Infantile and later-onset paediatric Pompe disease (glycogen storage disease type II)

CANADIAN PAEDIATRIC SURVEILLANCE PROGRAM
100-2305 St-Laurent Blvd.
Ottawa, ON K1G 4J8
Tel: (613) 526-9397, ext. 239
Fax: (613) 526-3332
cpsp@cps.ca
www.cpsp.cps.ca

REPORTING INFORMATION
(To be completed by CPSP staff)

Report number:	
Month of reporting:	
Province:	
Today's date:	

Please complete the following sections for the case identified above.
Strict confidentiality of information will be assured.

CASE DEFINITION FOR POMPE DISEASE (PD)

Report any patient **(new or previously diagnosed)** less than 18 years of age meeting the following criteria:

1. **Genetic criteria:** Pathogenic mutations affecting both *GAA* genes (encodes the acid alpha-glucosidase protein) as determined by sequence analysis or deletion/duplication analysis

AND/OR

2. **Biochemical criteria:** Measurement of acid alpha-glucosidase (GAA) enzyme activity performed on one or more of:
   - Dried blood spot GAA enzyme activity assay
   - Whole blood GAA enzyme activity assay
   - Skin biopsy (fibroblast culture) GAA enzyme activity assay
   - Muscle biopsy GAA enzyme activity assay

Exclusion criteria

Clinical evidence of proximal muscle weakness without genetic or biochemical confirmation of disease

NOTE: DATA ARE BEING COLLECTED ON INCIDENT AND PREVALENT CASES. PLEASE REPORT ALL NEW AND/OR PREVIOUSLY DIAGNOSED CASES.

SECTION 1 – DEMOGRAPHIC INFORMATION

1.1 Date of birth: ____ /_____ /_____
DD MM YYYY

1.2 Sex: Male___ Female___

1.3 First 3 digits of patient's postal code: ___ ___ ___

1.4 Population groups (check all that apply):

☐ Arab	☐ Black	☐ Chinese	☐ Filipino
☐ Japanese	☐ Korean	☐ Latin American	☐ White
☐ First Nations	☐ Inuit	☐ Métis	☐ Unknown
☐ Southeast Asian (e.g., Vietnamese, Cambodian, Laotian)	☐ South Asian (e.g., East Indian, Pakistani, Sri Lankan)	☐ West Asian (e.g., Iranian, Afghan)	☐ Other, specify: _____

1.5 Is this child the first known case of this disease for his/her family? Yes___ No___ Unknown___

SECTION 2 – DIAGNOSIS

2.1 Age at symptom onset: ____months OR ____years

2.2 Age at diagnosis: ____months OR ____years

2.3 Date of clinical diagnosis: ____ /_____ /_____
DD MM YYYY

2.4 Date of biochemical OR genetic confirmation: ____ /_____ /_____
DD MM YYYY

2.5 Who made the initial diagnosis?
___Paediatrician ___Paediatric neurologist ___Physiatrist

___Developmental paediatrician ___Metabolic specialist ___Geneticist
___Adult neurologist ___Other (specify): _____

2.6 Was an electromyography (EMG) done? Yes___ No___ Unknown___
If yes, were results: Normal___ Abnormal___; (specify):_____

2.7 Was a muscle biopsy done? Yes___ No___ Unknown___
*If ye*s, what muscle was biopsied?_____
Normal___ Abnormal___; (specify): _____

2.8 Was a serum creatine kinase (CK) level done?: Yes___ No___ Unknown___
If abnormal, please provide value(s): _____

2.9 How was the diagnosis of Pompe disease made? (Check all that apply)
___Blood enzyme analysis (dried blood spot) ___Fibroblast (skin biopsy) enzyme analysis
___Genetic analysis (Sanger sequencing) ___Genetic analysis (whole exome sequencing)
___Blood enzyme analysis (whole blood) ___Muscle enzyme analysis
___Genetic analysis (NGS gene panel) ___Newborn screening
___Unknown

2.10 Residue enzyme activity (% normal) _____ Unknown___

2.11 Laboratory performing **enzyme** analysis:
___London Health Sciences Centre (London, ON) ___BC Children's Hospital (Vancouver, BC)
___CHU Sainte-Justine (Montreal, QC) ___Dynacare Next (Montreal, QC)
___Unknown ___Other, (specify):_____
___Not performed

2.12 Laboratory performing **genetic** testing:
___Sherbrooke, QC ___Edmonton, AB
___Unknown ___Other, (specify):_____
___Not performed

2.13 List mutation(s) in *GAA* genes: Unknown___
1. _____
2. _____

SECTION 3 – SYMPTOMATOLOGY AT DIAGNOSIS

3.1 Age at first symptom onset: Birth____ Months____ or Years____

3.2 Most prominent sign/symptom/laboratory abnormalities **AT** the moment of the diagnosis (select **ONE**):
___Hypotonia ___Feeding difficulty ___Respiratory distress
___Gross motor delay ___Cardiomyopathy/heart failure ___Hepatomegaly
___Proximal muscle weakness ___High serum creatine kinase

