RDGenePP

Rare Disease Gene Panel Program

Sample collection
and submission guidelines

Paper Submission only for remaining kits distributed before April 2024.

Content of the kits received:

1. Collection form

SERVIÇOS DE
TESTE DE DIAGNÓSTICO
DIAGNOSTIC TESTING SERVICES / SERVICIOS DE PRUEBAS DE DIAGNÓSTICO

Leia atentamente as instruções de coleta contidas neste formulário.
Read carefully the collection instructions described in this form.
Lea atentamente las instrucciones de recolección contenidas en este formulario.

Sem valor comercial. / No commercial value. / Sin valor comercial.

000000000000

LOT CP003.7043143.20.08 31/12/2039

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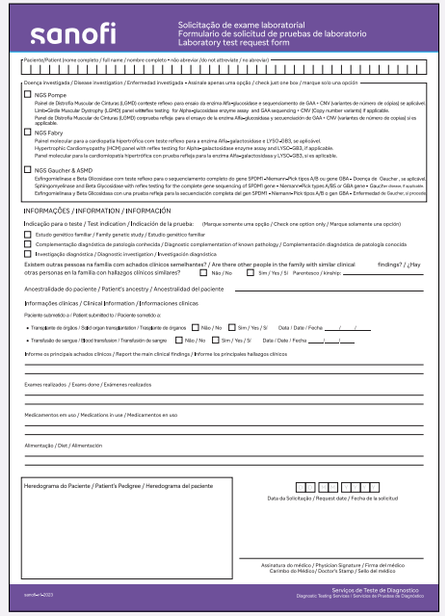
2. Test request form



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Solicitação de exame laboratorial
Formulario de solicitud de pruebas de laboratorio
Laboratory test request form

Paciente/Patient (nome completo / full name / nombre completo • não abreviar /do not attreviate / no abreviar)

Doença investigada / Disease investigation / Enfermedad investigada • Assinale apenas uma opção / check just one box / marque solo una opción

☐ NGS Pompe
Painel de Distrofia Muscular de Cinturas (LGMD) contendo reflexo para ensaio da enzima Alfa-glicosidase e sequenciamento de GAA + CNV (variantes de número de cópia) se aplicável.
Limb-Girdle Muscular Dystrophy (LGMD) panel withreflex testing for Alpha-glucosidase enzyme assay and GAA sequencing + CNV (Copy number variants) if applicable.
Panel de Distrofia Muscular de Cinturas (LGMD) conprueba reflejo para el ensayo de la enzima Alfa-glucosidasa y secuenciación de GAA + CNV (variantes de número de copias) si es applicable.

☐ NGS Fabry
Painel molecular para a cardiopatia hipertrófica com teste reflexo para a enzima Alfa-galactosidase e LYSO-Gb3, se aplicável.
Hypertrophic Cardiomyopathy (HCM) panel with reflex testing for Alpha-galactosidase enzyme assay and LYSO-GB3, if applicable.
Panel molecular para la cardiomiopatía hipertrófica con prueba reflejo para la enzima Alfa-galactosidasa y LYSO-GB3, si es aplicable.

☐ NGS Gaucher & ASMD
Esfingomielinase e Beta Glicosidase com teste reflexo para o sequenciamento completo do gene SPDM1 •Niemann•Pick tipos A/B ou gene GBA • Doença de Gaucher , se aplicável
Sphingomyelinase and Beta Glycosidase with reflex testing for the complete gene sequencing of SPDM1 gene • Niemann•Pick types A/B) or GBA gene • Gaucher disease, if applicable
Esfingomielinasa y Beta Glicosidasa con una prueba reflejo para la secuenciación completa del gen SPDM1 •Niemann•Pick tipos A/B o gen GBA • Enfermedad de Gaucher, si procede

INFORMAÇÕES / INFORMATION / INFORMACIÓN

Indicação para o teste / Test indication / Indicación de la prueba: (Marque somente uma opção / Check one option only / Marque solamente una opción)
☐ Estudo genético familiar / Family genetic study / Estudio genético familiar
☐ Complementação diagnóstica de patologia conhecida / Diagnostic complementation of known pathology / Complementación diagnóstica de patología conocida
☐ Investigação diagnóstica / Diagnostic investigation / Investigación diagnóstica

