

# The University of Chicago Genetic Services Laboratories



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CLIA #: 14D0917593 CAP #: 18827-49

## SNRPN Duplication Analysis for Autism

### Clinical Features:

The phenotype associated with proximal 15q duplications includes autism, mental retardation/developmental delay, ataxia, hypotonia, and seizures. However, there are individuals with this duplication who only have an autism-spectrum disorder [2]. Approximately 1% of autistic individuals have this cytogenetic alteration [1].

### Inheritance:

To date, all proximal 15q duplications in individuals with autism have been maternally derived/inherited. Identification of such a duplication has important implications for recurrence risk assessment.

### Molecular Genetics:

Autism affects approximately 1 in 500 individuals. However, despite the high incidence, the causes of autism are poorly understood. To date, several single-gene conditions and several cytogenetic alterations have been associated with autism. However, in the majority of individuals, autism does not have a recognized etiology. Recent studies have indicated that changes in the proximal region of chromosome 15q, including duplications, may be the most common identifiable cause of autism [1]. Dicentric additional marker chromosomes containing 15q material are easily recognized by routine chromosome analysis. However, many interstitial, proximal 15q duplications would likely be missed by this method. We offer FISH analysis using a SNRPN probe for duplications of the proximal 15q region. Sample submission paperwork and instructions are included in this memo.

### Additional Resources:

#### Autism Speaks

2 Park Avenue  
11th Floor  
New York, NY 10016  
Phone: (212) 252-8584  
Fax: (212) 252-8676  
E-mail: [contactus@autismspeaks.org](mailto:contactus@autismspeaks.org)  
[www.autismspeaks.org](http://www.autismspeaks.org)

#### Autism Society of America

7910 Woodmont Avenue, Suite 300  
Bethesda, Maryland 20814-3067  
Phone: 301-657-0881 or  
1-800-3AUTISM (1-800-328-8476)  
[www.autism-society.org](http://www.autism-society.org)

### Test methods:

Sample specifications:	5 cc of blood in a green top (sodium heparin) tube
Cost:	\$325
CPT codes:	88230, 88271, 88291, 88273
Turn-around time:	12 days

## Results

You will be informed of the results of your case as soon as it has been completed. Results, along with an interpretive report, will be faxed and mailed to the referring physician. Additional reports will be provided as requested. All abnormal results will be reported by telephone.

## Laboratory Faculty and Staff:

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## References:

1. Schroer et al., Am J Med Genet 76:327-336, 1998.
2. Cook et al., Am J Hum Genet 60:928-934, 1997.

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