Instructions for contract of Sanofi HRS

Lysosomal Storage Diseases Laboratory

National Taiwan University Hospital

The aims of the National Taiwan University Hospital (NTUH) lysosomal storage disease (LSD) laboratory are to bring diagnoses to patients to enhance their management and treatment.

Tests available for dried blood spot (DBS)

- Fabry disease
- Pompe Disease
- Gaucher Disease
- Mucopolysaccharidoses (MPSs): type I & II
- Measuring DBS Lyso-Gb3 (Fabry Disease), Lyso-GL1 (Gaucher Disease) and Lyso-SM (Niemann-Pick type A/B disease)

Tests available for dried blood spot (DBS)

Genetic confirmation of the following diseases

- Fabry disease
- Pompe Disease
- Gaucher Disease
- Mucopolysaccharidoses (MPSs): type I & II
- Niemann-Pick type A/B disease

Procedures

1. **Fill in test request form on NTUH Lab Testing Service e-Platform according to the test request reference (page 9-10), email NTUH to inform the shipping date**

   Miss Teng, Chiao-ning: ntuh.lsdlab@gmail.com

   cc. Dr. Hwu: hwuwlntu@ntu.edu.tw

2. **Obtain informed consent from patients & physicians (Page 3-4)**
3. **Prepare samples and the package (Page 5-6)**
4. **Fill/Sign in the invoice form (Page 7-8)**
5. **Copy the invoice form and the Air Waybill and email them to NTUH**
6. **Send the package in room temperature which contains:**
   A. Samples
   B. Test request form (printed from e-platform)
   C. Inform consent form
   D. Invoice form

8. **Results will be sent through e-platform in 4 weeks**

Informed Consent Form (For customs clearance)

To be completed by the patient/parent/guardian

I understand that Lysosomal Storage Diseases are not common in our region and related diagnostic and monitoring tests are not performed in our places. Therefore I agree to have my blood and urine samples to be sent to National Taiwan University Hospital lysosomal storage disease laboratory for testing.

Printed name of the patient : _____

Signature of the patient/parent/guardian : _____

Date: _____/_____/_____

Day Month Year

知情同意書（通關用）

由病人/家屬/監護人填寫

我了解各溶小體儲積症在此地區並非為常見的疾病，因此無法提供相關的診斷或監測檢驗項目。我同意將本人的血液及尿液檢體送到臺大醫院生化遺傳研究室作檢測用途。

病人姓名：_____

病人簽名：_____

日期：_____/_____/_____

日 月 年

To be completed by the Health Professional

We promise that we have informed this patient/parent/guardian regarding results and limitations of this test and get patient's informed consent before sample collection. We assure you that these human samples are not infected by HBV, HCV, HIV-1, or HIV-2 and the exportation of those samples is not restricted in my country.

Printed name of person who obtains consent : _____

Signature of person who obtains consent : _____

Affiliation: _____

Date: _____/_____/_____

Day Month Year

由醫療人員填寫

我們承諾已將本檢測結果和檢測限制告知病人/家屬/監護人，並在檢體採集前獲得患者的知情同意。我們保證，這些人類檢體未感染 HBV、HCV、HIV-1 和 HIV-2，並且這些檢體的出口在我國不受限制。

醫療人員姓名：_____

醫療人員簽名：_____

醫療機構：_____

日期：_____/_____/_____

日 月 年

Prepare and send dried blood/urine spots

Prepare dried blood spots (DBS)

1. Blood sampling by either way (blood clotting is not acceptable)

- Venous blood with or without heparin
- Finger/heel prick with or without a heparin capillary tube

2. Apply blood to the center of the circle on a newborn screening card

- Mixed well blood sample
- Allow the blood to soak through the circle
- Fill all circles
- Do not apply blood twice to a same circle
- Do not apply to the other side of the circle

3. Dry the filter for 4 hours

- Avoid sun shine. Do not heat
- Filter can be dried overnight in an air conditioned room

4. Seal in a plastic bag and store frozen.

Sending dried blood

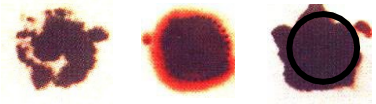
1. Put in a FedEx envelop

2. Can be sent at room temperature but need to avoid heating of the samples

3. Send by International Priority

Correct:

Incorrect:

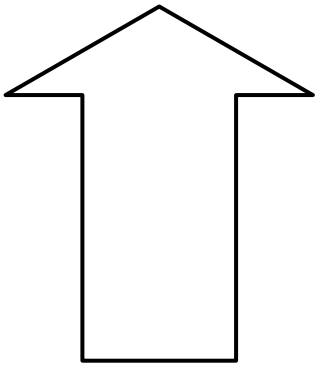


Label of package

Testing for lysosomal storage disease

Non-infectious

Store at 15~25°C



UP

Invoice form (Please fill in the QTY and calculate and sum the VALUE)

