DISEASE:
Symptomatic form of fragile X syndrome in female carriers

NAME:	Symptomatic form of fragile X syndrome in female carriers
DESCRIPTION:	A rare genetic disease characterized by a variable clinical phenotype which includes similar features but is typically less severe than in affected males. Patients may present with mild to borderline intellectual disability, anxiety, social phobia, selective mutism, attention deficit hyperactivity disorder, language deficit, neurologic signs and symptoms (such as seizures, hypotonia, and clonus), ophthalmologic anomalies (strabismus, refractive errors), and facial dysmorphism (including long face, prominent forehead, large, prominent ears, and mandibular prognathism).
ORPHACODE:	449291
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	FMR1
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RELATED CONTENT

Related Genetic Tests

- Fragile X syndrome and fragile X-associated disorders (FXTAS, FXPOI)
- Fragile X syndrome/FXPOI/FXTAS - CGG repeat expansion
- Fragile X syndrome/FXPOI/FXTAS - FMR1 CGG repeat expansion
- Fragile X syndrome/POF/FXTAS - CGG repeat expansion
- Fragile X syndrome/POF/FXTAS - CGG repeat expansion
- Fragile X syndrome/POF/FXTAS - CGG repeat expansion
- Fragile X syndrome/POF/FXTAS - CGG repeat expansion
- Fragile X syndrome/POF/FXTAS - FMR1 gene CGG repeat expansion
- Premature Ovarian Failure/Primary Ovarian Insufficiency (POF/POI) (32 genes)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- fragile X messenger ribonucleoprotein 1

Related Gene Panels

- Premature Ovarian Failure/Insufficiency (32 genes) - VUB
