

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

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: _____

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

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 code 000X0126

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

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

Niemann Pick Type A/B Disease

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