



Division of Human Genetics

## JAG1 Sequence Analysis for Alagille Syndrome

### Molecular Genetics Laboratory

CLIA#: 36D0965791

Phone: (513) 636-4474

Fax: (513) 636-4373

Email: [moleculargenetics@cchmc.org](mailto:moleculargenetics@cchmc.org)

Additional information and test requisitions are available at:

[www.cincinnatichildrens.org/molecular-genetics](http://www.cincinnatichildrens.org/molecular-genetics)



*Helping you fit the pieces together*

### Shipping Instructions

Please enclose test requisition with sample. All information must be completed before sample can be processed.

Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday

Ship to:

Cytogenetics and Molecular Genetics Laboratories

3333 Burnet Avenue NRB 1042

Cincinnati, OH 45229

513-636-4474

Alagille syndrome is a complex disorder characterized by cholestasis and bile duct paucity, cardiac defects, skeletal abnormalities, ophthalmologic abnormalities and characteristic facial features. Less common features of Alagille syndrome include kidney abnormalities, neurovascular accidents and pancreatic insufficiency. Developmental delay and mild mental retardation are seen in a minority of individuals with Alagille syndrome. Mutations in *JAG1* result in broad phenotypic variability, even within families.

### INDICATIONS:

- Diagnostic testing in patients with suspected Alagille syndrome
- Targeted mutation analysis of at-risk relatives of patient with *JAG1* mutation
- Prenatal diagnosis of an at-risk fetus



## SPECIMEN:

At least 3 mls whole blood in purple top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Phlebotomist must initial tube to verify patient's identity.

## METHODOLOGY:

PCR-based sequencing of the coding regions and their exon/intron boundaries of the *JAG1* gene.

## CLINICAL SENSITIVITY:

PCR-based sequencing of the coding regions and their exon/intron boundaries of the *JAG1* gene detects ~ 88% patients with Alagille syndrome. 20p12 microdeletion syndrome, which includes deletion of the entire *JAG1* gene, is identified in approximately 7% of patients with Alagille syndrome and can be detected by fluorescence in-situ hybridization (FISH) analysis. Mutations in *NOTCH2* are associated with Alagille syndrome in approximately 1% of affected individuals.

## ANALYTICAL SENSITIVITY:

The sensitivity of PCR-based DNA sequencing is over 99% for the detection of nucleotide base changes, small deletions and insertions in the regions analyzed. Mutations in regulatory regions or other untranslated regions are not detected by this test. Multiple exon deletions, large insertions, genetic recombinational events and rare, primer site mutations may not be identified using this methodology. If the patient has received a liver transplant or recent blood transfusion, donor DNA may be present in the blood along with patient DNA (chimerism). In this case, additional testing may be required to rule out chimerism.

## TURN-AROUND TIME:

3-4 weeks

## COST:

(current 7/1/10-6/30/11)

Please call 1-866-450-4198 for current pricing or with any billing questions.

## CPT CODES:

- *JAG1* sequence analysis: 83890, 83898(x3), 83894(x3), 83891(x2), 83904(x49), 83909(x49), 83912
- Family specific analysis: 83890, 83898, 83894, 83891, 83904, 83912

## RESULTS:

Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.