Existem outras pessoas na família com achados clínicos semelhantes? / Are there other people in the family with similar clinical findings? / ¿Hay otras personas en la familia con hallazgos clínicos similares? ☐ Não / No ☐ Sim / Yes / Sí Parentesco / kinship: _____

Ancestralidade do paciente / Patient's ancestry / Ancestralidad del paciente _____

Informações clínicas / Clinical information / Informaciones clínicas

Paciente submetido a / Patient submitted to / Paciente sometido a:
• Transplante de órgãos / Solid organ transplantation / Trasplante de órganos ☐ Não / No ☐ Sim / Yes / Sí Data / Date / Fecha ___/___/___
• Transfusão de sangue / Blood transfusion / Transfusión de sangre ☐ Não / No ☐ Sim / Yes / Sí Data / Date / Fecha ___/___/___

Informe os principais achados clínicos / Report the main clinical findings / Informe los principales hallazgos clínicos

Exames realizados / Exams done / Exámenes realizados

Medicamentos em uso / Medications in use / Medicamentos en uso

Alimentação / Diet / Alimentación

Heredograma do Paciente / Patient's Pedigree / Heredograma del paciente

☐☐/☐☐/☐☐☐☐
Data da Solicitação / Request date / Fecha de la solicitud

Assinatura do médico / Physician Signature / Firma del médico
Carimbo do Médico / Doctor's Stamp / Sello del médico

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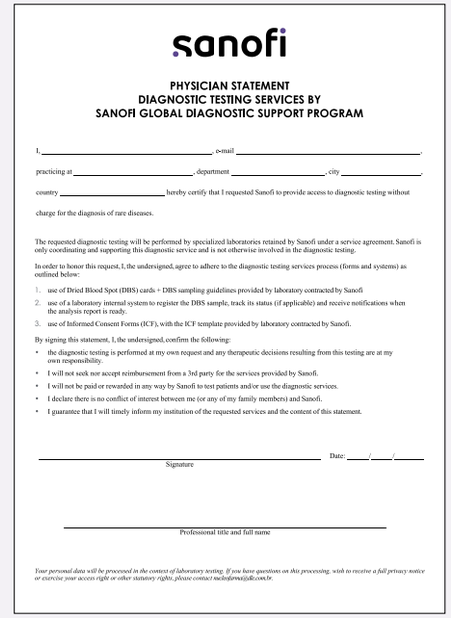
Serviços de Teste de Diagnostico
Diagnostic Testing Services / Servicios de Pruebas de Diagnóstico

3. Physician's statement



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PHYSICIAN STATEMENT
DIAGNOSTIC TESTING SERVICES BY
SANOFI GLOBAL DIAGNOSTIC SUPPORT PROGRAM

I, _____, e-mail _____,

practicing at _____, department _____, city _____,

country _____ hereby certify that I requested Sanofi to provide access to diagnostic testing without charge for the diagnosis of rare diseases.

The requested diagnostic testing will be performed by specialized laboratories retained by Sanofi under a service agreement. Sanofi is only coordinating and supporting this diagnostic service and is not otherwise involved in the diagnostic testing.

In order to honor this request, I, the undersigned, agree to adhere to the diagnostic testing services process (forms and systems) as outlined below:

1. use of Dried Blood Spot (DBS) cards + DBS sampling guidelines provided by laboratory contracted by Sanofi
2. use of a laboratory internal system to register the DBS sample, track its status (if applicable) and receive notifications when the analysis report is ready.
3. use of Informed Consent Forms (ICF), with the ICF template provided by laboratory contracted by Sanofi.

By signing this statement, I, the undersigned, confirm the following:

- the diagnostic testing is performed at my own request and any therapeutic decisions resulting from this testing are at my own responsibility.
- I will not seek nor accept reimbursement from a 3rd party for the services provided by Sanofi.
- I will not be paid or rewarded in any way by Sanofi to test patients and/or use the diagnostic services.
- I declare there is no conflict of interest between me (or any of my family members) and Sanofi.
- I guarantee that I will timely inform my institution of the requested services and the content of this statement.

