CONGENITAL MYOTONIC DYSTROPHY (CMD)

CANADIAN PAEDIATRIC SURVEILLANCE PROGRAM

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Mother

3.1 Age: _____ years

Web site: www.cps.ca/english/cpsp

REPORTING INFORMATION (To be completed by the CPSP Senior Coordinator)								
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 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.

		EMOGRAPHIC INFORMATION					
1.1	Date o	ate of birth: / / 1.2 Sex: Male Female					
1.3	Provin	Province/Territory of residence:					
ECTIO	N 2 – M	ATERNAL HISTORY OF MYOTONIC DYSTROPHY					
2.1	Clinica	l information:	No	Yes	Unknown		
	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	amnioce	entesis	number of repeats		

Parity: G ____ P ___ A ____

SECTION 3 – PREGNANCY AND BIRTH HISTORY (CONT'D)

3.2	Clinical information:	No	Yes Unknowr	1				
	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							
	If yes, please specify: bradycardia ta	achycardia	scalp gas					
	3.2.6 Type of delivery: vaginal forceps ut unknown	ilization	vacuum extractio	on caesarian				
New	vborn							
3.3	Gestational age: weeks Birth weig	ght: gr	ams					
3.4	Apgar score: 1 min 5 min 10 min							
3.5	Cord gas: pH: base excess:		Unknown					
3.6								
	If yes, please specify time from birth: (minutes / hours)							
3.7								
	Intubation and ventilation Cardiac compression Unknown							
3.8	Resuscitation medications: No Yes Ur	nknown						
	If yes, please specify:							
SECTIO 4.1	ON 4 – PATIENT DIAGNOSIS Child is the first diagnosis in the family: No Y If yes, please specify: Date of genetic confirmation	/	/					
4.2	EMC: Not dono Dono (recults):	DD MM						
4.2	EMG: Not done Done (results):							
4.4	Biopsy: Not done Done (results): or classification E1 E2 E3 E4							
7,7	deficite testing. Number of eTd repeats.	or classifica						
SECTIO	ON 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:							
	Туре	Appro	ximate duration	Order Current				
	Oxygen							
	c-PAP							
	Assisted ventilation (any type, for any portion of da							
	Respiratory stimulants (type:							
	Other medications							
	(surfactant steroids nitrous oxide)							
	Surgery: Tracheostomy Other (type:							

SECTION 5 - INITIAL DIAGNOSES AND MANAGEMENT (cont'd) Feeding diagnosis (check all that apply): Delayed gastric emptying ____ Poor intestinal transport ____ Constipation ____ Gastro-esophageal reflux ____ Necrotizing enterocolitis __ Unknown Other, please specify: Feeding therapy: Normal oral feeding 5.4 If multiple therapies, please specify temporal order: **Type Approximate duration** Order Current Naso-gastric tube feeding Gastrostomy/jejunostomy tube feeding Total parenteral nutrition Prokinetic agent: (type: 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** Hospital stay: Total number of days: Days in intensive care unit: 6.1 Still in hospital ____ Home ___ Transfer to another facility, please specify: _____ 6.2 Age at last follow-up: ___ months / years 6.3 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

Thank you for completing this form.

City_____ Province_____ Postal code______
Telephone number____ Fax number_____

E-mail Date completed