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*Helping to identify
the best treatment
for newborns with
severe, persistent
hypoglycemia.*

Congenital Hyperinsulinism Evaluation



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 **Correlagen**[®]
Correlagen, Inc.

 **athena diagnostics**[®]
Testing You Can Count On.



Causes of Congenital Hyperinsulinism (CH)

(also known as PHHI, FHI, or nesidioblastosis)

- More than half of all CH is associated with mutation in any one of five autosomally encoded genes (*ABCC8*, *KCNJ11*, *GLUD1*, *GCK*, *HADHSC*).¹
- The most severe form of CH is typically caused by mutations in *ABCC8* (also known as *SUR1*) or *KCNJ11* (also known as *KIR6.2*).²
- Mutations in *ABCC8* or *KCNJ11* can cause both the diffuse and the focal form of CH.³
- Mode of inheritance varies between dominant (*GLUD1*, *GCK*, *ABCC8*), recessive (*KCNJ11*, *ABCC8*), and dominant after somatic loss-of-heterozygosity (*KCNJ11*, *ABCC8*).

Why genetic testing?

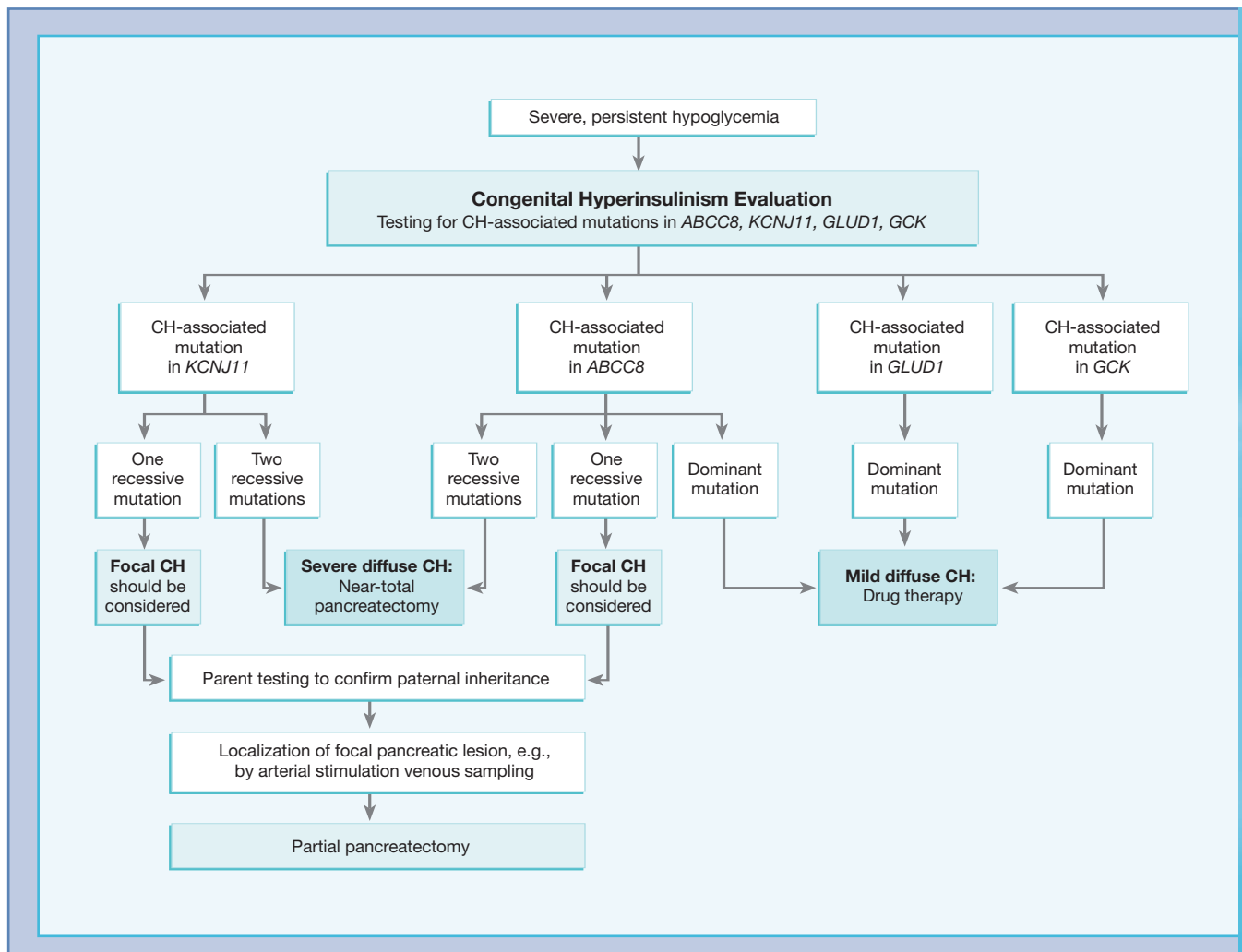
Genetic testing for mutations in *ABCC8*, *KCNJ11*, *GLUD1*, and *GCK*

