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) with automatic reflex to CDKN1C if negative

☐ Methylation analysis of 11p15.5 (analysis will also provide evidence for UPD11p15) with automatic reflex to CDKN1C if negative

☐ Methylation analysis of 11p15.5 (analysis will also provide evidence for UPD11p15) only

☐ 11p15.5 high resolution copy number analysis only (aCGH)

☐ CDKN1C sequencing analysis only

☐ Screening for a KNOWN FAMILIAL _______ methylation defect* _______ copy number defect* _______ CDKN1C mutation*

☐ Prenatal methylation and high resolution copy number analysis of 11p15.5 (will also provide evidence for UPD11p15) with automatic reflex to CDKN1C if negative**

☐ Methylation analysis of 11p15.5 (analysis will also provide evidence for UPD11p15) with automatic reflex to CDKN1C if negative only**

☐ Prenatal CDKN1C sequencing analysis only**

☐ Prenatal screening for a KNOWN FAMILIAL _______ methylation defect* _______ copy number defect* _______ CDKN1C mutation*

*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

CLINICAL INFORMATION

ICD-10 CODE(S):* ☐ Q89.8 Hemi hypertrophy ☐ P70.4 Neonatal hypoglycemia ☐ Z84.81 Family history of carrier of genetic disease

☐ Q38.2 Macroglossia ☐ Q79.59 Omphalocele ☐ Other:

☐ C64.9 Wilms tumor ☐ P08.1 Large for gestational age

ADDITIONAL CLINICAL SYMPTOMS:______________________________________________________________

If the test request is for FAMILIAL or PRENATAL 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 REGISTRATION FORM

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

PATIENT INFORMATION

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

STREET ADDRESS

CITY                                                                 STATE                ZIP

PHYSICIAN INFORMATION*

REFERRING PHYSICIAN            PHONE            FAX

GENETIC COUNSELOR            PHONE            FAX

EMAIL ADDRESS FOR COUNSELOR            EMAIL ADDRESS FOR PHYSICIAN

INSTITUTION AND DEPARTMENT

STREET ADDRESS            CITY                                                                 STATE                ZIP

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:

BILLING ADDRESS

BILLING ADDRESS

NAME OF INDIVIDUAL AUTHORIZING PAYMENT            PHONE            FAX
VERIFICATION OF CORRECTLY IDENTIFIED BLOOD TUBES

I am a participant in genetic DNA testing.

I have been shown the tubes containing my blood for this genetic testing and my name has been correctly placed on each one of these tubes.

I have signed a copy of the consent form regarding this genetic testing to be sent along with my blood samples. I have been given a copy of the consent form to keep.

Participant Name: __________________________________________

Participant/Parent Signature: _________________________________

Date: ________________

10/01/2016
INFORMED CONSENT: GENETIC TESTING FOR BECKWITH- WIEDEMANN SYNDROME (BWS)

Background: Beckwith-Wiedemann syndrome (BWS) is a genetic disorder characterized by overgrowth, specific physical findings, and an increased risk to develop tumors. 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. I understand that the testing will take approximately 4-6 weeks to complete. 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 mutation is detected in the 11p15.5 methylation pattern or the CDKN1C gene sequence. I understand that this means that there is a high risk to developing clinical symptoms of BWS. This result will also confirm a clinical diagnosis of BWS.

**NEGATIVE:** The analysis did not detect a molecular alteration associated with BWS. This result reduces the likelihood that you/your child/fetus has a mutation causing 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:** The laboratory could detect an alteration of currently unknown significance, called a “variant of unknown significance (VUS)”. Our laboratory will work with your physician to help determine if the VUS can be further classified as to whether it is disease-associated for BWS.

Disclosure Policy: Results will be reported to me only through the physician or genetic counselor who requested the testing due to the complexity of DNA based testing. The results of genetic testing are protected by the Health Insurance Portability and Accountability Act (HIPAA) of 1996 (Public Law 104-191). Release of test results is limited to authorized personnel as required by law. Additionally, results can be released to other medical professionals or other parties with my written consent.

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 our current knowledge of the variant. My signature below acknowledges my voluntary participation in this test, but in no way releases the laboratory and staff from their professional and ethical responsibility to me. Furthermore, the DNA analysis performed at the University of Pennsylvania Genetic Diagnostic Laboratory is specific for Beckwith-Wiedemann syndrome 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 (i.e. Genetic Information Nondiscrimination Act (GINA) of 2008 (Public Law 110-233). There are currently no laws specific to discrimination based on genetic information for life insurance, long term care, or disability insurance companies. Additional information about GINA can be found on the Genetics Public & Policy Center’s website at www.dnapolicy.org.
**Use of Specimens:** Any blood or tissue specimens obtained for the purposes of this genetic testing become the exclusive property of the Genetic Diagnostic Laboratory. After the specific tests requested have been completed and reported, the laboratory may dispose of, retain, or preserve these specimens for research or for validation in the development of future genetic tests. In all circumstances described previously, my identity will be protected and research results will not be provided to me or to any other party. If use of this genetic material results in a scientific publication, it will not contain any identifying information. Indicate consent or denial to the above by initialing below. Refusal to consent to research will not affect the reporting of my genetic results.

_____ I consent to the use of my DNA sample for future test validation and/or research purposes.

_____ I do not consent to the use of my DNA sample for future test validation or research purposes.

In the event that my sample is used for research purposes, the laboratory may wish to contact my physician/genetic counselor for additional information regarding my sample. This includes, but is not limited to, information on personal health and family history as it relates to genetic testing. If there are new developments in the field, my physician/genetic counselor may be contacted by the Genetic Diagnostic Laboratory staff to offer me the opportunity to have additional clinical testing. Indicate consent or denial to the above sentence by initialing below. My refusal to consent to research will not affect the reporting of my genetic results.

_____ I consent to be contacted by the Laboratory in the future for research purposes.

_____ I do not consent to be contacted by the Laboratory in the future for research purposes.

**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/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 and the associate risks and alternatives.

Patient’s Printed Name:_____________________________________

Patient’s Signature:_____________________________________

DOB:_____________ Date:_____________

(Or parent/Guardian if patient is a minor)

Name and Relationship:_____________________________________

(Parent/Guardian if patient is a minor)

10/01/2016