Belgian Society of Human Genetics absl-vzw

Guidelines for analysis of the FMR1 gene

Indications for FMR1 analysis (diagnostic)

- Individuals of either sex with developmental delay or unexplained intellectual disability
- Women who have reproductive or fertility problems
- Individuals with the suspected diagnosis of fragile X tremor/ataxia syndrome (FXTAS)

Indications for FMR1 analysis (carrier testing)

- Individuals at risk for being carrier because of a family history of either i) fragile X syndrome or possible X-linked mental retardation; or ii) fragile X tremor/ataxia syndrome (FXTAS); or iii) premature ovarian failure
- Women entering an IVF program
- Female gamete donors

The above mentioned guidelines were prepared by the Belgium Society of Human Genetics Work Group on "Good Clinical Practice", modified by Geert Mortier, reviewed and approved on 13/07/2012 at the Meeting of the High Council for Anthropogenetics (now College for Medical Geneticists).