenylpropionylglycine, suberylglycine, hexanoylglycine, and medium chain dicarboxylic acids (C6>C8>C10).
3. Molecular genetic studies confirming the presence of the 985 A>G mutation, or other less common mutations.
4. Skin fibroblasts acylcarnitine probe assay demonstrating accumulation of characteristic acylcarnitines.
5. Skin fibroblasts enzyme studies showing reduced activity of MCAD.

In the presence of the following clinical features or biochemical findings:

1. Vomiting, hepatomegaly and altered sensorium.
2. Hypoglycemia, elevated liver enzymes.

SECTION 1 – DEMOGRAPHIC INFORMATION

1.1 Date of birth: _____ / _____ / ______  1.2 Sex: Male ___  Female ___

1.3 Province/Territory of residence: ______________________________

1.4 Ethnicity of parents

Mother:
First Nations _____  Innu _____  Inuit _____  Métis _____  Asian _____  Black _____  Caucasian _____
Latin American _____  Middle Eastern _____  Other (specify): _____________________________________

Father:
First Nations _____  Innu _____  Inuit _____  Métis _____  Asian _____  Black _____  Caucasian _____
Latin American _____  Middle Eastern _____  Other (specify): _____________________________________

SECTION 2 – FAMILY HISTORY

2.1 Presence of similarly affected siblings or other relatives in the family

No ___  Yes ___  Unknown ___

If yes, specify, number _____  and relationship to patient ______________________________

2.2 History of sudden unexplained death of a child in the family

No ___  Yes ___  Unknown ___

If yes, specify, number _____  and relationship to patient ______________________________
SECTION 3 – MEDICAL HISTORY

3.1 Gestational Age:
<37 weeks ___ or ≥37 weeks ___

3.2 First suspicion of a MCAD deficiency diagnosis:
\[ \frac{DD}{MM/YY} \] or age: _____ months _____ years

3.3 Reasons for initiating MCAD deficiency diagnostic investigations (please check ALL that apply)
___ routine newborn screening
___ clinical symptoms
___ family history of affected sibling(s)/relative(s)
___ post-mortem investigations
___ other (please specify) _______________________________________________________

SECTION 4 – CLINICAL FINDINGS

4.1 Please indicate ALL signs and symptoms at the first clinical episode of MCAD deficiency:

<table>
<thead>
<tr>
<th>Signs and Symptoms</th>
<th>No</th>
<th>Yes</th>
<th>Unknown</th>
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<tbody>
<tr>
<td>Hepatomegaly</td>
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<td>Apnea</td>
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<td>Respiratory arrest</td>
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<td>Cardiac arrest</td>
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<tr>
<td>Other (please specify)</td>
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</tbody>
</table>

SECTION 5 – INVESTIGATIONS DONE AT PRESENTATION

5.1 Blood sugar
No ___ Yes ___ Unknown ___
↓ If yes, please specify lowest value: _______ mmol/L

5.2 Urinary ketones
No ___ Yes ___ Unknown ___
↓ If yes, please specify: Absent ____ Present ___

5.3 Liver function tests – ALT/AST
No ___ Yes ___ Unknown ___
↓ If yes, please specify results – ALT: _______ U/L
    - AST: _______ U/L

5.4 Blood ammonia levels
No ___ Yes ___ Unknown ___
↓ If yes, please specify results: _______ µmol/L

5.5 Carnitine levels
(total and free)
No ___ Yes ___ Unknown ___
↓ If yes, please specify results – Total: _______ µmol/L
    - Free: _______ µmol/L
SECTION 5 – INVESTIGATIONS DONE AT PRESENTATION (cont’d)

5.6 Plasma acylcarnitine profile  No ___ Yes ___ Unknown ___ 
   ↓
   If yes, please specify results: Normal ____ Abnormal ____

5.7 Urinary organic acids  No ___ Yes ___ Unknown ___ 
   ↓
   If yes, please specify results: Normal ____ Abnormal ____

5.8 Genetic mutation analysis  No ___ Yes ___ Unknown ___ 
   ↓
   If yes, please specify:
   homozygous for 985 A>G ___ heterozygous for 985 A>G ___ no mutation detected ___
   other mutation detected (please specify): ____________________________________________

5.9 Skin fibroblasts enzyme study  No ___ Yes ___ Unknown ___ 
   ↓
   If yes, please specify: ________________________________________________

SECTION 6 – MANAGEMENT AND OUTCOME

6.1 At the first clinical episode, the child was:
   Admitted to hospital  No ___ Yes ___ Unknown ___ 
   ↓
   If yes, please specify length of stay: _____ days _____ weeks

   Admitted to intensive care unit  No ___ Yes ___ Unknown ___ 
   ↓
   If yes, please specify length of stay: _____ days _____ weeks

6.2 Medical status at time of reporting, if available:
   Normal No ___ Yes ___ Unknown ___ 
   Seizures No ___ Yes ___ Unknown ___ 
   Hemiparesis No ___ Yes ___ Unknown ___ 
   Speech problems No ___ Yes ___ Unknown ___ 
   Developmental delay No ___ Yes ___ Unknown ___ 
   Others, please specify________________________

6.3 Child is deceased: No ___ Yes ___ Unknown ___ 
   ↓
   If yes, age at time of death: ___ weeks ___ months
   Cause of death: ________________________________

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.