

# FMR1 gene mutations in patients with fragile X syndrome and obligate carriers: 30 years of experience in Chile

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## Resumen

Fragile X syndrome (FXS) is the most common form of inherited intellectual disability (ID) and co-morbid autism. It is caused by an amplification of the CGG repeat (>200), which is known as the full mutation, within the 5'UTR of the FMR1 gene. Expansions between 55-200 CGG repeats are termed premutation and are associated with a greater risk for fragile X-associated tremor/ataxia syndrome and fragile X-associated premature ovarian insufficiency. Intermediate alleles, also called the grey zone, include approximately 45-54 repeats and are considered borderline. Individuals with less than 45 repeats have a normal FMR1 gene. We report the occurrence of CGG expansions of the FMR1 gene in Chile among patients with ID and families with a known history of FXS. Here, we present a retrospective review conducted on 2321 cases (2202 probands and 119 relatives) referred for FXS diagnosis and cascade screening at the Institute of Nutrition and Food Technology (INTA), University of Chile. Samples were analysed using traditional cytogenetic methods and/or PCR. Southern blot was used to confirm the diagnosis. Overall frequency of FMR1 expansions observed among probands was 194 (88%), the average age of diagnosis was 88 +/- 54 years. Of 119 family members studied, 72 (60%) were diagnosed with a CGG expansion. Our results indicated that the prevalence of CGG expansions of the FMR1 gene among probands is relatively higher than other populations. The average age of diagnosis is also higher than reference values. PCR and Southern blot represent a reliable molecular technique in the diagnosis of FXS.

## Palabras clave

**KeyWords Plus:** PREMATURE OVARIAN FAILURE; CGG ALLELE SIZE; MENTAL-RETARDATION; TREMOR/ATAXIA SYNDROME; PREMUTATION ALLELES; MOLECULAR DIAGNOSIS; FULL-MUTATION; METHYLATION; MALES; MOSAICISM

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