



FXTAS DNA Test (Fragile X-Associated Tremor/Ataxia Syndrome)

Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS) is a neurological disorder that can affect older men who carry a fragile X premutation. FXTAS is characterized by progressive tremor, gait ataxia, and cognitive decline. The carrier frequency of men with a fragile X premutation is estimated to be 1 in 760 in the general population. One study suggests that up to 30% of male carriers with a fragile X premutation may develop FXTAS.

FXTAS is distinct from fragile X syndrome, the most common cause of inherited mental retardation. However, both conditions arise as a consequence of a CGG trinucleotide repeat expansion in the FMR1 gene. The number of CGG repeats varies from 6 to approximately 40 in normal alleles. Premutations have between approximately 55 and 200 CGG repeats. Full mutations have more than 200 repeats and are usually associated with abnormal FMR1 methylation, resulting in fragile X syndrome. Both males and females with a premutation are at risk for fragile X syndrome in future generations.

Males with a premutation do not typically display characteristics of fragile X syndrome and usually have a normal IQ. Those who eventually develop FXTAS are generally over 50 years of age. They often present with tremor and may have difficulty with handwriting or using eating utensils. They may exhibit unsteadiness with walking and experience frequent falling. As these symptoms gradually progress, cognitive deficits may appear including decreased memory retrieval, distractibility, and dementia.

Men with FXTAS may be misdiagnosed as having Parkinson disease, Alzheimer disease, or senile dementia. A correct diagnosis may lead to early intervention and treatment. The FXTAS DNA Test should be considered in the evaluation of adult males with intention tremor and/or late-onset ataxia. For those who are found to have a fragile X premutation, genetic counseling is recommended to address the risk for future generations to have fragile X syndrome. In families known to have fragile X syndrome, testing of grandfathers of affected children can identify those men at risk for FXTAS.

Indications for Testing

- Tremor and/or ataxia in males aged > 50 years
- Males aged > 50 years with a family history of fragile X syndrome

Special Aspects of our Service

- Rapid turnaround time
- Detailed reports with genetic interpretation, recommendations, and education
- Genetic consultation by board-certified genetic counselors and geneticists

FXTAS DNA Testing Services

- Methodology:
 - Southern blot and PCR analysis
- Results include:
 - Presence/absence of the mutation
 - CGG repeat number
- Specimen requirements:
 - 10 ml whole blood in a lavender top tube (EDTA), room temperature
- Turnaround time: 7-11 days

Please call Kimball Genetics for more information

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