BAYLOR GENETICS

UCD GENETIC TESTING PROGRAM
SPONSORED BY HORIZON THERAPEUTICS PLC

BAYLOR GENETICS
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HOUSTON, TX 77021-2024

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UCD GENETIC TESTING PROGRAM REQUISITION

This requisition form can be used to submit an order for the UCD Genetic Testing Program, a sponsored testing program for genetic disorders brought to you by Horizon Therapeutics and Baylor Genetics. Please review the ordering options and complete all sections of this form.

PATIENT INFORMATION (COMPLETE ONE FORM FOR EACH PERSON TESTED)

Patient Last Name _____ Patient First Name _____ MI _____ Date of Birth (MM / DD / YYYY) ____ / ____ / ____

Address _____ City _____ State _____ Zip _____ Phone _____

Accession # _____ Hospital / Medical Record # _____

Biological Sex: ◯ Female ◯ Male ◯ Unknown

Gender identity (if different from above): _____

FAMILY HISTORY

Is there family history of disease for which the patient is being tested? ◯ Yes ◯ No

If yes, describe below and attach the family pedigree and/or clinical notes.

RELATIVE'S RELATIONSHIP TO THIS PATIENT	MATERNAL OR PATERNAL	DIAGNOSED CONDITION	AGE AT DIAGNOSIS

PAYMENT

◉ INSTITUTIONAL BILLING

Horizon Therapeutics (Institution Name) HPDI (Institution Code)

REPORTING RECIPIENTS

ORDERING CLINICIAN

Ordering Clinician _____ Institution Name _____

Email _____ Phone _____ Fax _____

ADDITIONAL RECIPIENTS

Name _____ Email _____ Fax _____

Name _____ Email _____ Fax _____

ORDERING OPTIONS

UCD GENETIC TESTING PROGRAM

For individuals that meet the eligibility criteria below and wish to receive the program specific genetic testing panels. Note, you must select below the appropriate eligibility criteria for this patient.

☐ Suspected diagnosis of UCD

UCD FAMILY FOLLOW-UP TESTING

For relatives of program participants who received a Pathogenic/Likely Pathogenic result or approved VUS who want to receive gene specific family follow-up testing at no additional charge (see page 3).

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STATEMENT OF MEDICAL NECESSITY (REQUIRED)

This test is medically necessary for the risk assessment, diagnosis, or detection of a disease, illness, impairment, symptom, syndrome, or disorder. The results will determine my patient's medical management and treatment decisions. The person listed as the Ordering Physician is authorized by law to order the test(s) requested herein and will be used for quality assessment purposes only. I confirm that I have provided genetic testing information to the patient and they have consented to genetic testing. The medical professional will retain evidence that the patient consented to genetic testing. The medical professional warrants that (i) he/she will not seek reimbursement for this no charge test from any third party, including but not limited to government healthcare programs; (ii) participation in the Program will not influence the his/her medical decisions; (iii) he/she is not obligated to purchase or prescribe any product or service offered by a sponsor of the Program; (iv) he/she is not obligated to participate in or to encourage patients to participate in any clinical trial or other research program conducted by a sponsor; and (v) he/she will participate in the Program in accordance with applicable laws. I attest that I am authorized under applicable law to order this test.

By signing this form, the medical professional acknowledges that the individual/family member authorized to make decisions for the individual (collectively, the "Patient") has been supplied information regarding and consented to undergo genetic testing, substantially as set forth in Provider's Informed Consent for Genetic Testing (http://baylorgenetics.com/sponsored-testing/ucd). The medical professional (i) warrants that he/she will not seek reimbursement for this no-cost test from any third party, including but not limited to federal healthcare programs and (ii) will inform the Patient that he/she shall not seek reimbursement for this no-cost test from any third party, including but not limited to federal healthcare programs. In addition to the above, I attest that I am the ordering physician, or I am authorized by the ordering physician to order this test, or I am authorized under applicable state law to order this test. I further attest that the Patient meets eligibility criteria for testing under the Program.

Physician's Printed Name _____ Physician's Signature _____ Date (MM / DD / YYYY) ____ / ____ / ____

SAMPLE (REQUIRED)

SAMPLE TYPE

◯ Blood in EDTA-tube (purple-top) ◯ Buccal Swab ◯ Saliva

Special Cases: ☐ History of/current hematologic malignancy in patient

DATE OF COLLECTION (MM/DD/YYYY)

____ / ____ / ____

NOTE: Extracted DNA/RNA will only be accepted if the isolation of nucleic acids for clinical testing occurs in a CLIA-certified laboratory or a laboratory meeting equivalent requirements as determined by the CAP and/or the CMS.