- Can identify most cases (85%) of CH that require pancreatectomy.¹
- Distinguishes between *ABCC8*- or *KCNJ11*-related CH, which typically requires pancreatectomy, and *GLUD1*- or *GCK*-related CH, which typically responds well to drug therapy (see Figure 1).
- Can help to differentiate between the diffuse and the focal form of *ABCC8*- or *KCNJ11*-associated CH,⁴ so that infants with focal CH can be treated with a partial pancreatectomy rather than a near-total pancreatectomy (see Figure 1).
- **Please Note:** Demonstrating paternal inheritance of a CH-associated mutation can strengthen a diagnosis of focal CH (see Figure 1). For that reason, blood samples should be submitted for the proband and both parents whenever possible.

Indications for testing:

- Severe, persistent hypoglycemia in newborns or infants
- Family history of CH

*Figure 1:
An approach to CH testing and related therapeutic decision-making*



*For complete ordering information,
please see the reverse side.*

References: 1. Dunne MJ, et al (2004) *Physiol Rev* 84:239-75. 2. Stanley CA, et al. (2002) *J Clin Endocrinol Metab* 87:4857-9. 3. Fournet JC, et al. (2001) *Am J Pathol* 158:2177-84. 4. Suchi M, et al. (2003) *Pediatr Dev Pathol* 6:322-33.

For a brief review on CH, please visit www.athenadiagnostics.com/DR

Congenital Hyperinsulinism Evaluation

Typical Presentation:	Severe, persistent hypoglycemia in newborns
Synonyms:	Persistent Hyperinsulinemic Hypoglycemia of Infancy (PHHI), Familial Hyperinsulinism (FHI), Nesidioblastosis, Focal Adenomatosis
Indications for Testing:	<ul style="list-style-type: none">• Severe, persistent hypoglycemia in newborns or infants• Family history of CH

TEST DETAILS

Test Code:	819
Test Includes:	ABCC8 (CH) DNA Sequencing Test, #827 KCNJ11 (CH) DNA Sequencing Test, #826 GLUD1 (CH) DNA Sequencing Test, #822 GCK (CH) DNA Sequencing Test, #823
Test Turnaround:	7-14 days

TECHNICAL INFORMATION

Methodology:	Polymerase Chain Reaction (PCR), DNA sequencing of entire protein coding regions of genes
Patents:	United States Patent No. 6,054,313

SHIPPING CONSIDERATIONS

Specimen Type:	Whole blood – blood samples should be submitted for proband and both parents, whenever possible
Volume:	10 mL (pediatric minimum: 2 mL)
Collection Tube:	Yellow or lavender top tubes
Stability:	Hemolysis may compromise DNA recovery and integrity after 48 hrs
Storage Conditions:	For short periods (until shipped) at 4°C
Shipping Conditions:	Overnight at room temperature (specimen arrival must be less than 24 hrs after collection); ship Monday through Thursday only

Call Athena Diagnostics' Customer Service
Representatives to order the Congenital
Hyperinsulinism Evaluation (#819) at:

800-394-4493 x2



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