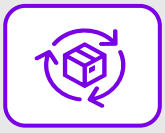
One Rare HCPs Guide

One Rare HCPs Guide is the tool to support DBS kits, samples pick-up request and others.

Step 1 Order via accessing the online form



Step 2 DBS kits order process

Step 3 How to fill test request form

Step 4 How to collect DBS samples

Step 5 Pick-up sample via FedEx

Step 6 How to follow-up status/report?

Step 7 Any Q&A

For Healthcare Professionals Only

MAT-SG-2600188-4.0-08/2026

sanofi

Step 1 Order via accessing the online form

One Rare Diagnostic Kits Request Form ← *Click here to access the form*

One Rare Diagnosis Request Form (Singapore)

This form supports Sanofi's One Rare Diagnosis Program for rare disease diagnosis in Singapore. For detailed process guidance, refer to the program manual here. Please ensure all NTUH forms are downloaded and completed, here.

When you submit this form, it will not automatically collect your details like name and email address unless you provide it yourself.

Which diseases are you planning to send samples for? *

- [ ] Pompe
- [ ] Fabry
- [ ] MPS I/II
- [ ] ASMD
- [ ] Gaucher

Full Name (First Name, Last Name) *

Enter your answer

Email Address *

Enter your answer

Step 1 Order via access online form

One Rare Diagnostic Kits Request Form ← *Click here to access the form*

Contact Number *

Enter your answer

Which Health Care Professional (HCP) are you? *

- ◯ Physician
- ◯ Nurse
- ◯ Technician
- ◯ Genetic Counsellor
- ◯ Pharmacists

Hospital/Institute Name *

If "Other" was chosen, Delivery Address (Please specify building, department, and floor) *

Enter your answer

Delivery Remarks (Building Name, Department, Floor, Attn Name) *

Enter your answer

Step 1 Order via access online form

One Rare Diagnostic Kits Request Form ← *Click here to access the form*

No. of DBS Kits (DBS 1 kit contains 10 cards, min. 1 kit required per delivery)

- Requests received before 3:00 PM will be delivered within one (1) Business Day

- Requests received after 3:00 PM will be processed and delivered within the next Business Day*

- ◯ 1 kit (10 cards)
- ◯ 2 kits (20 cards)
- ◯ 3 kits (30 cards)
- ◯ 4 kits (40 cards)
- ◯ 5 kits (50 cards)

Please note when planning timeline:

Requests received before 3:00 PM will be delivered within one (1) Business Day

Requests received after 3:00 PM will be processed and delivered within the next Business Day

☐ I confirm I will attach the required NTUH forms when submitting samples to NTUH.

☐ I confirm that I have read and understood the Personal Data Protection Notice, and I consent to the collection, use and disclosure of my personal data in the manner as described in the Personal Data Protection Notice

☐ I have read the Terms and Conditions and Sanofi Privacy Policy, understand their terms, and I agree and intend to be legally bound by them. I understand that if I do not accept such Terms and Conditions and Sanofi Privacy Policy, Sanofi is unable to grant me access to the Platform and provide me with related service

☐ I confirm that I am a licensed healthcare professional under the laws of Singapore

Submit ← ***Tick all boxes and fill all required information to submit***

Step 2.1 DBS kits order process

1. DBS kit order will be done via the One Rare Diagnosis Request Form.
2. DBS kits are ordered in packs of 10, with a minimum order of 1 kit (10 DBS cards).
3. Designate a contact person's details (Name, Phone number, and email) if not the same as HCP's

DBS kit orders before 3PM, will be delivered within 1 business day whereas orders after 3PM, will be processed and delivered next business day

Please plan accordingly

DBS kit

Each kit consists of

- ***10 DBS cards***
- ***10 zip lock bags***
- ***10 desiccants***
- ***Instructions for use***

Step 2.2 Assembling your DBS card(s) for sampling and shipment

Pack each DBS card **individually**

(1 DBS card + 1 Desiccant inside 1 zip lock bag)

1 Assembled DBS card

Step 3 How to Fill Test Request Form

Step 3.1 Fill and email request form on NTUH Lab Testing Service

•Ensure to include the request form when sending to NTUH to ensure correct test

•Email NTUH to inform shipping date

•Notice: Limit on Fabry disease DBS tests *(α-galactosidase A, Lyso-Gb3, GLA genetic)*

•Unlimited tests remain available for Pompe, Gaucher, MPS1, MPS2, and ASMD.

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.

See appendix for full-sized form

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

Once both forms have been filled and signed,

Send form to:

Miss Min Hui: (minhui@ntuh@gov.tw)

Step 3 How to Fill Test Request Form

Step 3.2 Obtain informed consent from patients and physicians

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

- Fill consent form
- Send to Miss Min Hui (minhui@ntuh@gov.tw) alongside the request form

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

See appendix for full-sized forms

Once both forms have been filled and signed,

Send form to:

Miss Min Hui: (minhui@ntuh@gov.tw)

Step 4.1 How to collect DBS samples?

