

Pompe patient information collection

Access to accurate, up-to-date medical information is imperative for you and your physician. To get this medical information, scientists, patients and industry have been working on a survey and a registry.

The IPA/Erasmus MC Pompe Survey is patient driven. Patients are the key role players in this questionnaire and provide scientists with data. The Pompe Registry is an electronic disease registry for data obtained through the patients' physicians. It provides immediate access to data and tools to assist physicians in the management of the disease. The Pompe Registry informational website is accessible over the Internet by Pompe patients and their physicians.



Both the IPA/Erasmus MC Pompe Survey and the Pompe Registry are very important initiatives to help learn more about Pompe disease and are complementary to each other. This handout introduces you to the goals of both databases, how they work and where you can find out more.

Q What is the IPA/Erasmus MC Pompe Survey?

A In 2002, the International Pompe Association (IPA) and Erasmus University Medical Center (MC) in Rotterdam, Netherlands jointly started the 'Pompe Survey' project, involving the development and distribution of a collection of questionnaires for older patients with Pompe disease. The purpose of this study was to describe the adult form of Pompe disease: the natural course, the severity of disease in the patient population, and the use of

(medical) care. A second goal was to develop and test measurement scales for the assessment of disease severity and the evaluation of changes over time. It is important to continue (or start) participation when you begin enzyme replacement therapy so that the effects of ERT can be followed from a patient's perspective.

A questionnaire was developed covering topics such as diagnosis, disease history, development in childhood, mobility, respiratory problems, specific symptoms, daily activities and use of care. Existing

Other names for Pompe disease

Acid alpha-glucosidase deficiency, acid maltase deficiency (AMD), glycogen storage disorder (GSD) type II, glycogenosis II, and lysosomal alpha-glucosidase deficiency. In different parts of the world, Pompe may be pronounced "pom-PAY," "POM-puh," or "pom-PEE."

questionnaires were added for the measurement of fatigue (fatigue severity scale), the level of handicap (Rotterdam 9-items handicap scale), and quality of life (SF-36 health survey).

Q How does the IPA/Erasmus MC Pompe Survey work?

A To participate in this patient driven survey, you can contact your patient organization, the IPA secretariat or the Pompe Center at the Erasmus MC in Rotterdam via www.pompecenter.nl. When your request for participation has been received you will be asked to sign an 'informed consent'. This is a letter that explains what the study is all about, the objectives and patient confidentiality. It can be returned directly to Erasmus MC or forwarded through your patient organization. After the Pompe Center in Rotterdam receives the informed consent document, you will receive the questionnaire at home. When you have finished filling in the questionnaire, you can send it back to the Pompe Center or your patient organization. The data you have provided will be entered in the computer and analyzed at Erasmus MC. If you agree, you will be asked to fill in the same questionnaire on an annual basis. This ongoing input provides valuable follow-up information on the natural course of the disease and the effect of your Myozyme treatment.

Q How confidential is the IPA/Erasmus MC Pompe Survey?

A The information you provide to the Pompe Center is confidential. Information from which your identity can be derived will be kept separate from the answers on the questionnaires, in a special database. This

database can only be accessed by a physician or researcher from the Erasmus MC Pompe Center. Every participant in the Survey has a unique study number. For the purpose of data analysis only this number (and not your name) will be used.

Q What will be done with the IPA/Erasmus MC Pompe Survey data?

A Every individual answer will be linked to the answers of other Pompe patients to get a good group overview. This approach supplies scientists with good individual and group follow-up. The IPA/Erasmus MC Pompe Survey results will be published in scientific journals so other physicians and researchers working on Pompe disease can benefit from the compiled data. You can find an overview of these publications at the Pompe Center website at www.pompecenter.nl under 'publications'.

Q How can I participate in the IPA/Erasmus MC Pompe Survey?

