Children's Memorial Hospital
Center for Autonomic Medicine in Pediatrics & Molecular Diagnostic Laboratory
2300 Children's Plaza, Chicago, Illinois 60614-3394
Phone: 773.880.8188 & 773.880.3015
CLIA: 14D0424464

REQUEST FOR CONGENITAL CENTRAL HYPOVENTILATION SYNDROME (CCHS) *PHOX2B* SEQUENCING*

PHOX2B Gene Sequencing Test

The gene sequencing test is performed by the Center for Autonomic Medicine and the Molecular Diagnostic Laboratory at Children's Memorial Hospital (CMH). The entire coding sequence together with the intronic splice sites are sequenced and analyzed. This test will detect polyalanine-repeat mutations as well as known and novel non-polyalanine repeat mutations. **For further clarification about this test and its clinical utility or about CCHS, please contact Dr. Weese-Mayer at** DWeese-Mayer@ChildrensMemorial.org.

Specimen Preparation

Three to 9 cc of blood in an EDTA vacutainer is required. Call Dr. Mike Yu in the laboratory if less than 3cc of blood is available (773.880.3015). Please have the blood sent at room temperature by overnight delivery service (*e.g.* Federal Express). Because of transportation issues, it is best not to obtain blood on Friday, Saturday or Sunday. If kept overnight before sending, the blood should be refrigerated. **DO NOT FREEZE**. No refrigeration is needed for same day deliveries. A turnaround time of 1 to 2 weeks is anticipated for the sequencing test. Please include the requesting physician's complete mailing address, e-mail address, and phone and fax numbers. A FULLY completed requisition must accompany EACH blood sample.

Payment Arrangements

The patient can pre-pay with a cashier's check or credit card or we can bill the referring institution. Pre-payment is required for all samples from outside the U.S. Children's Memorial Hospital will not bill third party payors (e.g. insurance, Medicare, Medicaid) for this testing. Payment is the responsibility of the submitting entity, and CPT codes are provided here as a reference (83890x1, 83912x3, 83904x6, 83898x3, 83892x3, 83891x6). **For queries about pricing, the sequencing test, specimen requirements, or this requisition, please contact Dr. Jennings at** Ljennings@ChildrensMemorial.org.

***All test results will be reviewed by Drs. Weese-Mayer and Jennings (Children's Memorial Hospital, Northwestern University) and Dr. Berry-Kravis (Rush University Medical Center). Additional information and medical records may be requested**.

Last Revised 06/09/08

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PLEASE THOROUGHLY COMPLETE THIS FORM TO AVOID DELAYING THE PROCESSING OF THE SAMPLE.

❑ **I am ordering sequencing for the entire coding region and intronic splice sites of the *PHOX2B* gene**

Patient Information: ***If submitting samples from multiple family members, please attach a pedigree.***

Name of patient: _____
last *first*

Date of Birth: ___/___/____ **Gender:** ❑ **Male** ❑ **Female**
MM/DD/YYYY

Ethnicity: ❑**Caucasian** ❑**African American** ❑**Hispanic** ❑**Asian Pacific Islander**
❑**Native American** ❑**Other**_____

Home phone number: _____ **Cell phone number:** _____

Address: _____

City: _____ **State:** _____ **Zip:**_____ **Country:** _____

Date of Blood Draw: ___/___/____ **Date Blood sent:** ___/___/____
MM/DD/YYYY *MM/DD/YYYY*

Reason for ordering test: _____

Diagnoses (check all that apply):

❑ **CCHS**	❑ **Asleep ventilator needs**
❑ **Hirschsprung disease**	❑ **Mechanical ventilation via tracheostomy**
❑ **Neuroblastoma**	❑ **BiPAP via mask ventilation**
❑ **Other neural crest tumor**	❑ **Negative pressure ventilation**
❑ **Apparent life threatening events (ALTEs)**	❑ **Diaphragmatic pacers**
❑ **Unresolved apnea of prematurity**	❑ **Awake ventilator needs**
❑ **Unresolved apnea of infancy**	❑ **Mechanical ventilation via tracheostomy**
❑ **Cardiac Pacemaker**	❑ **Diaphragmatic pacers**
❑ **Esotropia** ❑ **Exotropia** ❑ **Fixed and dilated pupils**	❑ **Tracheostomy**
Age at diagnosis of alveolar hypoventilation: _____	
Other pertinent information:	

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Physician Information: ***(results will be sent to physician)***

Name: (last)_____ **(first)**_____
Phone: _____ **Fax:** _____
Address: _____
City: _____ **State:** _____ **Zip:** _____ **Country:** _____
e-mail: _____

Billing Information: **CMH will not bill third party payors (e.g. insurance, Medicare, Medicaid) for this testing. Payment is the responsibility of the submitting entity.**

Pre-payment by cashier's check or credit card is r*equired for all samples referred from outside of U.S.*

❑**Check enclosed (payable to Children's Memorial Hospital)**
❑**Credit Card** ❑**Visa** ❑**MasterCard** ❑**Discover**

Account #: _____ **Expiration date:** ___/___
MM/YY

Card Holder's Name: _____**Phone #:** _____

Signature: _____ **Date:** ___/___/____
MM/DD/YYYY

❑**Institutional Billing**

Contact Person/Title:_____
Name of Institution:_____
Section/Laboratory:_____
Address: _____
City:_____ **State:**_____ **Country:**_____ **Zip:**_____
Phone: _____ **FAX:**_____
e-mail:_____

Shipping Information:

HLA and Molecular Diagnostic Laboratories
Mailbox: 262 Room: M0.55
Children's Memorial Hospital
2300 Children's Plaza
Chicago, IL 60614-3394
Phone: 773.880.3015
Fax: 773.880.3790

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