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**Lab Dept:** Anatomic Pathology

**Test Name:** CONGENITAL HYPERINSULINISM EVALUATION SEQUENCING

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***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.

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***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:** Monday – Thursday by 15:00 only

**Turnaround Time:** 7 days

**Special Instructions:** Blood samples should be submitted for proband & both parents whenever possible. Collect specimens Monday – Thursday by 15:00 ONLY.

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

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***Specimen***

**Specimen Type:** Whole blood

<b>Container:</b>	Lavender top (EDTA) tube Alternate tube: Yellow-top (ACD) tube
<b>Draw Volume:</b>	10 mL (Minimum: 2 mL) blood
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine venipuncture.
<b>Special Processing:</b>	Lab Staff: <b>Do Not</b> Centrifuge. Specimen should remain in the original collection container. Store and ship at room temperature on MONDAY – THURSDAY. Specimens may be refrigerated overnight. Forward promptly. Specimens are stable for 48 hours.
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Mislabeled or unlabeled specimens

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### ***Interpretive***

<b>Reference Range:</b>	Interpretive report
<b>Critical Values:</b>	N/A
<b>Limitations:</b>	N/A
<b>Methodology:</b>	Polymerase Chain Reaction (PCR), DNA sequencing of entire protein coding regions of genes.
<b>References:</b>	<a href="#">Athena Diagnostics Laboratory</a> October 2013 (800) 394-4493 Fax: (508) 753-5601
<b>Updates:</b>	2/4/2013: CPT update 10/9/2013: CPT update