REQUEST FOR BECKWITH-WIEDEMANN SYNDROME (BWS) TESTING

Please provide the following information. We cannot perform your test without ALL of this information. PLEASE PRINT ALL ANSWERS.

**TEST REQUESTED**

- Methylation and high resolution copy number analysis of 11p15.5 (will also provide evidence for UPD11p15); if negative, reflex to \textit{CDKN1C} sequence analysis
- Methylation analysis of 11p15.5 (will also provide evidence for UPD11p15); if negative, reflex to \textit{CDKN1C} sequence analysis
- Methylation analysis of 11p15.5 (will also provide evidence for UPD11p15) \textbf{only}
- 11p15.5 high resolution copy number analysis \textbf{only (aCGH)}
- Sequence analysis of \textit{CDKN1C} gene \textbf{only}
- Site specific analysis (familial) \underline{m}ethylation defect* \underline{c}opy number defect* \underline{C}DKN1C *
- Prenatal methylation of 11p15.5 (will also provide evidence for UPD11p15) \textbf{with} automatic reflex to \textit{CDKN1C} if negative**
- Prenatal methylation analysis of 11p15.5 (analysis will also provide evidence for UPD11p15) \textbf{only}**
- Prenatal \textit{CDKN1C} sequencing analysis \textbf{only}**
- Prenatal site specific analysis (familial)** \underline{m}ethylation defect* \underline{c}opy number defect* \underline{C}DKN1C *

*Required information
**Please call the laboratory prior to sending a prenatal sample. Please refer to the test catalog for prenatal sample requirements on www.med.upenn.edu/genetics/gdl

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**CLINICAL INFORMATION**

- ICD-10 CODE(S):* 
  - Q89.8 Hemi hypertrophy
  - Q38.2 Macroglossia
  - C64.9 Wilms tumor
  - P70.4 Neonatal hypoglycemia
  - Q79.59 Omphalocele
  - P08.1 Large for gestational age
  - Z84.81 Family history of carrier of genetic disease

- ADDITIONAL CLINICAL SYMPTOMS:

If the test request is for site specific FAMILIAL ANALYSIS for a KNOWN MUTATION:

Name of person previously tested and relationship:

Was the previous testing performed at the Genetic Diagnostic Laboratory?  Yes  No

Result (Please include a copy of the result):*

**PATIENT INFORMATION**

Sample Collection Date & Time: __________________________

FIRST NAME  MI  LAST NAME  BIRTH DATE (MM/DD/YYYY)  GENDER

- ANCESTRY
  - Western/Northern European
  - Central/Eastern European
  - Latin American/Caribbean
  - African
  - Asian
  - Jewish (Ashkenazi)
  - American Indian
  - Near East/Middle Eastern
  - Native Hawaiian or Pacific Islander
  - Specify countries: ______________________________________________________________
  - Other: ______________________

**GENETIC DIAGNOSTIC LABORATORY**

UNIVERSITY OF PENNSYLVANIA SCHOOL OF MEDICINE
DEPARTMENT OF GENETICS

415 Anatomy Chemistry Building • 3620 Hamilton Walk • Philadelphia, PA 19104
Tel: (215) 573-9161 • Fax: (215) 573-5940 • Email: gdl@pennmedicine.upenn.edu

CLIA ID: 39D0893887
**PATIENT INFORMATION**

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**PHYSICIAN INFORMATION***

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**PAYMENT OPTIONS*** (must choose one) [a receipt will be mailed to the patient for self-pay options]

- [ ] I have enclosed a check payable to the “Genetic Diagnostic Laboratory” for $ ____________________

- [ ] Please charge my credit card for the amount of $ ____________________
  - [ ] VISA
  - [ ] Master Card
  - [ ] Discover
  - [ ] American Express
  - Card Number: ____________________ Exp date: ____________
  - Name of cardholder as it appears on card: ____________________

- [ ] I have Pennsylvania Medicaid. A copy of my Medicaid card is attached.

- [ ] INSTITUTIONAL BILLING: The Institution where my testing originated has agreed to pay all charges for the testing.
  - INCLUDE Billing Address, Person Authorizing Payment, Telephone, and Fax below:

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INFORMED CONSENT: GENETIC TESTING FOR BECKWITH- WIEDEMANN SYNDROME (BWS)

Background: Beckwith-Wiedemann syndrome (BWS) is characterized by overgrowth, specific physical findings, and an increased risk to develop tumors in childhood. Early diagnosis of BWS allows for increased screening for Wilms tumor, hepatoblastoma and other associated benign and malignant tumors.

The molecular basis of BWS is complex and involves changes in several genes on chromosome 11p15.5. Methylation analysis is used to detect specific differences in patients with BWS. If methylation analysis is negative, then sequence analysis of CDKN1C can be performed. Testing will not identify all molecular changes associated with BWS. A negative test does not change a clinical diagnosis of BWS.

