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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:**       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.

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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>	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.
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine blood collection
<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 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.
<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>	Sanger sequencing
<b>References:</b>	<a href="#">Athena Diagnostics Laboratory</a> March 2018 (800) 394-4493 Fax: (508) 753-5601
<b>Updates:</b>	2/4/2013: CPT update 10/9/2013: CPT update