

## Specialty Clinical Laboratories

1050 Forest Hill Road  
Staten Island, NY 10314

### Laboratory of Molecular Cytogenetics

Milen Velinov, M.D.,Ph.D.

Director,SCL Genetic Testing, Dept. Hum.Genetics

Telephone: 718-494-5219/Fax: 718-494-1026

E-mail: : [milen.velinov@opwdd.ny.gov](mailto:milen.velinov@opwdd.ny.gov)

#### PATIENT'S INFORMATION

<u>Last Name</u>	<u>First Name</u>	<u>M.I.</u>	<u>D.O.B.</u> / /	<u>SEX</u> <input type="checkbox"/> M <input type="checkbox"/> F	<u>DIAGNOSIS: ICD-9CM</u>
<u>Address</u> (Street)	(Apt. #)	(Telephone #)	<u>FACILITY CONSECUTIVE # / PATIENT ID #</u>		
(City)	(State)	(Zip)			

#### PERSON RESPONSIBLE FOR BILL (OUTPATIENTS ONLY)

<u>Last Name</u>	<u>First Name</u>	<u>INSURANCE CARRIER COMPLETED INSURANCE FORM OR COPY OF MEDICAID CARD REQUIRED</u> I.D. #			
<u>Address</u> (Street)	(Apt. #)	(Telephone #)			
(City)	(State)	(Zip)			

#### PHYSICIAN INFORMATION

<u>Last Name</u>	<u>First Name</u>	<u>MMIS#</u> Or Lic. #	<u>State</u>	<u>Number</u> Facility
<u>Address</u> (Street)	(Apt. #)	(City)	(State)	(Zip)
Fax ( ) Telephone ( )	<u>PHYSICIAN'S SIGNATURE</u>			<u>DATE:</u> / /

#### By law, test cannot be performed without physician and patient authorization

Collection Date / /	<input type="checkbox"/> Initial Study <input type="checkbox"/> Follow-up	<b>Specimens Accepted Monday – Friday</b>
------------------------	--	---

#### PLEASE CHECK TEST(S)

- Krabbe disease-Patient GALC common mutation  
 Krabbe disease-Parental carrier testing for the common GALC mutation

The result from the testing for GALC activity (Krabbe disease enzyme) was

- positive  
 negative  
 not done

#### SPECIMEN INFORMATION

- Whole Blood: One Purple-Top Tube (sodium EDTA)  
 Submit specimens immediately in sterile tubes on blue ice (not frozen)

...For Lab Use Only...

Accession Number	Date:	By:	Reviewed By:
Date Rec'd in Lab			
M#			

## Consent form

### Specialty Clinical Laboratories

1050 Forest Hill Road  
Staten Island, New York 10314

### Laboratory of Molecular Cytogenetics

Milen Velinov, M.D.,Ph.D.  
Director, SCL DNA Laboratory

Telephone: 718-494-5219/Fax: 718-494-1026  
E-mail: [milen.velinov@opwdd.ny.gov](mailto:milen.velinov@opwdd.ny.gov)

#### Consent for testing for the common 30 kb deletion of the GALC gene associated with Krabbe disease

##### **Purpose of test**

To determine if your child has the common 30 kb deletion of the gene GALC, associated with Krabbe disease. You/your child will be asked to donate a blood sample by venipuncture.

##### **Description of the disease/condition**

Krabbe disease is a severe neuro-degenerative disorder that presents with regression in the development, joint stiffness, seizures and other neurological manifestations.

##### **Result interpretation and test limitations**

A positive test result is an indication that you/your child have two gene mutations leading to Krabbe disease or you/your child is a carrier of one mutation for the disease.

The test identifies gene mutations in 35-45% of the gene copies in individuals that are already diagnosed with Krabbe disease. The rest of these individuals may have gene changes that are not identifiable with this testing. Failure of this test is also possible due to technical limitations/mishandling, but it is unlikely.

##### **Specimen retention**

The specimens tested will be retained in the laboratory for 60 days in case further/repeat testing is necessary. No tests other than those authorized or needed to confirm a result shall be performed on the sample unless an additional consent for testing is signed by you.

##### **Confidentiality of the obtained results**

The results of this testing are confidential and will be only sent to or discussed with the healthcare provider who requested the testing. The results may be released to other parties only after additional consent for release of information is signed by the patient's parent/legal guardian.

I understand the above and give consent for diagnostic testing

\_\_\_\_\_  
Signature of Subject/or Parent/Guardian

\_\_\_\_\_  
Date

\_\_\_\_\_  
Physician/Genetic Counselor

\_\_\_\_\_  
Date