Fragile X syndrome (FXS)

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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 FRAGILE X SYNDROME

Report any new patient less than 18 years of age with diagnosed fragile X syndrome (FXS) meeting the following criteria:

1) **Genetic criteria:** Male or female, with laboratory confirmation of a CGG repeat allele in the full mutation size range (>200 repeats), including mosaicism

AND

2) **Clinical criteria, one** of the following:
   - Global developmental delay, manifesting as the clinical impression of delays in two or more domains of development **or**
   - Intellectual disability, mild, moderate or severe, diagnosed through standardized psychological testing **or**
   - Asymptomatic infant, tested because of a positive family history, including prenatally diagnosed cases

Exclusion criteria

Clinical evidence of global developmental delays or intellectual disability with laboratory confirmation of a CGG repeat allele in the normal or premutation size range.

Month first seen _____

SECTION 1 – DEMOGRAPHIC INFORMATION

1.1 Date of birth: ____ /____ /_____ 1.2 Sex: Male ___ Female ___
DD MM YYYY

1.3 Province/Territory of residence: _____

1.4 Postal code of home address – first three digits only: __ __ __

1.5 Ethnicity

First Nations ___ Innu ___ Inuit ___ Métis ___ Chinese ___ Japanese ___

Other Oriental ___ East Indian ___ Black ___ Caucasian ___ Latin American ___

Middle Eastern ___ Other, specify: _____ Unknown ___

SECTION 2 – CLINICAL PRESENTATION

2.1 Age of patient at first parental concern: _____ years _____ months

2.2 Date of fragile X test result: ____ /____ /_____
DD MM YYYY

2.3 Did you order the diagnostic test: Yes ___ No ___

If not, specify who did: Family doctor ___ Other paediatrician ___ Neurologist ___ Geneticist ___

Psychiatrist ___ Other_____

SECTION 2 – CLINICAL PRESENTATION (cont'd)

2.4 Reason for referral to you (reporting physician) – check all that apply:

___ Depressed mood
___ Developmental delay (delayed milestones)
___ Intellectual disability
___ Attention deficit hyperactivity disorder
___ Anxiety
___ Autism spectrum features (social, language and repetitive behaviours)
___ Seizures
Other, specify: _____

2.5 Referral source

Family physician ___ Medical geneticist ___ Neurologist ___ Psychiatrist ___
Public health nurse ___ Family member ___ Speech therapist ___ Physiotherapist ___
Occupational therapist ___ Infant/child development professional ___
Other, specify: _____

SECTION 3 – CLINICAL FINDINGS

3.1 Physical features at presentation – check all that apply:

Tall stature (height >95-98 percentile) ___ Height _____ cm Percentile on growth chart _____
Macrocephaly (HC >98 percentile) ___ HC _____cm Percentile on growth chart _____
Long face ___ High-arched palate ___ Large or prominent ears ___ Macro-orchidism ___
Pes planus (flat feet) ___ Hyper-extensible finger joints ___

3.2 Medical diagnoses – check all that apply, current or past:

	Yes	No	Unknown
• Seizures	___	___	___
• Gastroesophageal reflux	___	___	___
• Cleft palate	___	___	___
• Mitral valve prolapse	___	___	___
• Inguinal hernia	___	___	___
• Scoliosis	___	___	___
• Sleep apnea	___	___	___
• Recurrent otitis media	___	___	___
• Hearing loss	___	___	___
• Speech/communication problems	___	___	___
• Eye/vision problems	___	___	___
If yes, specify: _____			
• ADHD	___	___	___
• Anxiety	___	___	___
• Autism spectrum disorder*	___	___	___
• Other mental health problems	___	___	___
If yes, specify: _____			

*Autism spectrum disorder confirmed using standardized assessment tools (ADI-R and ADOS)

SECTION 3 – CLINICAL FINDINGS (cont'd)

3.3 Indicate the classification of intellectual disability confirmed by standardized psychometric testing:

3.3.1 Age at most recent psychometric testing: _____ years _____ months

3.3.2 Full-scale IQ or general conceptual ability: _____

3.3.3 Description of classification of intellectual disability:

severity unspecified ____ mild (IQ 50-55 to 70) ____ moderate (IQ 35-40 to 50-55) ____

severe (IQ 20-25 to 35-40) ____ profound (IQ <20 or 25) ____

3.4 Indicate which of the following have been used in the patient's care – check all that apply:

	Yes	No	Unknown
• Developmental paediatrician	___	___	___
• Psychologist	___	___	___
• Psychiatrist	___	___	___
• Paediatric neurologist	___	___	___
• Clinical geneticist	___	___	___
• Genetic counseling of family members	___	___	___
• Occupational therapist	___	___	___
• Speech therapist	___	___	___
• Autism intervention program	___	___	___
• Support group for FXS	___	___	___
• Specialty FXS clinic	___	___	___

• Other, specify: _____

3.5 Current medications and non-prescription products

	Yes	No	Unknown
• Medications	___	___	___
• Non-prescription medications	___	___	___
• Natural health products	___	___	___
• Vitamins	___	___	___
• Other	___	___	___

If yes, specify name and dosage:

_____

_____

_____

SECTION 4 – FAMILY HISTORY

4.1 Indicate whether there are family members diagnosed with the following:

	Yes	No	Unknown
Fragile X syndrome	___	___	___
If yes, specify relationship to child: _____			
FXPOI (fragile X-associated primary ovarian insufficiency)	___	___	___
Associated with fragile X premutation in females and including irregular menstrual cycles, infertility, and menopause before age 40 years			
If yes, specify relationship to child: _____			

SECTION 4 – FAMILY HISTORY (cont'd)

	Yes	No	Unknown
FXTAS (fragile X-associated tremor-ataxia syndrome) Associated with fragile X premutation primarily affecting older males and including balance problems, intention tremor, and cognitive impairment	___	___	___

If yes, specify relationship to child: _____

SECTION 5 – ADDITIONAL COMMENTS OR FEEDBACK

_____

_____

_____

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

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

SECTION 6 – REPORTING PHYSICIAN

First name_____ Surname_____

Address_____

City_____ Province_____ Postal code_____

Telephone number_____ Fax number_____

E-mail_____ Date completed_____

Thank you for completing this form.

(FXS 2012-04)