Step 4.1 DBS Sample collection and storage (1/2)

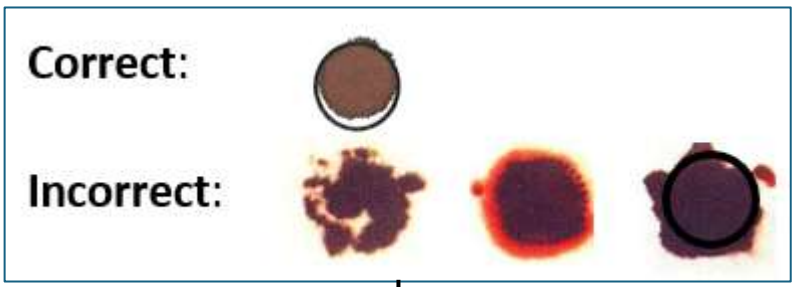
Please ensure **ALL information on the DBS card is filled before collection**
Blood collection on DBS Filter Paper Card can be done with 2 methods:

A. Patient < 6 months

1. Warm the heel site
2. Cleanse with disinfectant and dry
3. Puncture heel with safe lancet
4. Wipe away first drop with sterile gauze
5. Allow large hanging drop to form
6. Lightly touch drop to Centre of Circle 1
7. Let blood soak through and fill completely
8. Repeat for remaining 3 circles

B. Patient ≥ 6 months

1. Collect venous blood in collection tube
2. Invert tube 5 times to mix (no vortex mixer)
3. Set pipettor to 60 µL Hold tip above circle centre (close but not touching)
4. Gradually discharge blood to fill circle (single application)
5. Repeat for remaining 3 circles



- ✓ Apply only 1 drop of blood per circle on the DBS card.
- ✓ Be sure that the blood covers the whole circle and spreads on both sides of the filter paper.

Dry for 4 hours at room temperature

1. Avoid sunshine, do NOT heat
2. Filter can be dried overnight in an air conditioned room

Once dry, cover sample circles with paper flap
Seal in plastic bag with desiccant

✓ See next page for further storage guideline

General Guidelines:

- To perform the test only 4 blood drops are needed. This test can be done along with other standard blood tests. Capillary or venous blood can be used (EDTA or ACD anti-coagulant may be used, heparin should be avoided, if possible).
- For patients on ***dialysis*** its important to take the sample **before dialysis**. Blood taken during or directly after dialysis cannot be used for testing.
- **Do not use iodine containing disinfectants.**

Step 4.1 How to collect DBS samples?

Step 4.1 DBS Sample collection and storage (2/2)

DBS Sample storage guideline

Short term

Multiple studies demonstrate DBS samples can be stored at ambient temperature for prolonged periods with minimal enzyme activity losses

- **Recommended shipping timeline:**
  Ship samples to the lab as soon as possible
  Target: samples should arrive at lab within 1-2 weeks after collection

- **Critical note for Fabry disease biomarker (Lyso-Gb3/Lyso-GL-3):**
  May degrade after two weeks of storage
  Particularly important for women suspected of having Fabry disease
  Women typically present with low baseline levels of Lyso-Gb3/Lyso-GL-3
  Longer storage may result in false-negative results in women

Long term

- **Storage conditions:**
  DBS samples are stable for 6 months at -20°C
  Must be stored under dry conditions

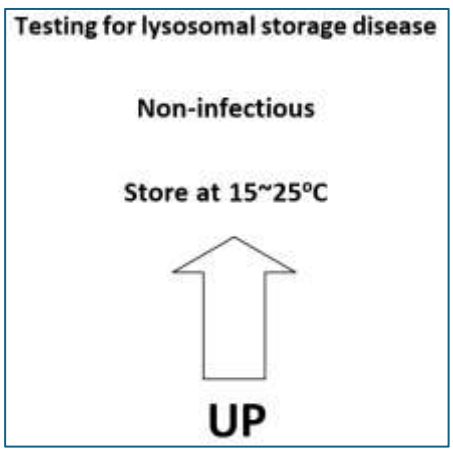
- **Storage procedure:**
  Freeze DBS samples in a plastic zip bag
  Include silica bag (desiccant) to protect against moisture
  Plastic bags and desiccants are included when receiving DBS kits
  Desiccant bags can be reused for storage

- **Important restriction:**
  Never store DBS samples at 4°C

Step 5 Pick-up sample with FedEx (1/2)

Sample pick-up via courier (FedEx)