INDICATION FOR TESTING (REQUIRED)

☐ Symptomatic (Summarize below or attach clinical notes and/or laboratory notes)

☐ Symptomatic with Positive Family History

☐ Asymptomatic

◯ Population Screening ◯ Positive Family History

Clinical Symptoms (select all that apply):

- ☐ Acute neonatal encephalopathy
- ☐ Confusion, irritability, slurred speech
- ☐ History of unexplained infant death
- ☐ Lethargy
- ☐ Protein avoidance
- ☐ Seizures
- ☐ Unexplained acute liver failure
- ☐ Unexplained cerebral palsy
- ☐ Cerebral edema
- ☐ Frequent headaches
- ☐ Hypotonia
- ☐ Nausea/recurrent vomiting
- ☐ Respiratory alkalosis
- ☐ Stupor/coma
- ☐ Unexplained altered mental status

LABORATORY FINDINGS (IF APPLICABLE)

See more for the example.

	Patient Value / Reference Range
☐ Elevated plasma ammonia	_____ / _____
☐ Elevation of urine orotic acid, if available:	_____ / _____
☐ Abnormal plasma citrulline	◯ Low ◯ High
☐ Elevated plasma arginine	_____ / _____
☐ Elevated plasma glutamine	_____ / _____

MOLECULAR DIAGNOSTIC TESTS

MASSIVELY PARALLEL SEQUENCING (BCM-MitomeNGS℠) PANELS

	TEST CODE	TEST NAME	SAMPLE TYPE *
☐	2110	Urea Cycle Disorders (UCD) Genetic Testing Program	BE, BUC, DNA

If test code 2110 is negative or only a variant of uncertain significance (VUS) is identified in the OTC gene, reflex to test code 3143 (OTC Deletion/Duplication Analysis).

If test code 2110 results have a single heterozygous variant (VUS, Pathogenic or Likely Pathogenic variants), reflex to the gene with the appropriate test below for further analysis.

TEST CODE	GENE	TEST NAME
3428	ARG1	ARG1 Deletion/Duplication Analysis
3158	SLC25A13	SLC25A13 Deletion/Duplication Analysis
3238	SLC25A15	SLC25A15 Deletion/Duplication Analysis
3348	CPS1	CPS1 Deletion/Duplication Analysis
3353	NAGS	NAGS Deletion/Duplication Analysis
3143	OTC	OTC Deletion/Duplication Analysis
2001	ASS1 / ASL	For deletion/duplication analysis for the ASS1 or ASL genes individually
2003	Multiple Genes	For multi-gene deletion/duplication analysis

Panels continued on next page

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CUSTOM FAMILY SEQUENCING INFORMATION

If testing of proband is needed, see separate requisition "Proband Custom Sequencing Analysis," which can be found at www.baylorgenetics.com.

Name of First Patient Studied | Relationship to Patient Studied | Baylor Genetics Lab # | Family #

This Family Member is Currently:

◯ **ASYMPTOMATIC**
◯ **SYMPTOMATIC**

If SYMPTOMATIC, please provide details. Please attach additional pages, if needed.

Include a pedigree showing familial relationships.

☐ Copy of Original Results Attached (REQUIRED)

CUSTOM FAMILY SEQUENCING TEST

TEST CODE	TEST NAME	GENE NAME (REQUIRED)	MUTATION/UNCLASSIFIED VARIANT (REQUIRED)
☐ 3141	OTC - Familial Mutation/Variant Analysis		
☐ 3346	CPS1 - Familial Mutation/Variant Analysis		
☐ 3426	ARG1 - Familial Mutation/Variant Analysis		
☐ 3351	NAGS - Familial Mutation/Variant Analysis		
☐ 6361	ASL - Familial Mutation/Variant Analysis		
☐ 3156	SLC25A13 - Familial Mutation/Variant Analysis		
☐ 3236	SLC25A15 - Familial Mutation/Variant Analysis		
☐ 6185	ASS1 - Familial Mutation/Variant Analysis		

SAMPLE SPECIFICATIONS TABLE

ABBREVIATION	SAMPLE NAME	RECOMMENDED AMOUNT (2 YRS - ADULT)	RECOMMENDED AMOUNT (NEWBORN - 2YRS)	SHIPPING INSTRUCTIONS	SPECIAL NOTES
BE	Blood in EDTA tube (purple-top)	3 - 5 cc	2 - 3 cc	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.	
BUC	Buccal Swab	See Special Notes	See Special Notes		Collected with ORAcollect•Dx (OCD-100) self-collection kit (provided by Baylor Genetics with instructions). It is highly recommended that the sample be collected by a healthcare professional. Buccal swab is an accepted sample type for FMR1 CGG Repeat Expansion Analysis (test code 6573) only.
CB	Cord Blood	N/A	1 - 2 cc		Ensure properly labeled. Also send 3 cc of maternal blood in properly labeled EDTA tube for MCC studies at no charge as needed.
DNA	DNA, Extracted	10 -15 ug	10 -15 ug		Minimal concentration of 50ng/uL; A260/A280 of ~1.7
SA	Saliva	See Special Notes	See Special Notes		Collected with Oragene DNA Self-Collection Kit.

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INFORMED CONSENT FOR MOLECULAR DIAGNOSTIC TESTING

INFORMED CONSENT FOR GENETIC TESTING

TEST INFORMATION

This consent form will provide you with information regarding genetic testing, which you should discuss with your healthcare provider or a genetic counselor. In order to ensure that you have understood the purpose and significance of genetic testing, we have provided information about the testing process and potential results below.

