INVITAE

UCD Genetic Testing Program
Genetic testing for urea cycle disorders (UCDs)
SPONSORED BY HORIZON PHARMA

ORDER ID
For Invitae internal use only

Requisition Form
UCD Genetic Testing Program TRF932-2

This requisition form can be used to submit a specimen for the UCD Genetic Testing Program, a complimentary testing program for urea cycle disorders sponsored by Horizon Pharma and performed by Invitae. Please confirm that the patient meets the eligibility requirements for the program. To submit orders for genetic testing outside of this program, please order through Invitae's online portal or use a standard requisition form, www.invitae.com/order-forms.

PROGRAM ELIGIBILITY:

A suspected diagnosis of a urea cycle disorder OR a family history of urea cycle disorder.

PATIENT INFORMATION

First name	MI	Last name
Date of birth (MM/DD/YYYY)	Sex ◯ M ◯ F	MRN (medical record number)

Ancestry ◯ Asian ◯ Black/African American ◯ White/Caucasian ◯ Ashkenazi Jewish ◯ Hispanic ◯ Native American ◯ Pacific Islande ◯ French Canadian ◯ Sephardic Jewish ◯ Mediterranean ◯ Other:

Phone	Email address
Address	City
State	ZIP code / Country

SPECIMEN INFORMATION

Label each tube with the patient's full name, date of birth, and specimen collection date.
A requisition form MUST accompany each specimen. www.invitae.com/specimen-requirements

Specimen type : ◯ Blood ◯ Saliva ◯ Assisted saliva ◯ DNA - source:

DNA must be extracted in a CLIA or other suitably certified laboratory

We are unable to accept blood/saliva from patients with:

- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Collection date (MM/DD/YYYY) *If not provided, date will be 1 day prior to our receipt of specimen. For DNA, provide date retrieved from archive.*

Special cases : ◯ History of/current hematologic malignancy ◯ Resubmission

REASON FOR TESTING

Previous results *(if applicable and not included in clinical criteria)*

ORGANIZATION INFORMATION

Organization name and address

Organization name	
Phone	Fax
Address	City
State / ZIP code	Country: **United States**

Primary clinical contact

Name	Role/title
Phone	NPI
Email address (for report access)	

Ordering physician

◯ **Same as primary clinical contact**

Name	NPI
Email address (for report access)	

Additional clinical or laboratory contact (optional)

Name	Email address (for report access)

INVITAE PARTNER CODE	UCD

Family history (optional; please fill out all that apply

☐ Family history of UCD

Familial gene and variant (if known): _____

If family member was tested at Invitae, RQ# (if known): _____

Clinical symptoms (optional; please check all that apply)

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

Laboratory findings

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	_____ /_____

ASSAY

Invitae is a CAP-accredited and CLIA-certified clinical diagnostic laboratory performing full-gene sequencing and deletion/duplication analysis using next-generation sequencing technology (NGS). Search for details on the analysis of any gene in our test catalog at **www.invitae.com/physician/search**.

Invitae continually updates its panels based on the most recent evidence. Please note that if an order is placed using an older version of this form, Invitae reserves the right to upgrade any ordered panel(s) to the current version(s).

TESTS INCLUDED IN THE PROGRAM

Test code	Test name	# of genes	Gene list
○ 06230	Invitae Hyperammonemia Panel	58	ABCD4, ACADM, ACADVL, ALDH18A1, ARG1, ASL, ASS1, BCKDHA, BCKDHB, BTD, CA5A, CPS1, CPT1A, CPT2, DBT, DLAT, DLD, ETFA, ETFB, ETFDH, GLUD1, GLUL, HADHA, HADHB, HCFC1, HLCS, HMGCL, IVD, LMBRD1, MCCC1, MCCC2, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, NAGS, OAT, OTC, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC7A7, TAZ, TMEM70, UMPS

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 Invitae's Informed Consent for Genetic Testing (www.invitae.com/patient-consent) and in connection with the Program. 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. The medical professional also hereby acknowledges that organization and clinician contact information provided in the order may be shared with third parties that may contact the medical professional directly in connection with the Program, and that they have made the Patient aware that third parties may contact their medical professional regarding de-identified information gathered through the Program. 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.

Medical professional signature	Date