

## COMPLETING A DRY BLOOD SPOT SCREENING TEST IS EASY WITH INNOMAR CLINICS'™ FREE TESTING SUPPORT

1



### INFORMED CONSENT/ CLINICAL DETAILS

- 1) **Obtain Patient Consent**  
HCP can sign the physician copies (3) indicating consent has been obtained  
**OR**  
Patient can sign the patient copies (3)
- 2) Affix one **barcode sticker** to each consent copy
- 3) **Insert (1) signed copy into the DBS kit** (lab will not perform test without this)
- 4) Place (1) copy in the patient's chart (for test result tracking)
- 5) Give (1) copy to the patient
- 6) Complete relevant **clinical information and contact details (incl. HCP email or fax)** on DBS card
- 7) **Patient signs Centogene DBS Requisition Form** (consent to be contacted for test scheduling)

### SCHEDULE TEST FOR YOUR PATIENT:

**FAX signed Centogene DBS requisition and signed Centogene consent to: 1-844-951-7284**

(Patient will be contacted to schedule the test at a nearby Innomar clinic )

**OR**

Call 1-844-691-7284  
(Mention you are calling to schedule a Centogene DBS test)

2



### PATIENT LEAVES WITH DBS KIT

(incl. **signed consent**)

**Innomar will contact patient directly to schedule test**

3



### PATIENT PROCEEDS TO CLINIC

on scheduled date with DBS kit for free testing

4



### HCP RECEIVES RESULTS

**4 weeks** later via an email link to a **secure online portal or by fax**

5



### TO FOLLOW UP ON TEST RESULTS:

report.support@centogene.com

### FOR GENERAL QUESTIONS:

info@dbskit.ca

DBS: dry blood spot

# HOW TO PERFORM A DRY BLOOD SPOT SCREENING TEST YOURSELF

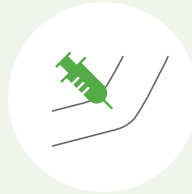
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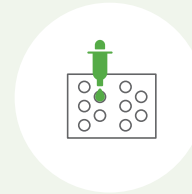
2



## COMPLETE VENOUS BLOOD DRAW USING EDTA PROCEDURES

(1 mL)

3



## ADD 50 µL (OR 3 DROPS) TO EACH SPOT,

ensuring uniform saturation and spot visible from back of card.

**Be careful not to overfill.**

4



## DRY AT LEAST 3 HOURS

at room temperature on a flat surface.

**Do not place in direct sunlight.**

5



## ONCE DRY, SEAL CARD IN PLASTIC ENVELOPE.

**Within 1 week, mail card and consent to lab for analysis.**

6



## HCP RECEIVES RESULTS

**4 weeks** later via an email link to a **secure online portal or by fax.**

7



## TO FOLLOW UP ON TEST RESULTS:

report.support@centogene.com

**FOR GENERAL QUESTIONS:**

info@dbskit.ca

# DBS TEST FAQ

## Q: WHAT DO THE DBS TEST RESULTS LOOK LIKE?

A: Sample test results from Centogene lab (for illustrative purposes only) below:

### POSITIVE RESULT

#### CLINICAL INFORMATION

Anhidrosis / hypohidrosis, functional abnormality of the gastrointestinal tract, paresthesia.



#### POSITIVE RESULT

Pathogenic or likely pathogenic variant(s) identified

#### INTERPRETATION

A heterozygous pathogenic variant was identified in the GLA gene. Additionally, the concentration of lyso-Gb3 biomarker was found within pathologic range. The genetic diagnosis of Fabry disease is confirmed.

#### RECOMMENDATIONS

- Genetic counselling is recommended
- Monthly analysis of the lyso-Gb3 biomarker concentration

### NEGATIVE RESULT

#### CLINICAL INFORMATION

Alpha-galactosidase activity was normal, but close to lower normal reference limit.



#### NO GENETIC DIAGNOSIS

#### INTERPRETATION

We have not detected any clinically relevant variant in your patient. The genetic diagnosis of Fabry disease is not confirmed.

### INCONCLUSIVE RESULT

#### CLINICAL INFORMATION

**No clinical information has been provided.**

**Please note that the quality of the interpretation of the patient's genetic data can be negatively influenced by missing clinical information.**



#### CARRIER STATUS

Variant(s) of uncertain significance identified

#### INTERPRETATION

A heterozygous variant of uncertain significance was identified in the GLA gene. Additionally, the concentration of lyso-Gb3 biomarker was found within normal range. This variant has recently been associated with multifocal white matter lesions (Lender et al., 2013). Further analyses are recommended to evaluate the clinical significance of the detected variant. These results are consistent with Fabry disease carrier status.

#### RECOMMENDATIONS




- Genetic counselling for your patient and other relevant family members, to explain the results and address any concerns.
- Testing a second independent sample from the patient in order to confirm the results (CMGS best practice guidelines).
- Routine analysis of lyso-Gb3 concentration.
- To evaluate the clinical significance of the detected variant, we recommend that you send us detailed clinical information about the patient.