A Pompe patients have participated in this project through the IPA-affiliated patient organizations in the Netherlands, Germany, the United Kingdom, France, the United States, Australia and Canada. Others have participated directly through Erasmus MC on an individual basis. In France, the study is carried out in co-operation with both the patient organization and the Institut de Myologie in Paris.

It is still possible to participate in the IPA/Erasmus MC Pompe Survey. The questionnaire is available in Dutch, English, French, and German. For more information visit the Pompe Center website www.pompecenter.nl or contact the IPA secretariat.

Q

What is a disease registry?

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A disease registry is a database of medical information on patients with a specific medical condition. The collective information from the registry is used by physicians to increase the understanding of the disease and to monitor the effectiveness of any disease support measures. The ultimate goal is to improve the clinical outcomes of patients like you, which is why physician and patient participation is critical to the success of the registry. Registries have proven to be especially valuable in rare diseases like Pompe.

Q

What is the Pompe Registry?

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The Pompe Registry, initiated in September 2004 and sponsored by Genzyme, is a global, confidential, observational, Internet-based disease registry program, established to increase the understanding of the natural course of Pompe disease and to monitor patient outcomes. The Pompe Registry is a voluntary program open to all Pompe patients, irrespective of therapy.

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What are the objectives of the Pompe Registry?

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The Pompe Registry has several objectives. To begin, it allows patients and their physicians to better understand the variability, progression, and natural history of Pompe disease. The Pompe Registry is a tool which helps to characterize and describe the global Pompe patient population and monitor the effectiveness of any and all patient support measures over time. Through the Pompe Registry, physicians can access patient-specific

online reports to monitor disease progression and enhance clinical decision-making. Physicians who access the Pompe Registry are also able to obtain the latest disease guidelines, publications, and practice approaches. What's more, the Pompe Registry provides a foundation for collaborative studies and publications to address new areas in the management of Pompe disease.

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How does the Pompe Registry work?

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Let's assume that you and your physician have agreed to participate in the Pompe Registry. When you have regular check-ups, some of the clinical information collected by your physician is documented in the Pompe Registry. Examples of these data include cardiac, muscle, respiratory and quality of life information.

Only your physician has access to this individual information. However, data on all Registry patients can be viewed collectively and in an anonymous way by all participating physicians.

All medical or healthcare professionals treating patients with Pompe disease are encouraged to enroll and follow their Pompe patients in the Pompe Registry.

Myozyme is not offered as part of Registry participation.

Q

How confidential is the Pompe Registry information?

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Your physician must ask you for the appropriate authorization according to national privacy regulations and other state and local laws relating to medical information,

before data is submitted to the Pompe Registry. To maintain patient confidentiality, all patients are referenced by a Registry identification (ID) number only, not by other unique identifiers.

All physician information is also confidential. Physician-specific patient data remains confidential and is not released to other physicians without prior written approval. No physician-to-physician data comparisons are made. The Pompe Registry also allows participating physicians to compare clinical information from their Pompe patients with the collective data in the Registry.

Pompe Registry data are analyzed and reported periodically, and on an ad hoc basis as specific requests for registry data are received from the medical community. Physicians are encouraged to collaborate, share observations, and generate hypotheses for evaluation, as well as assist in the collection of clinical data in an effort to guide and assess future therapeutic interventions. As a result, the Pompe Registry is a long-term program and will proceed indefinitely.

You can visit www.pomperegistry.com to learn more about the Pompe Registry.

also visit www.pomperegistry.com for more information and can download the Pompe disease Schedule of Assessments. Patients will also need to complete the Pompe Registry Patient Authorization form that you can obtain through your treating physician.

Q

Can I have access to my personal data in the Pompe Registry and/or IPA/Erasmus MC Pompe Survey?

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Your data in the Pompe Registry can only be accessed through your physician. In exceptional situations, data from the Pompe Survey can be accessed if the patient specifically requests it.

Q

How can I enroll in the Pompe Registry?

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The Pompe Registry is continuously enrolling new physicians and their patients. Interested patients are encouraged to speak with their physician for more information. Patients can

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