Commercial Invoice

INTERNATIONAL CARRIER: FedEx

AIRBILL NUMBER:

(NOTE: send 3 copies with each box. Each box should have its own set of attached documentation)

DATE OF EXPORTATION	EXPORT REFERENCES (i.e., order no., invoice no., etc.)
SHIPPER/EXPORTER SHIPPER:	CONSIGNEE (complete name and address) Wuh-Liang Hwu (Paul), M.D., Ph.D. Department of Medical Genetics Room 19004, 19F, Children's Hospital Building National Taiwan University Hospital No. 8 Chung Shan South Road Taipei 10041, Taiwan TEL: 02-23123456 ext 71939 FAX: 02-23314518 E-mail: hwuwlntu@ntu.edu.tw ; ntuh.lsdlab@gmail.com
COUNTRY OF EXPORT	
COUNTRY OF MANUFACTURE	
COUNTRY OF ULTIMATE DESTINATION	

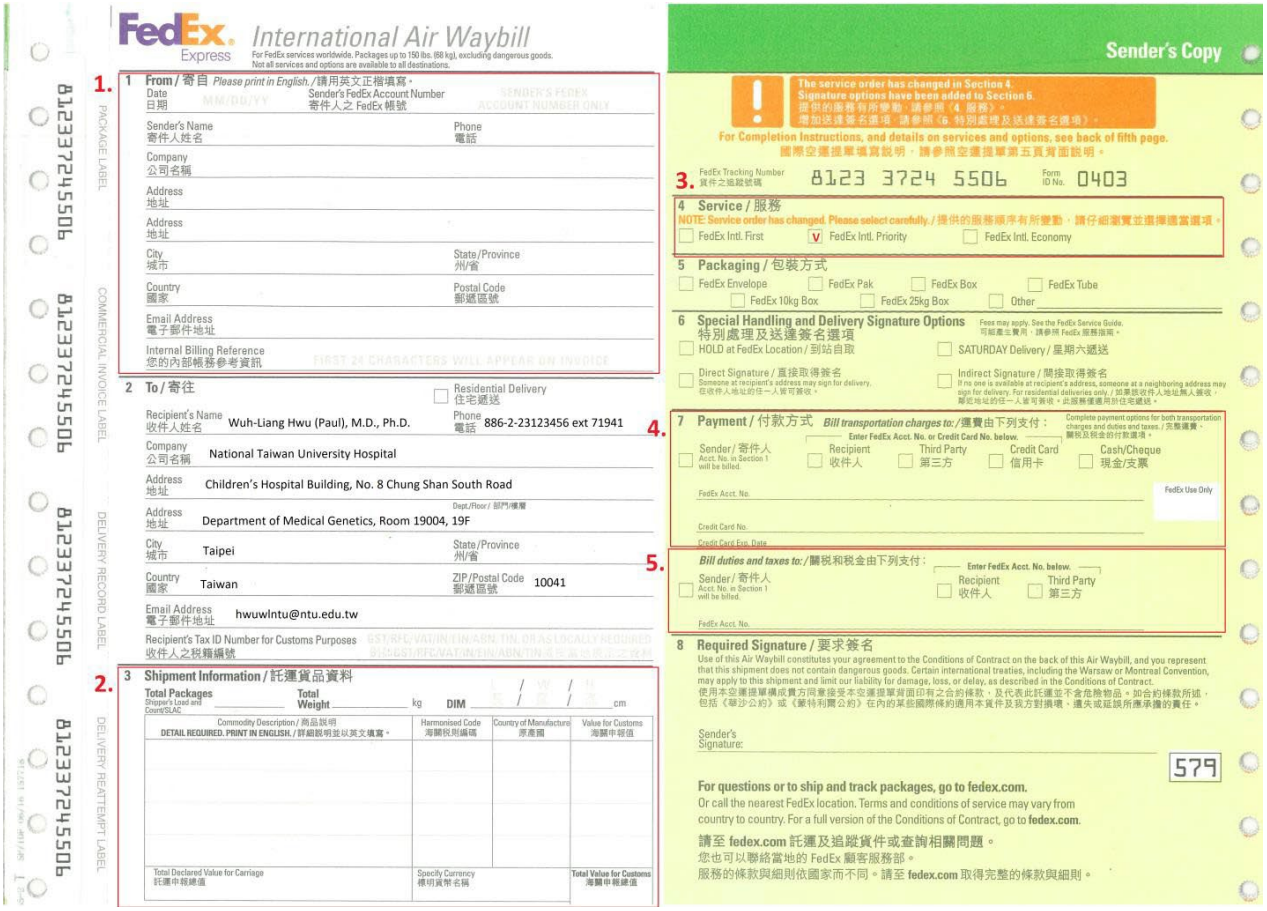
MARKS/ NOS.	NO. OF PKGS	TYPE OF PACKAGING	FULL DESCRIPTION OF GOODS	QTY	W	Unit of Measure	UNIT VALUE	TOTAL VALUE
	__1___	Urine blood spot Urine spot	Human urine Dried blood on paper (Human) Dried urine on paper (Human)	____ ____ ____		tube Sheet Sheet	US$ 1.00 US$ 1.00 US$ 1.00	US$_ US$ US$_____
	TOTAL NO. OF PKGS.					TOTAL WEIGHT		TOTAL INVOICE VALUE
	___1__					_____		US$_____