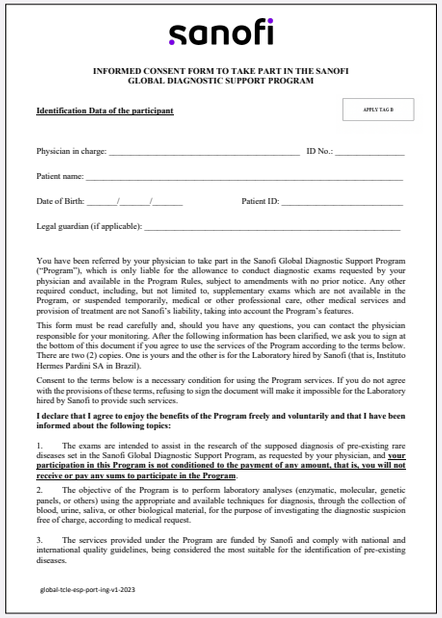
_____ Date: ___/___/___
Signature

_____
Professional title and full name

Your personal data will be processed in the context of laboratory testing. If you have questions on this processing, wish to receive a full privacy notice or exercise your access right or other statutory rights, please contact nucleoforma@dfe.com.br.

Content of the kits received:

4. ICF (Consent term) – 2 copies



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INFORMED CONSENT FORM TO TAKE PART IN THE SANOFI GLOBAL DIAGNOSTIC SUPPORT PROGRAM

Identification Data of the participant

APPLY TAG B

Physician in charge: _____ ID No.: _____

Patient name: _____

Date of Birth: _____/_____/_____ Patient ID: _____

Legal guardian (if applicable): _____

You have been referred by your physician to take part in the Sanofi Global Diagnostic Support Program ("Program"), which is only liable for the allowance to conduct diagnostic exams requested by your physician and available in the Program Rules, subject to amendments with no prior notice. Any other required conduct, including, but not limited to, supplementary exams which are not available in the Program, or suspended temporarily, medical or other professional care, other medical services and provision of treatment are not Sanofi's liability, taking into account the Program's features.

This form must be read carefully and, should you have any questions, you can contact the physician responsible for your monitoring. After the following information has been clarified, we ask you to sign at the bottom of this document if you agree to use the services of the Program according to the terms below. There are two (2) copies. One is yours and the other is for the Laboratory hired by Sanofi (that is, Instituto Hermes Pardini SA in Brazil).

Consent to the terms below is a necessary condition for using the Program services. If you do not agree with the provisions of these terms, refusing to sign the document will make it impossible for the Laboratory hired by Sanofi to provide such services.

I declare that I agree to enjoy the benefits of the Program freely and voluntarily and that I have been informed about the following topics:

1. The exams are intended to assist in the research of the supposed diagnosis of pre-existing rare diseases set in the Sanofi Global Diagnostic Support Program, as requested by your physician, and **your participation in this Program is not conditioned to the payment of any amount, that is, you will not receive or pay any sums to participate in the Program**.

2. The objective of the Program is to perform laboratory analyses (enzymatic, molecular, genetic panels, or others) using the appropriate and available techniques for diagnosis, through the collection of blood, urine, saliva, or other biological material, for the purpose of investigating the diagnostic suspicion free of charge, according to medical request.

3. The services provided under the Program are funded by Sanofi and comply with national and international quality guidelines, being considered the most suitable for the identification of pre-existing diseases.

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5. Plastic envelope for return

6. Silica gel bag

7. External envelope

Step 1:
Fill all the blanks in the forms within the kit.