1. Copy the Invoice form and the Air Waybill and **email them to NTUH**
2. Place any samples to be tested in a FedEx Pak including
   Test request form (printed from e-platform)
   Inform consent form
   Invoice form
3. Can be sent at room temperature, but must avoid heating of the samples
4. Send by **International Priority**



Label for package

See appendix for full-sized image

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

See appendix for full-sized image

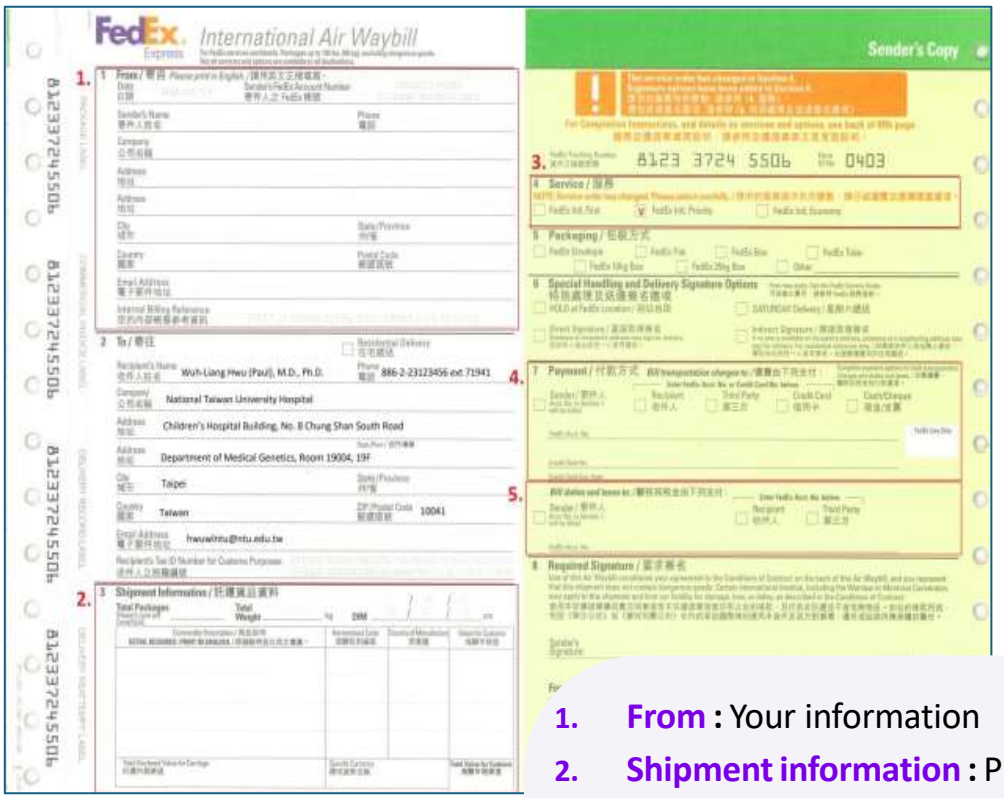
Delivery Exceptions

DBS samples should be shipped out as soon as possible, so **it arrives at the lab within 1-2 weeks after collection.**

Hence, take note of any delivery exceptions. For example, customs might hold the parcel for inspection (usually not rejected).

Step 5 Pick-up sample with FedEx (2/2)

FedEx Air Waybill can either be online or physical



Label for package
See appendix for full-sized image

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.

Copy this Air Waybill and the Invoice Form and **email them to NTUH**

Step 6 How to follow-up status/report

Email notification

If you had checked the box for **FedEx** email notifications, you would be sent email updates.
However, depending on the location of pickup, the email notification to inform you that the pickup has been successful may be sent by the following day once the sample has arrived.

Hence, do note that email notifications may be delayed and not as accurate.

FedEx Customer Service

Any urgent enquiries regarding your shipment can also call 1800-88-6363

Step 7 Any Q&A

For more information regarding DBS sample processing with NTUH:
E-Mail Miss Min Hui: minhui@ntuh.gov.tw

For NTUH form template download:
Click here

See next section for appendix
Thank you

Appendix A: 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

Appendix B: Informed Consent Form (For customs clearance)

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

Appendix C: NTUH Test Request Form

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.

Appendix C: NTUH Test Request Form

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

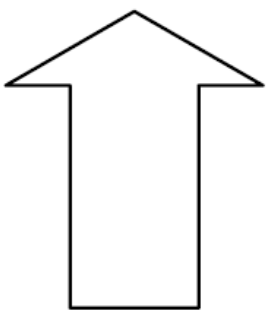
*Tick on this box indicates the agreement of automatic genetic analysis after an abnormal enzyme / biomarker test result, genetic test will not be performed if enzyme activity test or biomarker test is normal

Appendix D: Label of Package

Testing for lysosomal storage disease

Non-infectious

Store at 15~25°C



UP

Appendix E: Invoice Form (Please fill in 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

Appendix F: 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.