THESE COMMODITIES ARE LICENSED FOR THE ULTIMATE DESTINATION SHOWN.
DIVERSION CONTRARY TO TAIWAN, R.O.C. LAW IS PROHIBITED

I DECLARE ALL THE INFORMATION CONTAINED IN THIS INVOICE TO BE TRUE AND CORRECT

SIGNATURE OF SHIPPER/EXPORTER _____ DATE _____

Guide to fill in Air Waybill



1. **From :** Your information
2. **Shipment information :** Please fill in the sample type, QTY and the VALUE (Like invoice)
3. **Service :** Please check "FedEx Intl. Priority"
4. **Payment :** Please choose "Sender" or "Third Party"
5. **Bill duties and tax to :** Please choose "Sender" or "Third Party", if you forget to fill in this item, it may cause import delays.
   - Please remember to fill in the Fedex account number.

Test Request Form

Patient Initials:_____ **Hospital Chart number:** _____

Gender: _____ **Birth date:** _____ **(dd/mmm/yyyy)**

Sample collection date: _____ (e.g., 01/Jan/2018)

Physician Name: _____ Email Address: _____

Hospital Name: _____

NTUH Account code:_____

By enrolling in this program, I warrant I will neither seek nor accept payment (directly or indirectly) from a third party for the testing or related activities funded by Sanofi except that such other payment does not overlap the costs, expenses or efforts funded by Sanofi.

Clinical manifestations

- ☐ Bone dysplasia
- ☐ Bone fractures and thin cortex
- ☐ Joint contracture
- ☐ Mild hepatosplenomegaly
- ☐ Huge hepatosplenomegaly
- ☐ Muscle weakness
- ☐ Cardiomyopathy
- ☐ Facial dysmorphism
- ☐ Mental retardation
- ☐ Renal failure
- ☐ Pain
- ☐ Others:_____

Test list:

Fabry Disease

☐ Fabry Disease Enzyme Activity Test (DBS) – test code 000X0177

☐ Fabry Disease Lyso-Gb3 Test (DBS) – test code 000X0179

☐ *Fabry Disease GLA genetic test (DBS) – test code 000X0126

*Tick on this box indicates the agreement of automatic genetic analysis after an abnormal enzyme / biomarker test result, genetic test will not be performed if enzyme activity test or biomarker test is normal.

Pompe Disease

☐ Pompe Disease Enzyme Activity Test (DBS) – test code 000X0176

☐ Pompe Disease Glc4 Test (urine) – test code 000X0178

☐ *Pompe Disease GAA genetic test (DBS) – test code000X0126

*Tick on this box indicates the agreement of automatic genetic analysis after an abnormal enzyme / biomarker test result, genetic test will not be performed if enzyme activity test or biomarker test is normal.

Gaucher Disease

☐ Gaucher Disease Enzyme Activity Test (DBS) – test code 000X0183

☐ Gaucher Disease Lyso-GL1 Test (DBS) – test code 000X0179

☐ *Gaucher Disease GBA genetic test (DBS) – test code 000X0126

*Tick on this box indicates the agreement of automatic genetic analysis after an abnormal enzyme / biomarker test result, genetic test will not be performed if enzyme activity test or biomarker test is normal.

MPS I&II

☐ MPSI Enzyme Activity Test (DBS) – test code 000X0186

☐ *MPSI IDUA genetic test (DBS) – test code 000X0126

☐ MPSII Enzyme Activity Test (DBS) – test code 000X0187

☐ *MPSII IDS genetic test (DBS) – test code 000X0126

☐ Urinary GAGs analysis (DUS) – test code 000X0188

*Tick on this box indicates the agreement of automatic genetic analysis after an abnormal enzyme / biomarker test result, genetic test will not be performed if enzyme activity test or biomarker test is normal

Niemann Pick Type A/B Disease

☐ Niemann-Pick Type A/B Disease Enzyme Activity Test (DBS) – test code 000X0183

☐ Niemann-Pick Type A/BDisease Lyso-SM Test (DBS) – test code 000X0179

☐ *Niemann-Pick Type A/B Disease genetic test (DBS) – test code 000X0243

*Tick on this box indicates the agreement of automatic genetic analysis after an abnormal enzyme / biomarker test result, genetic test will not be performed if enzyme activity test or biomarker test is normal