

Before, Between & Beyond Pregnancy
**The National Preconception Curriculum and Resources Guide
for Clinicians**

**Guidance for Preconception Fragile X
Carrier Screening**

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This guidance should not substitute for clinical judgments or expert consultation

Summary of:

ACOG Committee Opinion

Screening for Fragile X Syndrome

Obstetrics & Gynecology. 2006. 107(6);1483-1485

Recommendations:

1. DNA-based molecular analysis (e.g., Southern blot analysis and polymerase chain reaction) is the preferred method of diagnosis for fragile X syndrome. In the rare cases where there is discordance between the triplet repeat number and the methylation status, the patient should be referred to a genetic specialist.
 2. Patients with a family history of mental retardation or a history of fragile X associated mental retardation should receive genetic counseling and should be offered genetic testing to assess their risk for having an affected child.
 3. Prenatal testing for fragile X syndrome by amniocentesis or CVS should be offered to all known carriers of the fragile X premutation or full mutation. Although it is reliable for determining the number of triplet repeats, CVS may not adequately determine the methylation status of the FMR1 gene.
 4. Testing for fragile X syndrome should be considered in any child with developmental delay of unknown etiology, autism, autistic-like behavior or any individual with mental retardation of unknown etiology.
 5. If a woman has ovarian failure or an elevated follicle-stimulating hormone level before the age of 40 years without a known cause, fragile X carrier screening should be considered to determine whether she has a premutation.
- ❖ ACOG does not currently recommend population based screening for fragile X syndrome, though some practices may choose to offer carrier screening to preconception patients based regardless of family history.