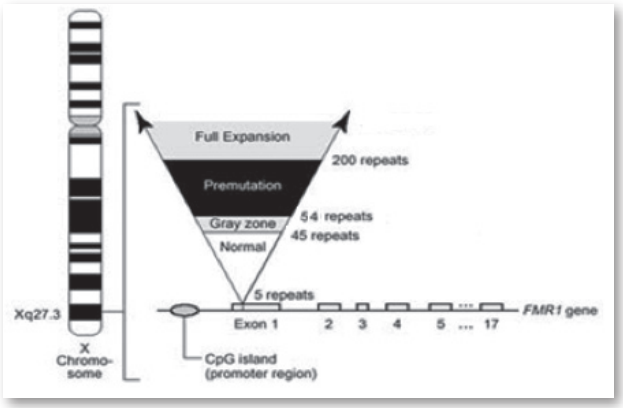
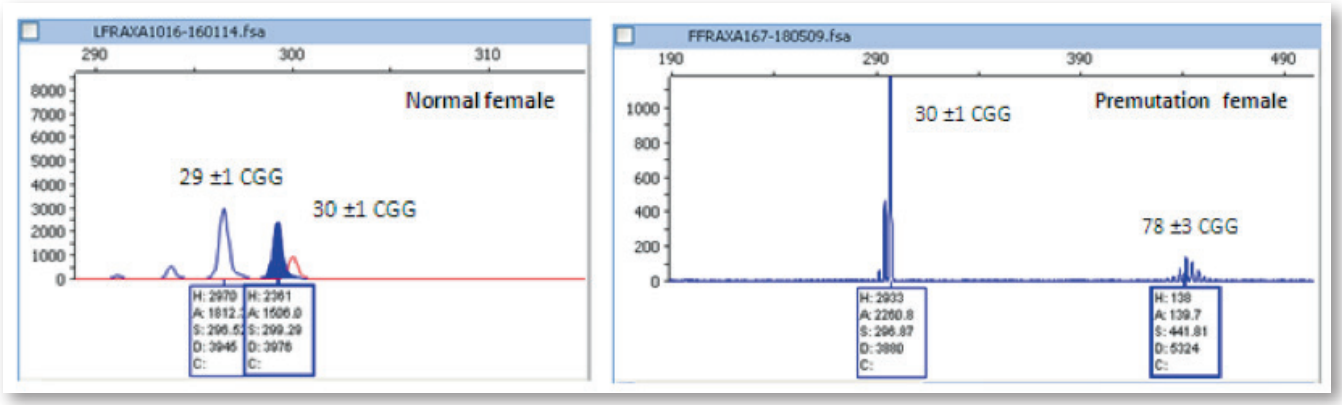
Fragile X syndrome

The Fragile X syndrome is the most common cause of inherited intellectual disability. Affected males present with mild to severe mental retardation with delay in language acquisition and/or behavioural problems being often the presenting symptoms. In addition to cognitive deficits, the FXS phenotype includes mild dysmorphic features and macroorchidism established around puberty. Behavioural disturbances including attention-deficit, hyperactivity, or autistic-like behaviour can often be observed. Approximately 50% of female carriers of the disease causing mutation will have mild to moderate mental disabilities.

It is caused by expansions of a (CGG) trinucleotide repeat in the 5'UTR of the FMR1 gene and subsequent abnormal methylation of neighboring CpG island leading to the loss of the protein product FMRP. Experteam offers complete system for the detection of FMR1 and FMR2 mutations on agarose gel as well as on a genetic sequencer (Applied Biosystems and Beckman Coulter).

Code	Name of kit	Technical specs	Amount of tests	Info	Additional info
FR.01	FraxA 1 Kit	Agarose Gel Electrophoresis kit	40	CE/IVD	Premutated Alleles
FR.02	FraxE 1 Kit	Agarose Gel Electrophoresis kit	40		Premutated Alleles
FR.01FL	FraxA 1 Kit - FL	Capillary Electrophoresis kit	40	CE/IVD	Discrimination of 1 repeat
FR.02FL	FraxE 1 Kit - FL	Capillary Electrophoresis kit	40		Discrimination of 1 repeat





Examples of capillary electrophoretic runs relative to a normal female carrying two normal FraxA alleles (29±1 and 30±1 CGG) and to a female carrying one normal allele and one premutated FraxA allele (30±1 and 78±3 CGG).