IPA/ Erasmus MC Pompe Survey

Information for patients with Pompe disease, 16 years or older

Dear Sir/Madam:

You have been asked to participate in the IPA/ Erasmus MC Pompe Survey, due to the fact that you have Pompe disease.

This written information is provided to you to help you decide whether or not you want to participate in the IPA/ Erasmus MC Pompe Survey. You may want to take your time reviewing this information and discuss it with your family. If you have any questions, at any time you may ask the investigators who are listed at the conclusion of this information letter.

The IPA/ Erasmus MC Pompe Survey (for convenience's sake, hereafter called the Pompe Survey) is a joint international study program of the International Pompe Association (IPA) and the Erasmus MC in Rotterdam. The IPA is a worldwide organization for various patient organizations for people with Pompe disease. The Erasmus MC is a university hospital that has performed studies on Pompe disease for more than 40 years.

The Pompe Survey is the continuation of a similar study, conducted since 2002 by the IPA and the Erasmus MC, called 'Investigation into the Clinical Condition of Juvenile and Adult (late onset) Pompe Patients.'

In this letter, we will explain what the Pompe Survey includes, and ask for permission for your participation in this study.

1. Aim of the Pompe Survey

The Pompe Survey has been designed to gather data about Pompe disease directly from the patients themselves through questionnaires. In this manner we'll be able to better understand what happens to patients with Pompe disease over time. By doing so, physicians could be assisted in the treatment for these patients. In addition, we want to find out how patients themselves experience the long-term effects of the available treatments and the supporting measures, including enzyme replacement therapy.

2. Design of the Pompe Survey

Who can participate in the Pompe Survey?

All individuals who have been diagnosed with 'Pompe disease' may participate in the study. Our goal is to have as many patients as possible from various countries participate.

What does it mean to participate?

If you decide to participate in this study, at the study's onset you will be asked to fill out an extensive questionnaire regarding your medical history and current physical condition, as well as a number of standardized questionnaires about the disease's effect on your activities and quality of life. After this initial questionnaire, every year you will receive a follow-up questionnaire with the standard questionnaires included.

The questionnaires can be filled out in different languages through a secure internet page. Each year you will receive a personal link to the online survey. Should this not be feasible for you, please inform us so that we can send you a paper version of the survey.

How long does it take to fill in the Survey?

Filling in the questionnaires will take approximately 1 to 1.5 hours of your time. The baseline questionnaire will probably take a little longer.

Linking with clinical data

The Pompe Survey collects the experiences of patients over time. In addition to this, your treating physician will be performing clinical measurements during follow-up visits to assess how you are doing. These clinical data are known in your hospital or by your treating physician, and sometimes also collated and analyzed locally or nationally to study the effects of enzyme replacement therapy.

Linking the survey data with such clinical datasets offers an opportunity to compare the data filled out by the patients themselves with the data collected by the attending physicians. This will give us even more understanding about the disease's course, and help us to assess the effects of the treatment even better.

For that reason, we ask in the attached consent form whether you agree to linking the survey data to your Pompe-specific clinical data. If you do, we can ask hospitals, treating physicians, or national registries to collaborate with us to link and analyze data for a group of patients. If you do not agree to this linkage you can still participate in this survey and you will receive the same level of care.

3. Advantages and Disadvantages of Participation

There are no direct medical benefits in connection with participation in the Pompe Survey. However, if you were to participate in the study, this could help physicians and scientific researchers to learn more about Pompe disease, what happens over time to people who suffer from this disease, and what the effects of available treatment for this disease are. This information may increase our understanding of Pompe disease, and can help physicians in the treatment of persons with the disease.

Additionally, participation in this study does not have any disadvantages and does not carry any risks for you. Your participation only requires the time you need to fill out the questionnaires.

What if you do not want to participate?

Participation in the Pompe Survey is completely voluntary. If you decide against participating, you are not required to state the reason for that decision. If you decide not to participate in the Pompe Survey, this will not in any way affect your continued treatment or support.

4. When does the study end?

The Pompe Survey is intended to last several years. At this time it is not known exactly how long this will be, but the study will in any event last until the end of 2032. However, you may withdraw your participation in the Pompe Survey at any time.

As this study is intended to last many years, we sometimes lose contact with participants, for example because of a change in contact details or a deterioration of health. Therefore we ask you to provide a contact person on the attached consent form. Should we not be able to reach you at your last known contact information we will contact this person to update your contact details and/or your status.

5. What happens after the study?

As a participant in the Pompe Survey, you will be informed from time to time of the results at a group level by the Erasmus MC and/or the International Pompe Association. This will be done in various ways, for instance via the International Pompe Association's website and through presentations at patient meetings. Results of the study will also be presented in scientific magazines and meetings.

6. What do we use your data for?

Which data do we store?

In addition to your answers to the questionnaires we also store your name, sex, date of birth, e-mail (or postal) address, country, and – if applicable – your clinical data obtained through linkage and the name of the hospital or doctor's office where you are treated for Pompe disease.

How do we protect your privacy?

Personal data, gathered in the course of this study, will be replaced with a code number and with initials. Only this code number will be used for analyzing the data, for study documentation and for reports or publications about this study. Reports, presentations and publications about this study will not show that you participated; results are never presented for an individual patient, only for groups of patients.

The only persons who know which code number has been assigned to you are those of the Pompe Survey Study team in the Erasmus MC. Additionally, the Pompe Survey's data may also be published in conjunction with clinical data; this will be on an anonymous basis as well.

