EUROPEAN MEDICINES AGENCY
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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant human acid α-glucosidase for the treatment of glycogen storage disease type II (Pompe's disease)

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Disclaimer

Please note that revisions to the Public Summary of Opinion are purely administrative updates. Therefore, the scientific content of the document reflects the outcome of the Committee for Orphan Medicinal Products (COMP) at the time of designation and is not updated after first publication.

On 14 February 2001, orphan designation (EU/3/01/018) was granted by the European Commission to Genzyme B.V., The Netherlands, for recombinant human acid α-glucosidase for the treatment of glycogen storage disease type II (Pompe's disease).

The sponsorship was transferred to Genzyme Europe BV, The Netherlands, in April 2002.

What is glycogen storage disease type II (Pompe's disease)?

Patients with glycogen storage disease type II (Pompe's disease) do not have enough of a protein called alpha-glucosidase. This protein is an enzyme, whose function is to break down glycogen (a "storage" carbohydrate) to glucose. If the protein is not present, glycogen builds up in certain tissues, such as the heart and muscle tissue (including the diaphragm, the main breathing muscle under the lungs). The progressive build-up of glycogen causes a wide range of signs and symptoms including progressive weakness of respiratory muscles and enlarged heart and liver. The condition is chronically debilitating and life-threatening.

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What is the estimated number of patients affected by the condition?

At the time of designation, glycogen storage disease type II affected approximately 0.137 in 10,000 people in the European Union (EU). This was equivalent to a total of around 5,200 people*, and is below the ceiling for orphan designation, which is 5 people in 10,000. This is based on the information provided by the sponsor and the knowledge of the Committee for Orphan Medicinal Products (COMP).

What treatments are available?

No medicinal products were authorised for the treatment of glycogen storage disease type II (Pompe's disease) in the Community at the time of submission of the application for orphan drug designation. Current management is restricted to symptomatic treatment.

How is this medicine expected to work?

α-glucosidase is the enzyme (molecule that speeds up chemical reactions in the body) missing in patients with glycogen storage disease type II (Pompe's disease). By replacing the deficient enzyme with a functioning one, recombinant human acid α-glucosidase, this medicine is expected to restore the normal break down of glycogen and ultimately improve the overall outcome of the patients.

What is the stage of development of this medicine?

The effects of recombinant human acid α-glucosidase were evaluated in experimental models.

At the time of submission of the application for orphan designation, one clinical trial in patients with glycogen storage disease type II (Pompe's disease) was completed.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 19 December 2000 recommending the granting of this designation.

Update: recombinant human acid α-glucosidase (Myozyme) has been authorised in the EU since 29 March 2006 for long-term enzyme replacement therapy (ERT) in patients with a confirmed diagnosis of Pompe disease (acid α-glucosidase deficiency).

More information on Myozyme can be found in the European public assessment report (EPAR) on the Agency's website: ema.europa.eu/Find medicine/Human medicines/European Public Assessment Reports

* Disclaimer: For the purpose of the designation, the number of patients affected by the condition is estimated and assessed on the basis of data from the European Union.
At the time of designation, this represented a population of 378,800,000 (Eurostat 2001).

Opinions on orphan medicinal product designations are based on the following three criteria:

- the seriousness of the condition;
- the existence of alternative methods of diagnosis, prevention or treatment;
- either the rarity of the condition (affecting not more than 5 in 10,000 people in the EU) or insufficient returns on investment.

Designated orphan medicinal products are products that are still under investigation and are considered for orphan designation on the basis of potential activity. An orphan designation is not a marketing authorisation. As a consequence, demonstration of quality, safety and efficacy is necessary before a product can be granted a marketing authorisation.

For more information

Sponsor's contact details:

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For contact details of patients' organisations whose activities are targeted at rare diseases see:

- Orphanet, a database containing information on rare diseases which includes a directory of patients' organisations registered in Europe.
- European Organisation for Rare Diseases (EURORDIS), a non-governmental alliance of patient organisations and individuals active in the field of rare diseases.

Translations of the active ingredient and indication in all official EU languages[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Recombinant human acid α-glucosidase	Treatment of Glycogen Storage Disease type II (Pompe's disease)
Danish	Gensplejset human syre α-Glukosidase	Behandling af glycogenoplagringslidelse type II (Pompe's sygdom)
Dutch	Recombinante humaan zure α-glucosidase	Behandeling van de glycogeenstapelingsziekte type II (Pompe-ziekte)
Finnish	Rekombinoituva ihmisen happo α-glukoosidaasi	Tyyppi II glykogenoosin (Pompen tauti)hoito
French	α-glucosidase acide humaine recombinante	Traitement de la glycogénose de type II (maladie de Pompe)
German	Rekombinante humane saure O-Glukosidase	Behandlung der Glykogenspeicherkrankheit Typ II (Pompe-Krankheit)
Greek	Ανασυνδυασμένη ανθρώπινη όξινη α-γλυκοζιδάση	Θεραπεία της Γλυκογόνωσης τύπου II (Νόσος του Pompe)
Italian	α-glucosidasi acida umana ricombinante	Trattamento della glicogenosi, tipo II (malattia di Pompe)
Portuguese	α-glucosidase ácida humana recombinante	Tratamento da glicogenose de tipo II (Doença de Pompe)
Spanish	α glucosidasa ácida humana recombinante	Tratamiento de la enfermedad de almacenamiento del glucógeno tipo II (enfermedad de Pompe)
Swedish	Rekombinant human syra alfa-glukosidas	Behandling av glykogen upplagringssjukdom typ II (Pompes sjukdom)

[1] At the time of designation