Fragile X syndrome (FXS)

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.

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:

<table>
<thead>
<tr>
<th></th>
<th>Yes</th>
<th>No</th>
<th>Unknown</th>
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</thead>
<tbody>
<tr>
<td>Seizures</td>
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<tr>
<td>Gastroesophageal reflux</td>
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<tr>
<td>Cleft palate</td>
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<td></td>
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<td>Mitral valve prolapse</td>
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<td>Inguinal hernia</td>
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<td>Scoliosis</td>
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<td>Sleep apnea</td>
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<tr>
<td>Recurrent otitis media</td>
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<tr>
<td>Hearing loss</td>
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<tr>
<td>Speech/communication problems</td>
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<td>Eye/vision problems</td>
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</tbody>
</table>

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:

<table>
<thead>
<tr>
<th>Service</th>
<th>Yes</th>
<th>No</th>
<th>Unknown</th>
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<tbody>
<tr>
<td>Developmental paediatrician</td>
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<tr>
<td>Psychologist</td>
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<tr>
<td>Psychiatrist</td>
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<tr>
<td>Paediatric neurologist</td>
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<tr>
<td>Clinical geneticist</td>
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<tr>
<td>Genetic counseling of family members</td>
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<td>Occupational therapist</td>
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<td>Speech therapist</td>
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<td>Autism intervention program</td>
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<td>Support group for FXS</td>
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<td>Specialty FXS clinic</td>
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<td>Other, specify:</td>
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</table>

3.5 Current medications and non-prescription products

<table>
<thead>
<tr>
<th>Product</th>
<th>Yes</th>
<th>No</th>
<th>Unknown</th>
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</thead>
<tbody>
<tr>
<td>Medications</td>
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<tr>
<td>Non-prescription medications</td>
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<tr>
<td>Natural health products</td>
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<tr>
<td>Vitamins</td>
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<tr>
<td>Other</td>
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</table>

If yes, specify name and dosage:

____________________________________________________________________________________
____________________________________________________________________________________
____________________________________________________________________________________

**SECTION 4 – FAMILY HISTORY**

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

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Yes</th>
<th>No</th>
<th>Unknown</th>
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</thead>
<tbody>
<tr>
<td>Fragile X syndrome</td>
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<tr>
<td>FXPOI (fragile X-associated primary ovarian insufficiency)</td>
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</table>

If yes, specify relationship to child: ____________________________
SECTION 4 – FAMILY HISTORY (cont'd)

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)