GENETIC DIAGNOSTIC LABORATORY
UNIVERSITY OF PENNSYLVANIA SCHOOL OF MEDICINE
DEPARTMENT OF GENETICS
560 Clinical Research Building • 415 Curie Boulevard • Philadelphia, PA 19104
Tel: (215) 573-9161 • Fax: (215) 573-5940• Email: gdllab@pennmedicine.upenn.edu
CLIA ID: 39D0893887

# REQUEST FOR WAGR SYNDROME TESTING

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

**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: _____

## CLINICAL INFORMATION

**ICD-10 CODE(S):*** ☐ C64.9 Wilms tumor ☐ Q52.9 Genital anomaly (female) ☐ R62.50 Developmental delay

☐ Q13.1 Aniridia ☐ Q55.9 Genital anomaly (male) ☐ Other: _____

☐ Q64.9 Urinary anomaly ☐ F79 Intellectual delay _____

ADDITIONAL CLINICAL SYMPTOMS: _____

_____

*If the test request is for 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):* _____

## TEST REQUESTED

☐ 11p13 high resolution copy number analysis

*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

GENETIC DIAGNOSTIC LABORATORY
UNIVERSITY OF PENNSYLVANIA SCHOOL OF MEDICINE
DEPARTMENT OF GENETICS
560 Clinical Research Building • 415 Curie Boulevard • Philadelphia, PA 19104
Tel: (215) 573-9161 • Fax: (215) 573-5940• Email: gdllab@pennmedicine.upenn.edu
CLIA ID: 39D0893887

# 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 | PHONE

## 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

GENETIC DIAGNOSTIC LABORATORY
UNIVERSITY OF PENNSYLVANIA SCHOOL OF MEDICINE
DEPARTMENT OF GENETICS
560 Clinical Research Building • 415 Curie Boulevard • Philadelphia, PA 19104
Tel: (215) 573-9161 • Fax: (215) 573-5940• Email: gdllab@pennmedicine.upenn.edu
CLIA ID: 39D0893887

# INFORMED CONSENT: GENETIC TESTING FOR WAGR SYNDROME

**Background:** Wilms tumor, aniridia, genitourinary anomalies, and range of developmental delays (WAGR) is a rare contiguous gene deletion syndrome. Diagnosis of WAGR allows for increased screening for Wilms tumor and determine any other appropriate medical management. The molecular basis of WAGR involves deletions of genes on chromosome 11p13, in particular the *PAX6* and *WT1* genes.

**Purpose:** I, or my child/fetus, will be tested for alterations in chromosome 11p13 as described above. The purpose of this genetic testing is to determine whether I, or my child/fetus, have specific molecular changes associated with WAGR.

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

**POSITIVE:** A clinically significant alteration is detected in the 11p13 region. I understand a positive molecular result is highly associated with WAGR, which can confirm a clinical diagnosis.
**NEGATIVE:** The analysis did not detect a molecular alteration associated with WAGR. This result reduces the likelihood that I/my child/fetus has a mutation related to WAGR. 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 WAGR.

**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 _____

**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)*

Initials _____