INVITAE

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

Urea Cycle Disorder Genetic Testing Program Eligibility Criteria

FM179-1

People with a suspected diagnosis of a urea cycle disorder or a family history of urea cycle disorders are invited to take part in the UCD Genetic Testing Program, sponsored by Horizon Pharma and performed by Invitae.

The patient must meet the eligibility criteria below to qualify for the program.

Please fill out, print, and sign this checklist and include it when sending in the specimen. Additional information about the ordering process can be found at www.invitae.com/UCD.

To submit orders for genetic testing outside of this program, please order through Invitae's online portal or use a standard requisition form, accessible at www.invitae.com/order-forms.

Patient name	Date of birth (MM/DD/YYYY)

UCD GENETIC TESTING PROGRAM ELIGIBILITY CRITERIA

A suspected diagnosis of a urea cycle disorder or a family history of urea cycle disorders.

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

Patient value/reference range

Laboratory findings:

- ☐ Elevated plasma ammonia _____ /_____
- ☐ Elevation of urine orotic acid, if available: _____ /_____
- ☐ Abnormal plasma citrulline ☐ LOW ☐ HIGH
- ☐ Elevated plasma arginine _____ /_____
- ☐ Elevated plasma glutamine _____ /_____

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**