Form 1:
Collection Form

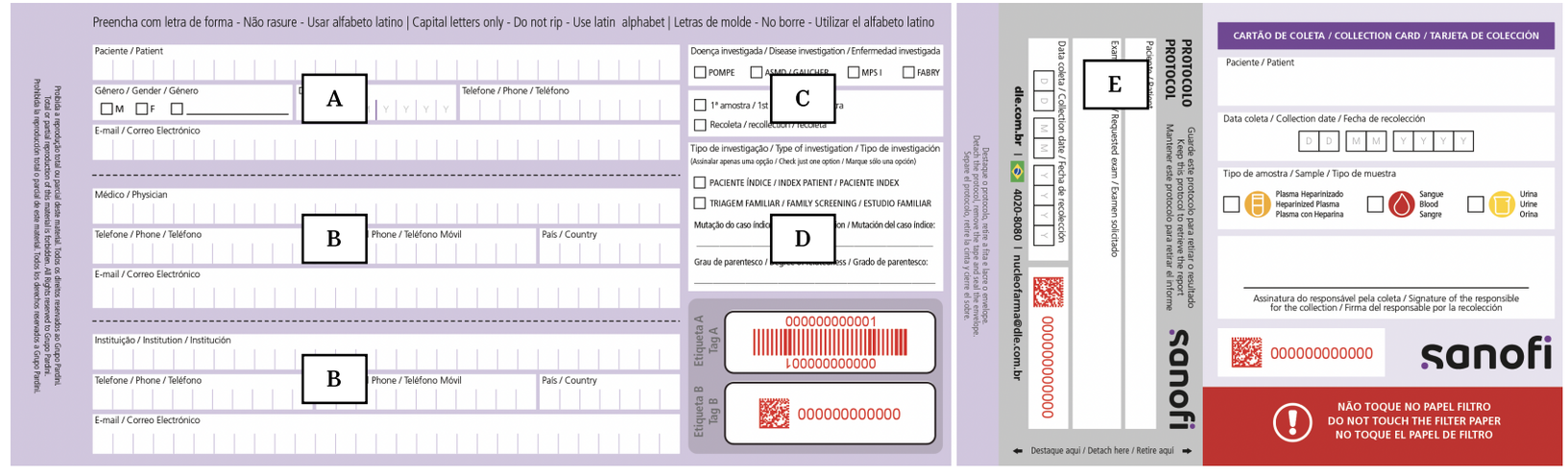
A) Patient information
Use capital letters. Do not abbreviate.

B) Clinic/Physician Information
Important: Be careful in writing the e-mail with readable letter to ensure receipt of results.

C) Select Test.

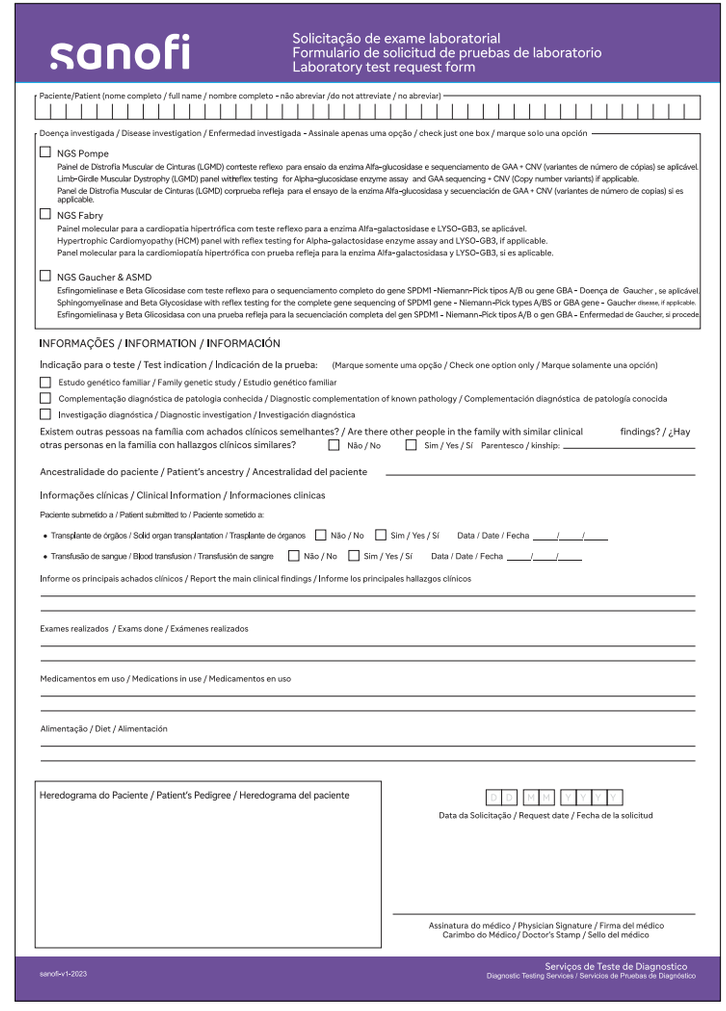
D) Inform Sample Collect Sample.

