



*The International Pompe Association (IPA) encourages patients to participate in the IPA/Erasmus MC Pompe Survey. The Survey is designed to better understand the progression of the disease and its effect on quality of life. Patient-reported data is gathered through paper and/or online questionnaires that have to be filled in by patients once a year.*

The **IPA/Erasmus MC Pompe Survey** (“the Survey”) is a joint international study of the International Pompe Association (IPA), Erasmus MC and Genzyme Corporation. It is the continuation of a study conducted since 2002 by the IPA and the Erasmus MC: “Investigation into the Clinical Condition of Juvenile and Adult (late onset) Pompe Patients”, also known as the Pompe Questionnaire.

## **Who drives the IPA/Erasmus MC Pompe Survey?**

- The **IPA** is a worldwide federation of patient associations supporting people with Pompe disease.
- The **Erasmus Medical Center** is a university hospital in Rotterdam (NL), having performed studies for more than 30 years on Pompe disease.
- **Genzyme Corporation** is the manufacturer of alglucosidase alfa, a drug for enzyme replacement therapy which has been the official treatment for Pompe disease since 2006, and is registered under the brand name Myozyme®.

## **Goal of the IPA/Erasmus MC Pompe Survey**

The Survey was designed to gather data about Pompe disease directly from patients through questionnaires with the objective to better understand the progression of disease over time. Understanding the disease allows physicians to anticipate the future needs of their patients and provides a baseline against which investigational therapies can be tested. Another goal is to find out how patients themselves experience the long-term effects of the available treatments and the supporting measures, including enzyme replacement therapy with Myozyme®. Further information and results from previous surveys can be found on the website [www.pompecenter.nl](http://www.pompecenter.nl).

## **The Design of the IPA/Erasmus MC Pompe Survey**

Who can participate? – All individuals aged 16 and above, with a confirmed diagnosis of Pompe disease, may participate in the Survey. The goal is to include as many patients as possible from various countries. All patients who participated in the earlier version of the Survey (approximately 300) will be asked to participate again, afterwards other patients will be invited to join. In future it is intended to include patients younger than 16 years of age.



What kind of information is collected? – 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 effect of the disease on your activities and quality of life. After this initial questionnaire, you will receive a follow-up questionnaire with the standardized questionnaires included every year.

You will be asked which language you prefer for filling out the questionnaire, and whether you prefer a paper version or an online version through a secured internet page (preferred) to fill out and return/submit the questionnaire. Currently the Survey is available in Dutch, German, English and French. In 2010 Spanish and Italian translations will become available, and a survey for children under 16 years of age will be developed.

## **Participation**

If you have previously participated in the Survey you will receive an invitation letter with a new informed consent form some time after October 2009, the exact date will depend on your country of residence (Netherlands, United Kingdom, Germany, Australia, USA, Canada). If you are not yet enrolled but you are considering participating, you should receive extensive information through IPA and/or your national patient organization by end of 2009/beginning of 2010 (the countries above plus Spain / Latin America, Italy, France). If your native language is not available you will be welcome to participate through one of the available languages.

## **Data ownership and use of data**

Data resulting from the Survey are administered and held securely by the Erasmus MC. Both IPA, Erasmus MC, and Genzyme are entitled to use the anonymised data either individually or jointly with others, for the evaluation of Pompe disease and its treatment in accordance with the Survey protocol only, after approval of the Pompe Survey Steering Committee. Members of the Committee are Prof. Ans van der Ploeg, Dr. Suyash Prasad, and Thomas Schaller (as of 2009).

## **What is new in this version of the Survey?**

So far the Survey was available as a paper version only which became more and more difficult to handle with the number of participants growing. The online version allows to expand the number of participants and makes it easier to collect consistent information. A review of the Survey over the past years resulted in some questions being changed or removed. As a result, the Survey now is shorter and more relevant. Further advantages of the online version are full language support for Survey and documentation, as well as easier and faster completion of the Survey because only those questions are asked that are relevant to you.



## **Keeping you informed**

As a participant in the Survey, you will be informed regularly of the results on a group level by the Erasmus MC and/or the IPA. This will be done in various ways, for instance through a newsletter, through the website of the International Pompe Association ([www.worldpompe.org](http://www.worldpompe.org)) and through presentations at patient meetings.

## **Cooperation with and linkage to the Pompe Registry**

The Pompe Registry is a study in which clinical data is gathered from patients with Pompe disease through their attending physician. Its goal, for one, is to gather the results of assessments that are already performed for medical reasons. In the Pompe Registry, the natural course of the disease and the effect of enzyme replacement therapy on people with Pompe disease are followed over time.

The Pompe Registry and the IPA/Erasmus MC Pompe Survey pursue the same goal. However, one important difference between these two databases is that for the Pompe Registry the data is obtained via the attending physician, whereas for the Survey, the patients themselves provide the data with regard to the effects of the disease on their everyday lives.

If you want, you can also be enrolled into the Pompe Registry, or perhaps you are already a participant. In the future we hope to link both databases of the IPA/Erasmus MC Pompe Survey and the Pompe Registry together to enable greater insights be made into the disease. If you decide to participate in the Pompe Registry, an informed consent will be asked from you, in a similar way as for the Survey.

More information on the Pompe Registry can be found on the website [www.lsdregistry.net/pomperegistry](http://www.lsdregistry.net/pomperegistry).



## Contact

For questions about the *IPA/Erasmus MC Pompe Survey* please contact one of the following persons:

- A representative of your national patient organization  
(see [worldpompe.org/index.php/affiliates](http://worldpompe.org/index.php/affiliates))
- Thomas Schaller  
Höhefeldstr. 26  
76356 Weingarten  
Germany  
Phone +49(7244)1230  
E-mail: [schaller@glykogenose.de](mailto:schaller@glykogenose.de)  
or any other member of the IPA Board  
(see [worldpompe.org/index.php/who\\_we\\_are/board](http://worldpompe.org/index.php/who_we_are/board))
- Dr. Deniz Güngör, Physician-Researcher  
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To avoid errors in translation and to minimise misunderstandings, you are encouraged to direct your requests primarily through your national patient organization.

If you have further questions about the *Pompe Registry* please contact your treating physician, a researcher at your national Pompe center (if available), a representative of your national patient association or a Board member of the IPA. Contact information for Genzyme representatives is given on the website [www.lsdregistry.net/pomperegistry](http://www.lsdregistry.net/pomperegistry).

## Acknowledgement

Deniz Güngör, Erasmus MC, made valuable contributions to this information, which is highly appreciated by the IPA.

(December 2009)