Your personal information will be stored in a separate file, independent from the questionnaires, on a secure drive within the Erasmus MC. With your permission, this information will only be used to enable linking the Pompe Survey's dataset with clinical datasets, to send you the annual survey, and if necessary to be able to contact you. In the event that you are contacted, this will only be done by the Pompe Survey Study team in the Erasmus MC, or through your national patient organization or a contact person at the International Pompe Association.

The Pompe Survey has been designed in a way that the confidentiality of the patient information in the Survey's database meets the EU Directives in the area of Privacy and Protection of Patient information.

Who can view your data?

Some people can review your name and other personal data without a code. These are people who check whether the study is carried out correctly and reliably. This can be an auditor working for or hired by the IPA or the Erasmus MC, or national and international supervisory authorities (e.g. the Healthcare and Youth Inspectorate). These persons will keep your data confidential. We ask you to give permission for such review.

The study data will be managed in compliance with European General Data Protection Regulations and Erasmus MC's Privacy Regulation.

How long will we store your data?

With your permission, the data will be stored for a maximum of 15 years after the study's close.

Can you withdraw your consent for the use of your data?

Even if you give your permission at this time, you may withdraw it at any time without stating a reason. However, you'll have to inform us of this in writing (see point 9 for contact details). After that, no new data about you will be entered in our database and you will not receive further questionnaires. If you wish to have any information entered previously deleted from the database as well, you will have to state this clearly in your letter of withdrawal. If you do not state this specifically, we will use your anonymous data for possible publications.

Do you want to know more about your privacy?

Do you want to know more about your rights regarding the processing of personal data? Please see https://www.autoriteitpersoonsgegevens.nl/en

If you have questions about your rights? Or a complaint about how your personal data have been processed? Please contact the research team at the Erasmus MC (contact information in point 9 below).

7. Costs and Compensation

You will not incur any costs nor receive any compensation for your participation in this study.

8. Insurance

The Erasmus MC Medical Ethics Committee has waived the requirement for taking out an insurance policy that would cover any damages caused by the study to the subject. The reason for the waiver is that the Committee is of the opinion that this study and the nature thereof pose no risk to the subject.

9. Do you have further questions?

If you have questions or complaints about the Pompe Survey, please contact the following persons:

Employees of the research team at the Erasmus MC:

Erasmus MC-Sophia, Pompe Survey Team
Dr. Molewaterplein 40; 3015 GD Rotterdam; the Netherlands
E-mail: pompe.survey@erasmusmc.nl

OR

A representative of the IPA in your country:

- Australia: Raymond Saich (president@australianpompe.org.au)
- Canada: Brad Crittenden (brad@pompecanada.com)
- France: Olivier Cavallero (olivier.cavallero@gmail.com)
- Germany: Thomas Schaller (schaller@mpompe.de)
- Italy: Fabio Di Pietro (fabio.dipietro@aiglico.it)
- New Zealand: Allyson Lock (nz.pompe@gmail.com)
- The Netherlands: Margo Peeters (margo.peeters@spierziekten.nl)
- UK: Hülya Apaydin (hulya.apaydin@pompe.uk)
- US: Marsha Zimmerman (marsha.zimmerman@amda-pompe.org)
- All other countries: please contact the Erasmus MC (pompe.survey@erasmusmc.nl)

You may also direct your questions to an independent physician who is not involved with the study, but is very familiar with it:

Dr. H.H. Huidekoper, pediatrician
Erasmus MC – Sophia; Dr Molenwaterplein 40; 3015 GD Rotterdam; the Netherlands
E-mail: h.huidekoper@erasmusmc.nl; Phone: 0031-107038112

For general information about your legal rights regarding the processing of your personal data you can visit the website of the Dutch Data Protection Authority in English. If you have questions or complaints about how your personal data have been processed, please first contact the research team at Erasmus MC. You can also contact the Data Protection Staff at Erasmus MC (0031-107034986) or the data protection authorities.

If you are dissatisfied with the study, you may contact the Erasmus MC's independent Grievance Committee. The Grievance Committee can be reached at telephone no. +31-10-7044108.

10. How do you give consent for this study?

Please take your time to think about this study. Would you like to participate? Please fill in the consent form attached to this information leaflet.

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Informed consent form for patients with Pompe disease, 16 years or older

- I have read the information leaflet. I understand the information. I have had the opportunity to ask additional questions. My questions have been answered well enough. I have had sufficient time to consider my participation.
- I am aware that participation is completely voluntary. I also know that I may withdraw my permission at any time without stating a reason and without this having any effect on the medical care or support that I am receiving.
- I know that some people may review all my data to check that the research is done properly. These people are mentioned in this information leaflet. I grant permission to these people to review my data to check the research.
- I grant the researchers permission to process my data for the goals as described in the information letter.
- I grant permission to store my data for a maximum of 15 years after the study's close.
- I grant permission for the data that I provide to the IPA/ Erasmus MC Pompe Survey to be linked to clinical data collected by my physician:

  ☐ Yes ☐ No

  If yes: I am treated in the following hospital or doctor's office:

- In the event that I am not reachable at my last known contact information: I grant permission for members of the study team and my national patient organization to approach a contact person. To enquire about my contact details and health status.

  ☐ Yes ☐ No

  If yes: contact person name: _____

  If yes: contact e-mail or telephone: _____

I want to participate in this study:

Signature:

Place: _____

Date: _____

My name is (patient): _____