

Select the best therapy for newborns and infants with insulin-sensitive hyperglycemia.

Neonatal Diabetes Mellitus Evaluation

What is Neonatal Diabetes Mellitus?

- Neonatal diabetes mellitus (NDM) is characterized by onset of transient (TNDM) or permanent (PNDM) insulin-sensitive hyperglycemia within the first three to six months of life.¹
- Autosomal dominant gain-of-function mutations in the gene *KCNJ11* (also known as *KIR6.2*) account for about 50% of all cases of PNDM.^{2,3,4} *KCNJ11*-related PNDM may be responsive to sulfonylurea therapy.⁵
- Autosomal recessive loss-of-function mutations in the gene *GCK* lead to insulin-dependent PNDM.⁶
- Autosomal recessive loss-of-function mutations in the gene *IPF1* cause pancreatic agenesis.⁷

(Of note, heterozygosity for loss-of-function mutations in *GCK* or *IPF1* is associated with MODY.^{8,9})

Why genetic testing?

By identifying the exact genetic cause of PNDM, the Neonatal Diabetes Mellitus Evaluation can

- Help the physician to select the most appropriate treatment:
 - *KCNJ11*-related PNDM may be treatable with oral hypoglycemics.^{2,5}
 - *GCK*-related PNDM requires insulin replacement therapy.
 - *IPF1*-related PNDM requires replacement of both endocrine and exocrine pancreatic functions.
- Alert relatives of patients with PNDM to their increased risk for PNDM and/or MODY.

Indications for testing:

- Insulin-sensitive hyperglycemia in newborns or infants
- Family history of neonatal diabetes mellitus

For complete ordering information, please see the reverse side.

References: 1. Shield JP (2000) *Horm Res* 53 (Suppl 1):7-11. 2. Gloyn AL et al (2004) *N Engl J Med* 350:1838-49. 3. Vaxillaire M et al (2004) *Diabetes* 53:2719-2722. 4. Massa O et al (2005) *Hum Mutat* 25:22-7. 5. Sagen JV et al (2004) *Diabetes* 53:2713-2718. 6. Njolstad PR et al (2001) *N Engl J Med* 344:1588-92. 7. Stoffers DA et al (1997) *Nat Genet* 15:106-10. 8. Froguel P et al (1993) *N Engl J Med* 328:697-702. 9. Stoffers DA et al (1997) *Nat Genet* 17:138-9.

For a brief review on NDM, please visit www.athendiagnosics.com/DR



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Neonatal Diabetes Mellitus Evaluation

- Typical Presentation:** Insulin-sensitive hyperglycemia in newborns
- Indication for Testing:**
- Insulin-sensitive hyperglycemia in newborns or infants
 - Family history of neonatal diabetes mellitus

TEST DETAILS

- Test Code:** 844
- Test Includes:** *IPF1* (NDM) DNA Sequencing Test #841
GCK (NDM) DNA Sequencing Test #842
KCNJ11 (NDM) DNA Sequencing Test #843
- 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. 6,274,310

SHIPPING CONSIDERATIONS

- Specimen Type:** Whole blood
- Volume:** 10 mL (pediatric minimum: 2 mL)
- Collection Tube:** Yellow or lavender top
- 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 Neonatal Diabetes Mellitus Evaluation (Test #844) at:

800-394-4493 x2



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