Lab Dept: Anatomic Pathology

Test Name: ALAGILLE WATSON SYNDROME (JAG1) KNOWN MUTATION

General Information

Lab Order Codes: JAGK (Blood), JAGKB (Saliva)

Synonyms: Cholestasis with Perpheral Pulmonary Stenosis; Arteriohepatic Dysplasia; Syndromatic Hepatic Ductular Hypoplasia; JAG1

CPT Codes: 81403 – Molecular pathology procedure, Level 4

Test Includes: JAG1 specific mutation testing in situations such as prenatal diagnosis, carrier testing and presymptomatic testing on family members at risk for the mutation. There must be a confirmed mutation analysis on an individual to proceed with this testing.
Note: Saliva specimens cannot be accepted for HDT array tests.

Logistics

Test Indications: Once a mutation is confirmed in an affected individual, prenatal diagnosis, carrier testing, and presymptomatic testing can be performed on family members who are at risk for the mutation. In some cases, particularly those with time constraints such as prenatal diagnosis, mutation confirmation in an affected family member can run concurrently with other samples from the same family.

Reasons for referral:
1. Clinical laboratory confirmation of clinical diagnosis.
2. Development of appropriate evaluation and management plan.
3. Evaluation of family members as possible donors for liver transplantation.
4. Identification of mutation carriers with milder manifestations.
5. Genetic counseling

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: CTGT – One Known Mutation

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours (Preferred draws are Sunday - Thursday as specimens can only be received at the reference lab Monday - Friday. Specimens collected Friday or Saturday will be held for shipment on Monday.)

Turnaround Time:	2 – 3 weeks
Special Instructions:	A CTGT signed request form must be sent with any patient or specimen to the laboratory. DNA can be prepared using saliva. Only samples collected in Oragene DNA Self-Collection Kit or Oragene Saliva Collection Kit for Young Children are accepted. Kits are available through: DNA Genotek (www.dnagenotek.com) or by phone (1-866-813-6354).

Specimen

Specimen Type:	Whole blood Saliva Note: Saliva specimens cannot be accepted for HDT array testing.
Container:	Blood: Lavender top (EDTA) tube Saliva collected using an Oragene Saliva Collection kit.
Draw Volume:	3 - 5 mL blood Saliva: follow kit instructions
Processed Volume:	Same as Draw Volume
Collection:	Blood: Routine venipuncture Saliva: Follow kit instructions
Special Processing:	Lab Staff: Send whole blood in original collection container or saliva collection kit, including signed consent form and requisition, with a cool pack during warm temperatures, via overnight or second-day courier so that the sample will arrive at CTGT on a weekday (Monday through Friday). Samples drawn on Friday or Saturday should be held at refrigerated temperatures for shipment on Sunday. Samples can be held at refrigerated temperatures for up to 7 days. **Do not** freeze.
Patient Preparation:	Blood: None Saliva: Follow kit instructions
Sample Rejection:	Unrefrigerated specimens older than 48 hours for blood; frozen specimens; mislabeled or unlabeled specimens

Interpretive

Reference Range:	No mutations detected
Critical Values:	N/A
Limitations:	N/A

Methodology:	Sanger Sequencing: Direct DNA sequencing of PCR products generated from genomic DNA
References:	CTGT November 2012 (484) 244-2900 Fax (484) 244-2904
Updates:	1/12/2011: CPT Updates 11/6/2012: Moved from GeneDx to CTGT.