# DBS TEST FAQ

## Q: HOW DO I EXPLAIN THE DBS TEST RESULTS TO MY PATIENT?

**A:** Disclosing a positive result can be challenging. Please consider informing your patient of a positive DBS result while explaining that they are being referred to a genetic/metabolic care treatment centre for further assessment. Let the patient know that a diagnosis cannot be confirmed until additional investigations have been completed.

## WHAT TO DO WHEN YOU RECEIVE THE DBS KIT RESULTS:

Result 	Interpretation 	Recommended course of action 
<b>POSITIVE</b>	A positive result suggests that the patient has the condition in question.	<ul style="list-style-type: none"><li>• Send referral with DBS results to nearest treatment centre for confirmatory investigations</li><li>• Genetic counselling is recommended</li></ul>
<b>NEGATIVE</b>	A negative result decreases the likelihood that the patient has the condition in question.	<ul style="list-style-type: none"><li>• Consider other possible causes for the patient's clinical presentation</li><li>• If clinical suspicion of the condition in question remains high, refer the patient to the nearest treatment centre for an assessment</li><li>• Consider referring your patient to a genetic counsellor for further explanation of the results</li></ul>
<b>INCONCLUSIVE</b>	An inconclusive result neither confirms nor rules out the condition in question.	<ul style="list-style-type: none"><li>• Consider repeating the DBS kit and/or</li><li>• Send referral with DBS results to nearest treatment centre for confirmatory investigation</li><li>• Consider referring your patient to a genetic counsellor for further explanation of the results</li></ul>

## DBS TEST FAQ

### **Q: WHAT IF I NEED HELP INTERPRETING THE DBS TEST RESULTS?**

**A:** Contact a consultant genetic counsellor at no charge (details below), or refer your patient, along with the DBS test results, to your nearest genetic/metabolic care treatment centre for further evaluation.

### **Q: I HAVEN'T RECEIVED MY DBS TEST RESULTS WITHIN THE EXPECTED 4-WEEK TIMEFRAME. HOW DO I FOLLOW UP?**

**A:** Email [report.support@centogene.com](mailto:report.support@centogene.com) and reference the barcode number from the DBS kit.

### **Q: WILL INCLUDING DBS RESULTS WITH A REFERRAL REDUCE THE WAIT TIME FOR MY PATIENT TO BE SEEN?**

**A:** Including your patient's DBS results in their referral to a genetic/metabolic care treatment centre is informative and may allow them to be seen more quickly.

### **Q: WHERE CAN I FIND MORE INFORMATION ON LYSOSOMAL STORAGE DISORDERS AND TREATMENT CENTRES?**

**A:**

**Fabry disease:**

[www.fabrycanada.com](http://www.fabrycanada.com)

**Gaucher disease:**

[www.gauchercanada.ca](http://www.gauchercanada.ca)

**MPS diseases:**

[www.mpssociety.ca](http://www.mpssociety.ca)

**Canadian Association of Genetic Counsellors:**

<https://www.cagc-accg.ca/>

For additional guidance regarding how to discuss DBS test results with your patient (at no charge), please contact consultant bilingual genetic counsellor:

Breanne Dale, MSc. CGC

Tel.: 416-953-4077

Email: [dalebreanne@gmail.com](mailto:dalebreanne@gmail.com)

**Confidentiality will be strictly maintained.**



# PHYSICIAN DBS CHECKLIST

## DID YOU REMEMBER TO:



- OBTAIN PATIENT CONSENT**
- Affix one **barcode** sticker to each of three (3) consent forms
- Insert one copy of the signed consent copy back into the kit** (lab will not conduct test without this)
- Keep one copy of the consent form for the patient file
- Give one copy of the consent form for the patient to keep



- COMPLETE RELEVANT CLINICAL INFORMATION AND CONTACT DETAILS (HCP EMAIL OR FAX) ON DBS CARD**



- SCHEDULE TEST FOR YOUR PATIENT**  
FAX signed Centogene DBS requisition and signed Centogene consent to:  
**1-844-951-7284** (Your patient will be contacted to schedule the test at a nearby clinic)  
**OR**
- Call 1-844-691-7284 (Mention you are calling to schedule a Centogene DBS test)

**Centogene DBS Requisition Form**

Please fax a signed copy of this form **AND** signed Centogene consent to:

**FAX: 1-844-951-7284**

(Patient will be contacted by Innomar to schedule test at a nearby clinic)

**Patient Information**

Last Name:	First Name:	Date of Birth: ____/____/____ DD    MMM    YYYY
Phone Number:	Best Time to Reach:	

**Physician Information**

Physician Last Name:	Physician First Name:	Telephone:
Fax:	Email:	

Preferred Method of Communication:  Phone  Fax  Email (to receive confirmation of test completion)

**Patient Consent**

I consent to be contacted by Innomar Strategies, the Administrator(s) and employees/consultants for the purposes of scheduling and conducting the DBS (Dry Blood Spot Testing). I understand I can withdraw consent at any time:     Yes     No    Initials \_\_\_\_\_

\_\_\_\_\_  
Signature of Patient or Legal Representative(s)

\_\_\_\_\_  
Printed Name of Patient or Legal Representative(s)

\_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_  
DD    MMM    YYYY  
Date