E) Protocol
The physician should keep the protocol to guarantee the traceability of sample via barcode.



Rare Disease Gene Panel Program

Form 2:
Test Request Form



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D D M M A A A A
Data da Solicitação / Request date / Fecha de la solicitud

Assinatura do médico / Physician Signature / Firma del médico
Carimbo do Médico / Doctor's Stamp / Sello del médico

sanofi-v1-2023

Serviços de Teste de Diagnostico
Diagnostic Testing Services / Servicios de Pruebas de Diagnóstico

Fill with Patient Name

Select Requested Exam and Test Indication

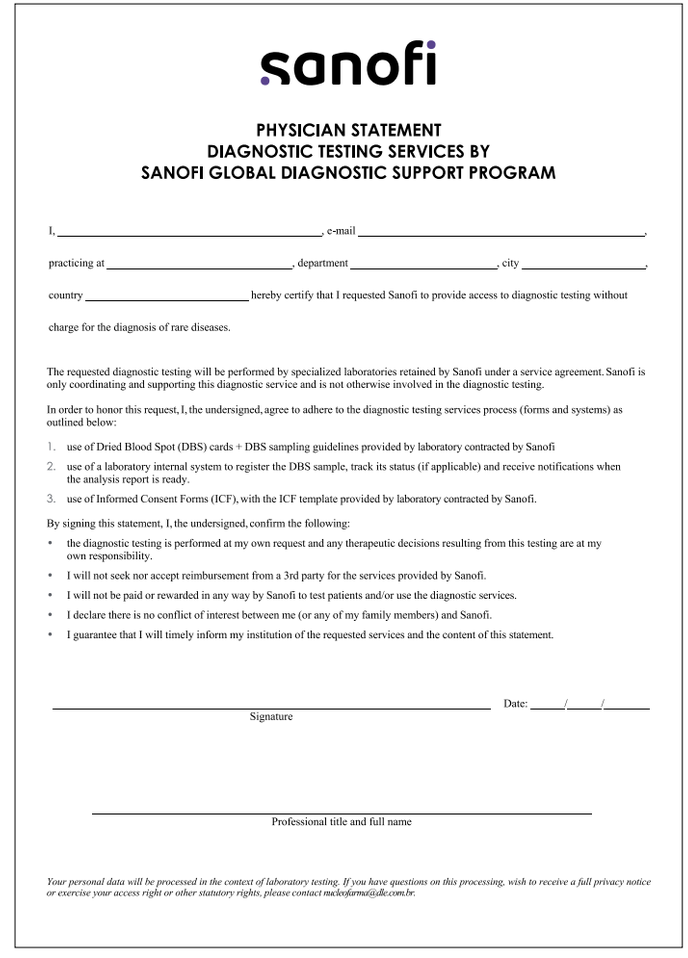
Fill with Clinical Information

Physician's signature

Form 3:
Physician's Statement

Fill with Physician's Information

Physician's signature



sanofi

PHYSICIAN STATEMENT
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- I declare there is no conflict of interest between me (or any of my family members) and Sanofi.
- I guarantee that I will timely inform my institution of the requested services and the content of this statement.