The purpose of genetic testing is to identify the cause of a suspected disease in you or your family. The testing analyzes your genetic material (DNA) for an abnormal change (variant) that could explain the disease you or members of your family are experiencing. Genetic testing can be a diagnostic test, which is used to identify or rule out a specific genetic condition. Genetic screening tests are used to assess the chance for a person to develop or have a child with a genetic condition. Genetic screening tests are not typically diagnostic, and results may require additional testing.

The purpose of this test is to see if you or your child may have a genetic variant or chromosome rearrangement. This may cause a genetic disorder or may determine the chance that you or your child will develop or pass on a genetic disorder in the future. "Your child" can also mean your unborn child, for the purposes of this consent.

In a genetic test, depending on the case, you can be tested for:

- A single gene/variant responsible for a specific, suspected genetic disease.
- Multiple genes in parallel.

The sample/specimen that is needed to perform the genetic test is stated in the test order form and is typically blood or purified DNA, but may also saliva or buccal swab.

RESULTS

There are several categories of test results that may be reported including:

- **Positive:** Positive or "abnormal" results mean there is a change in the genetic material found that is related to your/your child's medical issues or that you/your child are at an increased risk of developing the disorder in the future. It is possible to test positive for more than one genetic variant.
- **Negative:** Negative or "normal" results mean no relevant genetic change related to your/your child's medical issues was detected. This does not mean there is no genetic change, but it may mean that the type of testing performed could not detect it.
- **Results of Unclear Significance:** Testing can detect change(s) in DNA which we do not yet fully understand. These alterations are also referred to as variants of uncertain significance (VUS). Additional studies may be recommended if a VUS is identified in a gene that may be associated with your/your child's medical concerns.

CONSIDERATIONS AND LIMITATIONS

- Results may indicate affected status, increased risk to someday be affected with, and/or reproductive risk for a genetic disorder. It is important to understand that genetic tests, even if negative, are not exhaustive. It is not possible to exclude risks for all possible genetic diseases for yourself and your family members.
- A positive test result is an indication that the individual(s) being tested may be predisposed to or have the specific disease or condition which prompted testing. You might consider additional independent testing, consult a personal physician, or pursue genetic counseling.
- It is possible that the knowledge of the test results may result in psychological stress for you and your family. It is always recommended to discuss the results with your healthcare provider or genetic counselor.

PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION

- If several family members are tested, the correct interpretation of the results depends on the provided relationships between family members. In rare cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. If a discrepancy is identified, it may be necessary to report this to the physician who ordered the testing.
- Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to, mislabeled samples, inaccurate reporting of clinical/medical information, or rare technical errors.
- Results will only be released to a licensed healthcare provider, to those allowed access to test results by law, and to those authorized in writing.
- In rare cases, persons with genetic diagnoses have experienced problems with insurance coverage and employment. The U.S. Federal Government has enacted several laws that prohibit discrimination based on genetic test results by health insurance companies and employers. In addition, these laws prohibit unauthorized disclosure of this information. For more information, you can visit www.genome.gov/10002077.
- Samples will be retained in the laboratory in accordance with the laboratory retention policy.
- After testing is complete, the de-identified submitted specimen may be used for test development and improvement, internal validation, quality assurance, and training purposes. DNA specimens are not returned to individuals or to referring heath care providers unless specific prior arrangements have been made.

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INFORMED CONSENT FOR MOLECULAR DIAGNOSTIC TESTING

INFORMED CONSENT FOR GENETIC TESTING

PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION (CONT.)

- Samples from residents of New York State will not be included in the de-identified research studies described in this authorization and will not be retained for more than 60 days after test completion, unless specifically authorized by your selection. No tests other than those authorized shall be performed on the biological sample.
- Information including results, indications for testing and clinical status obtained from this testing may be shared with healthcare providers, scientists and healthcare databases or used in scientific publications or presentations, but the personal identifying information of all persons studied will not be revealed in such data sharing or publications/presentations.

RESEARCH & RECONTACT CONSENT

For more information on research at Baylor Genetics, please visit baylorgenetics.com. Please read the below statements carefully and check the appropriate box.

Note: If left blank, consent is interpreted as "NO."

☐ I agree to use of my de-identified specimen for research to improve genetic testing for all patients and contribute to scientific research.

☐ I am a New York State Resident, and I give Baylor Genetics permission to store my specimen in accordance to the laboratory retention policy for internal quality assurance and possible research studies.

☐ In addition to agreeing above, I agree to be contacted by Baylor Genetics regarding research opportunities.

PATIENT AUTHORIZATION

By signing this statement of consent, I acknowledge that I have read and understand the informed consent for genetic testing. I have received appropriate explanations from my physician regarding the purpose, scope, type and significance of the planned genetic testing and achievable results. All my questions have been answered and I have had the necessary time to make an informed decision about the genetic test.

I give permission to Baylor Genetics to conduct genetic testing as recommended by my physician.

Patient Signature

Date (DD/MM/YYYY)

Printed Name