

## **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.