_____ Date: _____/_____/_____
Signature

_____
Professional title and full name

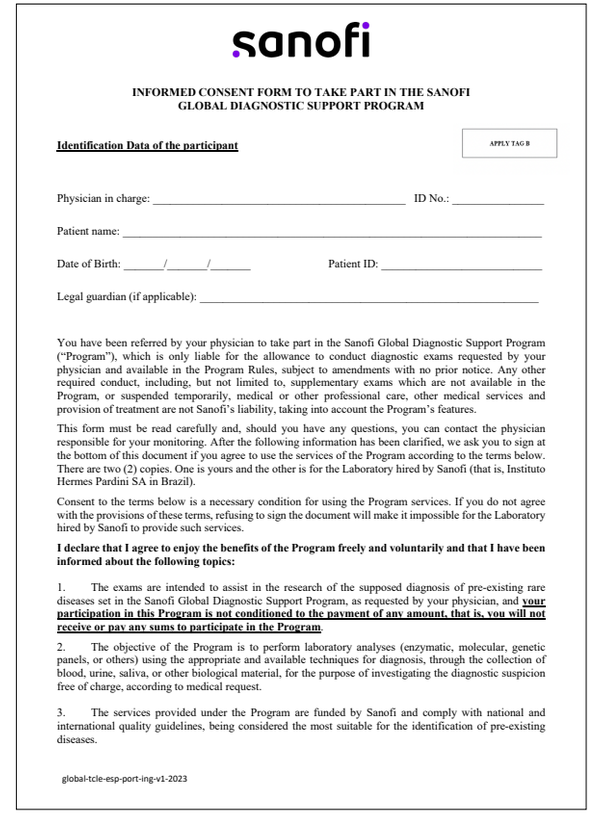
Your personal data will be processed in the context of laboratory testing. If you have questions on this processing, wish to receive a full privacy notice or exercise your access right or other statutory rights, please contact nucleofarma@dle.com.br.

Fill with Physician's Name and Professional Title

Rare Disease Gene Panel Program

Form 4:
Informed Consent Form (ICF)

Fill with Patient Information



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INFORMED CONSENT FORM TO TAKE PART IN THE SANOFI GLOBAL DIAGNOSTIC SUPPORT PROGRAM

Identification Data of the participant

APPLY TAG B

Physician in charge: _____ ID No.: _____

Patient name: _____

Date of Birth: _____/_____/_____ Patient ID: _____

Legal guardian (if applicable): _____

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← Apply Tag A

⚠ Patient or guardian signature on last page. **(mandatory)**

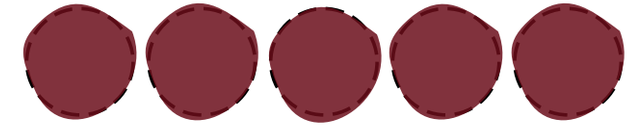
⚠ 2 copies (one copy must be attached to the test request form and the other to be given to the patient.

Step 2:
Sample Collection Process.

RDGenePP
Rare Disease Gene Panel Program

Step 2:
Tips for Successful Sample Collection

Successful sample



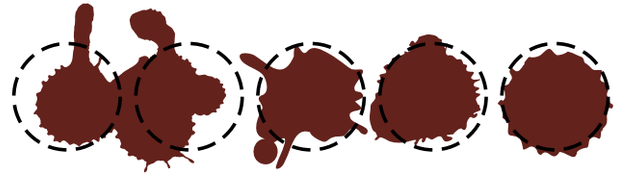
- Sufficient and homogeneous concentration of sample.
- Blood should be viewed in both sides of the filter.
- Sample could be drawed with needle or by capillary puncture.
- Important to fill all the paper filter circle with the blood.
- If the sample doesn’t meet the quality standard, a new sample collection will be asked.