Purpose: I, or my child/fetus, will be tested for alterations in several genes on chromosome 11p15.5 as described above. The purpose of this genetic testing is to determine whether I, or my child/fetus, have specific molecular changes associated with BWS.

Results: I understand that there are three possible results to this testing:

POSITIVE: A clinically significant alteration is detected in the 11p15.5 methylation pattern, copy number, or the CDKN1C gene sequence. I understand a positive molecular result is highly associated with BWS, which can confirm a clinical diagnosis.

NEGATIVE: The analysis did not detect a molecular alteration associated with BWS. This result reduces the likelihood that I/my child/fetus has a (epi)mutation related to BWS. Methods currently in use are unable to detect all mutations and I, or my child/fetus, may still have a molecular alteration that was not detected by the current technology.

VARIANT OF UNCERTAIN SIGNIFICANCE: The laboratory could detect an alteration of currently unknown significance, called a “variant of unknown significance (VUS)”. Our laboratory will work with my physician to help determine if the VUS can be further classified as to whether it is associated with BWS.

Disclosure Policy: The Genetic Diagnostic Laboratory will release my test results to the ordering healthcare provider or genetic counselor, and otherwise only as permitted by law. The results will be kept confidential to the extent allowed by law. If I provide separate written consent, the lab will release my test results to other medical professionals or third persons I want to receive my results.

Limitations: While genetic testing is highly accurate for detection of the majority of disease causing mutations, a small fraction of mutations may be missed by the current technology. Due to the nature of the testing, there is a small possibility that the test will not work properly or that an error will occur. Occasionally, testing may reveal a variant of unknown significance that is unable to be definitively interpreted as positive or negative for disease-association based on the current knowledge of the variant. The DNA analysis performed at the University of Pennsylvania Genetic Diagnostic Laboratory is specific only for the gene(s) analyzed and in no way guarantees my health.

There are federal laws in place that prohibit health insurers and employers from discriminating based on genetic information, such as test results. There currently are no federal laws prohibiting discrimination based on genetic information by life insurance, long term care, or disability insurance companies, but state laws may restrict this. I understand I can ask my ordering provider or genetic counselor for more information about how insurers might use genetic information.

Initials _______  
9/15/17
Use of Specimens After Clinical Test Performed: I understand my blood or tissue specimen will not be returned to me or the ordering healthcare provider, and becomes the property of the lab upon receipt. The laboratory is not a DNA banking facility; therefore this is no guarantee that samples will be available or usable for additional or future testing. Samples from New York residents will be disposed of 60 days after clinical testing is complete.

After the laboratory completes the ordered clinical test, the lab may retain and preserve the specimen to validate the development of future genetic tests or for future research or education purposes. The laboratory is committed to continuous improvement and therefore I understand my coded sample may be used to validate a new assay. If testing reveals a clinically significant result during the validation process of a new assay related to the original indication for testing, my health provider may be contacted. If the lab uses the specimen for future research or education purposes, the specimen will be de-identified by removing my personally identifying information. My name, address and other personal identifying information will not be linked to the samples, or the results of the research, and I will not be identified in any research results or publications. I will not receive a copy of the research results. I can decline for my sample to be retained at the lab by filling out “Research Opt Out” form found on the following website: http://www.med.upenn.edu/genetics/gdl/.

I understand the lab may wish to contact me, or my ordering healthcare provider, for additional information. The additional information may include, but would not be limited to, information about health and family history that might be relevant to the research. I understand I can decline future contact from the lab by filling out “Research Opt Out” form found on the following website: http://www.med.upenn.edu/genetics/gdl/.

Genetic Counseling provided by a qualified specialist (i.e. genetic counselor/ medical geneticist) is a recommendation for individuals proceeding with genetic testing. This service is available before and after genetic testing. Additionally, other testing or further physician consults may be warranted.

The Genetic Diagnostic Laboratory is also an available resource to ask more questions about this testing. The laboratory genetic counselor can be reached at 215-573-9161 and Arupa Ganguly, PhD, FACMG can be reached at 215-898-3122. I will be given a copy of this consent form to keep.

HEALTHCARE PROVIDER STATEMENT:

I have explained to _________________________________ the purpose of this genetic testing, the procedures required and the possible risks and benefits to the best of my ability.

_______________________________________________________
Printed Name of Professional Obtaining Consent

_______________________________________________________    ________________
Signature of Professional Obtaining Consent    Date

CONSENT OF PATIENT:

I have read and received a copy of this consent form. I agree to have genetic testing performed for myself, child or my fetus, and accept the risks. I understand the information provided in this document and I have had the opportunity to ask questions I have about the testing, the procedure, the associated risks and the alternatives.

Patient’s Printed Name: ____________________________________    DOB: ________________

Patient’s Signature: ____________________________________    Date: ________________
(or Parent/Guardian if patient is a minor)

Name and Relationship: ____________________________________
(Parent/Guardian if patient is a minor)