NTD | Eurofins Clinical Diagnostics

Patient Information — Final Report

PATIENT NAME: [redacted]
DATE OF BIRTH: [redacted]
CROSS REFERENCE #: [redacted]
REFERRING DIAGNOSIS: [redacted]
PATIENT ID: [redacted]
LABORATORY #: [redacted]

TYPE OF SPECIMEN: [redacted]
DATE COLLECTED: [redacted]
DATE RECEIVED: [redacted]
FINAL REPORT: [redacted]

REFERRING PHYSICIAN: [redacted]
ADDRESS: [redacted]

TELEPHONE: [redacted]
FAX NUMBER: [redacted]
REFERRING PRACTICE: [redacted]

TELEPHONE: [redacted]
FAX NUMBER: [redacted]

Fragile X: CGG Repeat Analysis

Results: No mutation detected.

Interpretation

A sample from this individual was referred for molecular testing for CGG expansion within the *FMR1* gene. Abnormal full expansion of the CGG repeat tract (>200 CGG repeats) and methylation of the *FMR1* promoter causes fragile X syndrome, an X-linked intellectual disability syndrome, in males. Females with an abnormal expansion have varying degrees of developmental delay. Repeat tract expansions in the premutation range (55-200 CGG repeats) are associated with *FMR1*-associated premature ovarian insufficiency (FXPOI) in females and *FMR1*-associated tremor ataxia syndrome (FXTAS) in males.[1,2] Premutation sized CGG repeat tracts are unstable and may expand to full mutation size when passed from mother to child.

PCR analysis detected a normal 29 and 30 CGG repeat within the *FMR1* gene. This results indicates that this individual does not have the typical mutation in *FMR1*, which accounts for >99% of the cases of fragile X syndrome. This analysis will only detect CGG repeat expansions and will not detect other types of mutations in *FMR1*. These results must be interpreted in the context of this individual's clinical profile. Genetic counseling is recommended.

If fragile X syndrome is still suspected in this individual, sequence analysis of the *FMR1* gene is available. For more information on *FMR1* gene sequencing, please visit eglgenetics.com or call (470) 378-2200 to contact a laboratory genetic counselor or consult with a laboratory director.

Reference range: Normal: ≤ 44 CGG repeats, Intermediate: 45-54 CGG repeats, Premutation: 55-200 repeats, Full mutation: >200 CGG repeats

Methodology: The DNA surrounding the CGG repeat in the *FMR1* gene was amplified by PCR and the size of the repeat was determined by capillary electrophoresis.

References:

1. Sherman, S. (2000) Am J Med Genet. 97:189-194.
2. Brouwer et al. (2009) Am J Med Genet B Neuropsychiatr Genet. 150B (6):782-98.

NOTE: Direct detection of fragile X syndrome is highly accurate. Possible diagnostic errors include sample mix-ups, genotyping errors, rare genetic variants which interfere with analysis, and other sources.

Pursuant to the requirements of CLIA '88, this test was developed and its performance validated by EGL Genetic Diagnostics LLC. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes.

Medical Consultant — Laboratory Director, Molecular Genetics

This case has been reviewed and electronically signed by a Laboratory Director.