Step 2:
Wrong Collection

Insufficient sample

Supersaturated sample

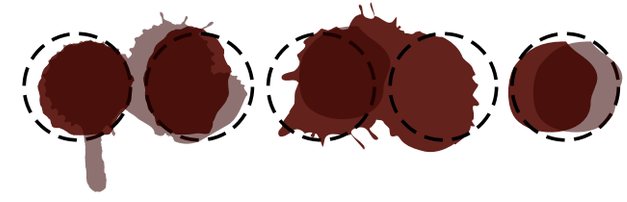
Overlapped sample



Diluted sample

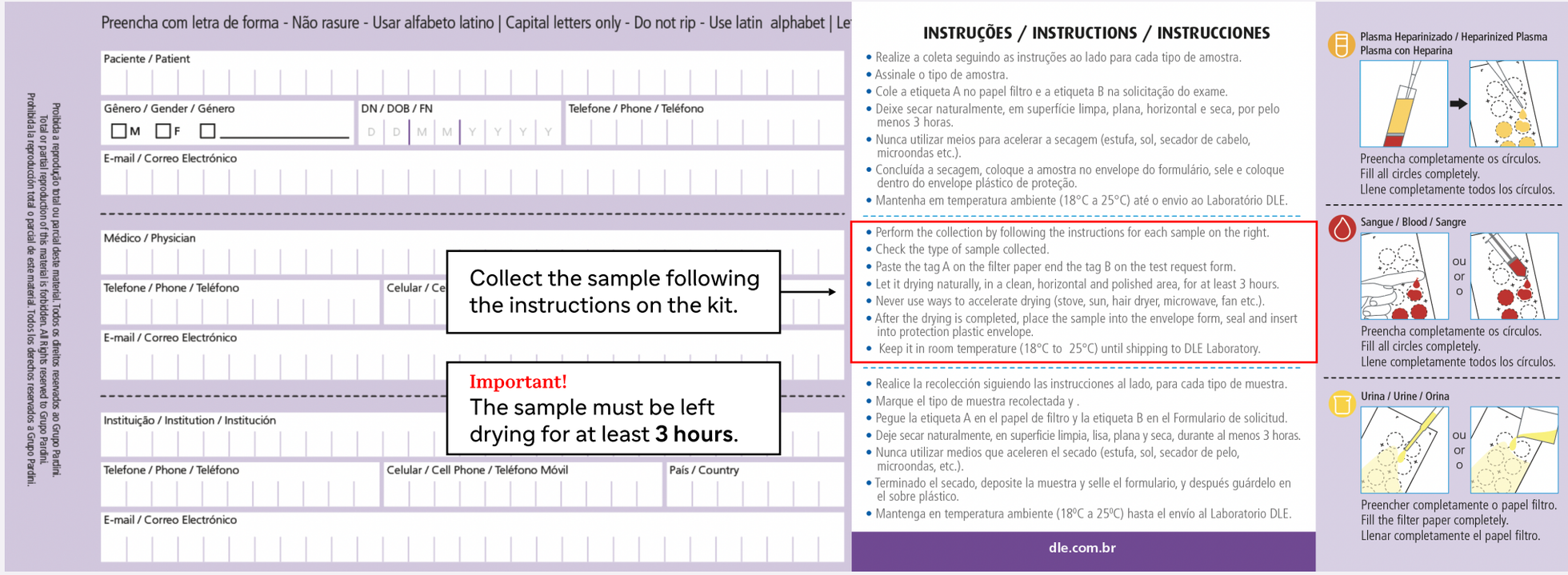
Wet sample

Coagulated sample



Rare Disease Gene Panel Program

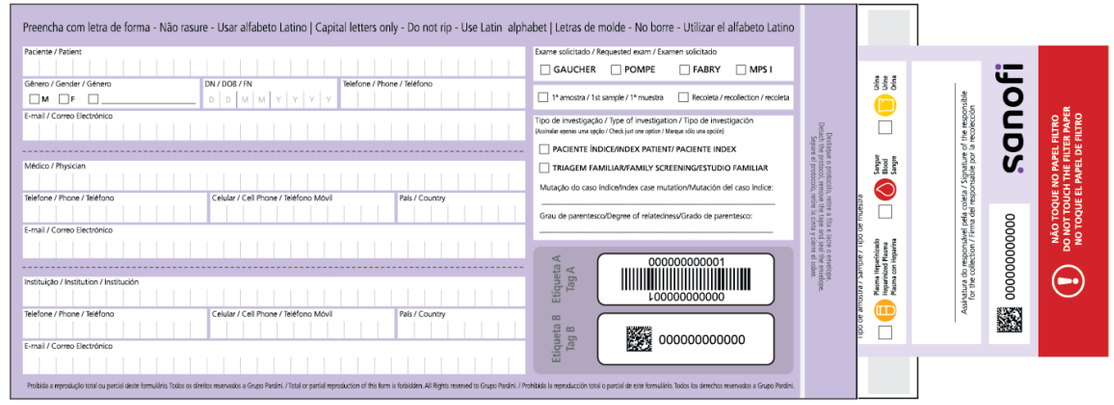
Step 2:
Additional Information



Step 3:
Sample Packaging and Submission.

Rare Disease Gene Panel Program

Step 3:
Correct placement



Place the dried blood spot card into the form envelope and seal it.

Insert the form envelope inside the plastic bag with silica gel and seal it.

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Rare Disease Gene Panel Program

MAT-GLB-2401586- v4.0 - 05/2024