DISEASE:
Fragile X syndrome

NAME:	Fragile X syndrome
DESCRIPTION:	A rare genetic disease associated with mild to severe intellectual deficit that may be associated with behavioral disorders and characteristic physical features including a high forehead, prominent and large ears, hyperextensible finger joints, flat feet with pronation and, in adolescent and adult males, macroorchidism.
ORPHACODE:	908
SYNONYMS:	FRAXA syndrome FXS FraX syndrome Martin-Bell syndrome
XREF(S):	Orphanet ICD-10 MeSH OMIM OMIM MedDRA
ANALYTE(S):	FMR1
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RELATED CONTENT

Related Genetic Tests

- Autism (gene panel)
- Epilepsy, seizures (gene panel)
- FMR1-premutation instability
- 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

- Autism (57 genes) - IPG
- Epilepsy, seizures (196 genes) - IPG
- Premature Ovarian Failure/Insufficiency (32 genes) - VUB
- essai2