3.3 Select **ALL** signs/symptoms/laboratory abnormalities **PRIOR** To diagnosis (check all that apply):
___Hypotonia ___Cardiomyopathy ___Feeding difficulty
___Failure to thrive ___Respiratory distress ___Never able to sit
___Never able to crawl ___Never able to stand ___Never able to walk
___Never able to run ___Gross motor regression ___Scapular winging
___Proximal weakness (arms) ___Rhabdomyolysis ___Proximal weakness (legs)
___Distal weakness (hands) ___Distal weakness (feet) ___Joint contracture(s)
___Hepatomegaly ___Scoliosis ___Spinal rigidity
___Enlarged tongue ___High serum creatine kinase
___Shortness of breath with exercise

3.4 Select all the medical devices used by the patient **PRIOR** to the diagnosis (check all that apply):
___Wheelchair ___NG or G-tube required ___Tracheostomy
___Non-invasive ventilation (i.e., CPAP or BiPAP required)

3.5 Serum creatine kinase level PRIOR to diagnosis:
Normal___ Abnormal___ Unknown___ Not performed___
If abnormal, please provide value(s):_____

SECTION 4 – MEDICAL SURVEILLANCE

☐ NOT APPLICABLE *(Mark this checkbox if this case represents an **incidence case** and medical surveillance data is not available)*

Cardiac:

4.1 Has the patient developed clinical signs/symptoms of cardiac dysfunction? Yes___ No___ Unknown___
If yes, please specify the age at which symptoms appeared and list the signs/symptoms:
_____

4.2 Has an electrocardiogram (ECG) and/or echocardiogram ever been performed? Yes___ No___ Unknown___
If yes, age of most recent echocardiogram: ____months, ____years
age of most recent ECG: ____months, ____years

4.3 If an ECG and /or echocardiogram was performed, select ALL positive findings on the most recent echocardiogram (check all that apply):
___Sinus arrhythmia ___Left ventricular hypertrophy
___Right bundle branch block ___Wolff-Parkinson-White (WPW) syndrome
___Other, specify:_____

Pulmonary:

4.4 Has the patient developed clinical signs/symptoms of respiratory dysfunction? Yes___ No___ Unknown___
If yes, please specify the age at which symptoms appeared and list the signs/symptoms:
_____

4.5 Has a pulmonary function test (PFT) ever been performed? Yes___ No___ Unknown___
If yes, age of most recent PFT: ____months, ____years
Most recent forced vital capacity (FVC): _____ L
Most recent forced vital capacity (FVC): _____ % predicted

4.6 Please list all respiratory supports ever required by patient:
___No supports required ___Non-invasive ventilation (night only)
___Non-invasive ventilation (night + some day time use) ___Tracheostomy and mechanical ventilation
___Non-invasive ventilation (>20 hours per day)

SECTION 5 – MEDICAL MANAGEMENT

5.1 After diagnosis, was the patient initiated on enzyme replacement therapy (ERT) ?
Yes___ No___ Unknown___
If no, specify reason:
___Patient/family refused treatment ___Patient was Cross-reactive immunological material (CRIM) negative
___Could not afford cost of ERT ___Unknown

5.2 Was clinical improvement noted following ERT initiation? Yes___ No___ Unknown___
If yes, what measure(s) was used to determine improvement?
_____
_____

5.3 Is the patient currently receiving ERT? Yes___ No___ Unknown___
If no, specify reason:
___Patient did not improve with ERT ___Patient was CRIM negative
___Patient/family refused further treatment ___Patient/family reported ERT-related side effects
___Could not afford cost of ERT ___Patient is deceased

5.4 What other therapeutic options were employed? (check all that apply)

___Digoxin ___Other antiarrhythmic ___Diuretics
___ACE-inhibitors ___Bronchodilators ___Cough Assist In-Exsufflator
___Glucocorticoids ___Beta agonists ___BiPAP
___CPAP ___Tracheostomy ___Nasoduodenal or gastrojejunal feeding
___Physical therapy ___Occupational therapy ___Speech therapy
___Respiratory therapy ___Orthotic intervention (e.g., ankle-foot orthosis)
___Vitamin D/Calcium supplementation

5.5 Has the patient ever experienced complications following anaesthesia? Yes___ No___ Unknown___

If yes, please describe the reason for the surgery and the anesthetic complication:

_____
_____
_____

SECTION 6 – OUTCOME

6.1 Age at last visit: ____months, ____years

6.2 Is the patient alive? Yes___ No___ Unknown___

If no, age of patient at death: ____months, ____years

Cause of death:
___Cardiac failure resulting in cardiorespiratory arrest
___Respiratory failure resulting in cardiorespiratory arrest
___Pneumonia
___Other, (specify) _____
___Unknown

___ **I agree to be contacted by the CPSP for further information.**

___ **I do not wish to be contacted by the CPSP for further information.**

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.