
Lab Dept: Anatomic Pathology

Test Name: CONGENITAL HYPERINSULINISM EVALUATION SEQUENCING

General Information

Lab Order Codes: CHIE

Synonyms: ABCC8 Sequencing; CGK Sequencing; GLUD1 Sequencing; KCNJ11 Sequencing

CPT Codes: 81407 – Molecular Pathology procedure, Level 8
81406 x2 – Molecular Pathology procedure, Level 7
81403 – Molecular Pathology procedure, Level 4

Test Includes: ABCC8 (CH) DNA Sequencing, GCK (CH) DNA Sequencing, GLUD1 (CH) DNA Sequencing, KCNJ11 (CH) DNA Sequencing. Detects mutations including point mutations, deletions, insertions, and rearrangements.

Logistics

Test Indications: Severe, persistent hypoglycemia in newborns or infants Family history of Congenital Hyperinsulinism..

Lab Testing Sections: Anatomic Pathology – Sendouts

Referred to: Athena Diagnostics (Athena Test: 819)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 5 - 7 days

Special Instructions: Blood samples should be submitted for proband & both parents whenever possible.

Please note if the patient is symptomatic or asymptomatic. If the patient is asymptomatic, complete and submit "[Informed Consent](#)" with the specimen.

Specimen

Specimen Type: Whole blood

Container:	Lavender top (EDTA) tube Alternate tube: Yellow-top (ACD) tube
Draw Volume:	Adult: 8 mL (Minimum: 6 mL) blood Pediatric (0-3 yrs): 2 mL (Minimum: 1 mL) blood Note: Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients 0-3 yrs have higher WBC, yielding more DNA per mL of blood.
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
Special Processing:	Lab Staff: Do Not Centrifuge. Specimen should remain in the original collection container. Store and ship at room temperature Monday – Thursday. Specimens collected Friday, Saturday or Sunday may be kept at room temp or refrigerated and shipped on Monday. Forward promptly. Specimens are stable for 10 days at room temp or refrigerated.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	Interpretive report
Critical Values:	N/A
Limitations:	N/A
Methodology:	Sanger sequencing
References:	Athena Diagnostics Laboratory March 2018 (800) 394-4493 Fax: (508) 753-5601
Updates:	2/4/2013: CPT update 10/9/2013: CPT update