

**Children's Hospital Boston  
DNA DIAGNOSTIC  
LABORATORY REQUISITION**



**DNA Diagnostic Lab**

[CLIA #: 22D0001844]  
Attention: Lab Control  
300 Longwood Ave., Farley 7  
Boston, MA 02115  
Phone # 617.355.7582  
Fax # 617.730.0338

**TO BE USED FOR PATIENTS OUTSIDE  
CHILDREN'S HOSPITAL**

*Institution* \_\_\_\_\_ *Client #* \_\_\_\_\_

**Billing should be sent to:** *(All tests will be billed to the institution)*

*Contact Person:* \_\_\_\_\_

*Address* \_\_\_\_\_

*City* \_\_\_\_\_ *State* \_\_\_\_\_ *Zip* \_\_\_\_\_

*Country* \_\_\_\_\_

*Phone #* \_\_\_\_\_ *Fax #* \_\_\_\_\_

**Report should be faxed to:**

*Attention:* \_\_\_\_\_

*Phone #* \_\_\_\_\_ *Fax #* \_\_\_\_\_

**Patient Name** \_\_\_\_\_ **MR #** \_\_\_\_\_

**DOB** \_\_\_\_\_ **Gender** \_\_\_\_\_ **Ethnicity** \_\_\_\_\_

**Date Specimen Collected** \_\_\_\_\_

**Referring Physician:** \_\_\_\_\_

**Pager/phone #** \_\_\_\_\_ **Fax #** \_\_\_\_\_

**Email:** \_\_\_\_\_

- *DNA tests require 3-5 cc of whole blood in purple top /EDTA tube.*
- *All specimens should be packed at room temperature and sent by overnight delivery.*
- *Send specimens with this Requisition completely filled out and an Informed Consent if applicable, or specimen will **NOT** be processed.*
- *All tests will have a professional interpretation.*

**CONGENITAL CRANIAL DYSINNERVATION DISORDER PANEL** *Check box for testing requested:*

- CFEOM1 (KIF21A) Mutation [Exon 8,20,21 – mutation hotspots only]
- CFEOM1 (KIF21A) Mutation [Exon 8,20,21. If no mutation is detected, the DNA sample will be transferred to the Engle lab where the remaining *KIF21A* exons will be screened. **Informed consent** required.]

*Confirmatory testing for individuals already enrolled in the Engle Lab study and for whom a mutation has been detected on a research basis:*

- CFEOM1 (KIF21A research mutation confirmation)
- CFEOM2 (PHOX2A research mutation confirmation)
- Okihiro syndrome-DRRS (SALL4 research mutation confirmation)

**CLINICAL EXAMINATION** Patient's CCDD is Sporadic\_\_ Familial\_\_ (who else is affected? \_\_\_\_\_)

Ptosis: Right\_\_ Left\_\_ Severe/moderate/mild? \_\_\_\_\_ Symmetric? \_\_\_\_\_

Primary globe position:

Vertical: Hyper/hypo/orthotropic? Right\_\_\_\_\_ Left\_\_\_\_\_

Horizontal: Exo/Eso/orthotropic? Right\_\_\_\_\_ Left\_\_\_\_\_

Describe residual globe movements \_\_\_\_\_

- Patient can elevate right\_\_ left\_\_ eye above the horizontal midline
- Patient has aberrant movements (describe) \_\_\_\_\_

Pupil and fundus exam: \_\_\_\_\_

Other findings: \_\_\_\_\_