

NLGN4X-related X-linked Susceptibility to Autism Spectrum Disorder: NLGN4X Gene Deletion/Duplication

Test Code: DNLG4

Turnaround time: 2 weeks

CPT Codes: 81404 x1

Condition Description

Mutations in the *NLGN3* (Xq13) and *NLGN4X* (Xq22.33 – also referred to as *NLGN4*) genes have been found in less than 1% of individuals with autism spectrum disorder (ASD). The clinical phenotype of affected individuals varies widely and can include autism of variable severity, X-linked intellectual disability without autism, Asperger syndrome, and pervasive developmental delay not otherwise specified (PDD-NOS). Onset may be gradual or abrupt and regression of milestones may occur. There are usually no dysmorphic features present. Carrier females are often unaffected, but neuropsychiatric disorders have been reported in a female carrying a deletion of exons 4-6 of *NLGN4X*.

For patients with suspected *NLGN4X*-Related X-Linked Susceptibility to Autism Spectrum Disorder, sequence analysis is recommended as the first step in mutation identification. For patients in whom mutations are not identified by full gene sequencing, deletion/duplication analysis is appropriate.

This testing is for mutation in the *NLGN4X* gene only.

References:

- Luitas & Persioco. (2009) J Med Genet 46:1-8.
- GeneReviews
- [OMIM #300427](#): *NLGN4X* gene
- [OMIM #300495](#): X-Linked Autism Disorder
- [OMIM #300497](#): X-Linked Asperger Syndrome

Genes

[NLGN4X](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of *NLGN4X*-Related X-Linked Susceptibility to Autism Spectrum Disorder in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of *NLGN4X*-Related X-Linked Susceptibility to Autism Spectrum Disorder in whom sequence analysis was negative.

Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

Detection

Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood (EDTA)

Specimen Requirements:

EDTA (Purple Top)

Infants and Young Children (2 years of age to 10 years old): 3-5 ml

Older Children & Adults: 5-10 ml

Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

Type: DNA, Isolated

Specimen Requirements:

Microtainer

3µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

Refrigerate until time of shipment in 100 ng/μL in TE buffer. Ship sample at room temperature with overnight delivery.

Related Tests

- Sequence analysis of the *NLGN4X* gene is available and is required before deletion/duplication analysis.
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Prenatal testing is available only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.
- X-Linked Intellectual Disability panels are available for 30, 60, and 90 genes.
- Sequencing and deletion/duplication analysis are also available for the *NLGN3* gene.