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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Glycosylation independent lysosomal targeting (GILT)-tagged recombinant human acid alpha glucosidase for the treatment of glycogen storage disease type II (Pompe's disease)

On 27 October 2011, orphan designation (EU/3/11/921) was granted by the European Commission to BioMarin Europe Ltd, United Kingdom, for GILT-tagged recombinant human acid alpha glucosidase for the treatment of glycogen storage disease type II (Pompe's disease).

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

Glycogen storage disease type II is an inherited disorder that is caused by the lack of an enzyme called hydrolase acid alpha glucosidase (GAA). This enzyme is contained in lysosomes (part of the body's cells which break down nutrients and other materials). It breaks down glycogen (carbohydrate stored in the body) into glucose. Without this enzyme, glycogen builds up in the body, typically in muscle cells, causing damage to the cells.

Glycogen storage disease type II is a long-term debilitating and life-threatening disease because it causes breathing and heart problems.

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

At the time of designation, glycogen storage disease type II affected approximately 0.2 in 10,000 people in the European Union (EU)[*]. This is equivalent to a total of around 10,000 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?

At the time of designation, human recombinant alglucosidase alpha enzyme (Myozyme) was authorised for the treatment of glycogen storage disease type II in the EU. Myozyme is an 'enzyme replacement therapy' that works by replacing the missing GAA enzyme.

[*]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 (EU 27), Norway, Iceland and Liechtenstein. This represents a population of 506,300,000 (Eurostat 2011).

7 Westferry Circus ● Canary Wharf ● London E14 4HB ● United Kingdom
Telephone +44 (0)20 7418 8400 **Facsimile** +44 (0)20 7523 7040
E-mail info@ema.europa.eu **Website** www.ema.europa.eu

The sponsor has provided sufficient information to show that GILT-tagged recombinant human acid alpha glucosidase might be of significant benefit for patients with glycogen storage disease type II because early studies show that it may target muscle cells more effectively than the existing treatment. This assumption will need to be confirmed at the time of marketing authorisation, in order to maintain the orphan status.

How is this medicine expected to work?

The medicine is an 'enzyme replacement therapy' which is expected to work by replacing the missing GAA enzyme in glycogen storage disease type II, helping to break down glycogen and stopping it building up in the body. The medicine is produced by a method known as 'recombinant DNA technology': it is made by cells that have received a gene (DNA), which makes them able to produce GAA. The medicine also contains a chemical tag (GILT) designed to enhance its uptake into the lysosomes.

What is the stage of development of this medicine?

The effects of GILT-tagged recombinant human acid alpha glucosidase have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with GILT-tagged recombinant human acid alpha glucosidase in patients with glycogen storage disease type II were ongoing.

At the time of submission, GILT-tagged recombinant human acid alpha glucosidase was not authorised anywhere in the EU for glycogen storage disease type II. Orphan designation of GILT-tagged recombinant human acid alpha glucosidase had been granted in the United States for glycogen storage disease type II.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 8 September 2011 recommending the granting of this designation.

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:

BioMarin Europe Ltd.
164 Shaftesbury Avenue
London WC2H 8HL
United Kingdom
Telephone: + 44 207 420 0800
Telefax: + 44 207 420 08 29
E-mail: biomarin-europe@bmrn.com

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	Glycosylation independent lysosomal targeting tagged recombinant human acid alpha glucosidase	Treatment of glycogen storage disease type II (Pompe's disease)
Bulgarian	Гликолизационно независима лизозомно-насочена-белязана рекомбинирана човешка кисела алфа-гликозидаза	Лечение на гликогеноза тип II (Болест на Помпе)
Czech	Značená rekombinantní lidská kyselá alfa-glukosidasa cílená do lyzosomu nezávisle na glykosylaci	Léčba glykogen střádavé choroby typu II (Pompeho choroba)
Danish	Glykosyleringsuafhængig lysosomal targeteret tagged rekombinant human sur alfa-glucosidase	Behandling af glycogenoplagringslidelse type II (Pompe's sygdom)
Dutch	GILT (Glycosylatie independente lysosomiale "targeting") gelabelde recombinante humane zure alfa-glucosidase	Behandeling van de glycogeenstapelingsziekte type II (Pompe-ziekte)
Estonian	GILT (glycosylation independent lysosomal targeting)-märgistusega rekombinantne inimese happeline alfaglükosidaas	Glükogenoos II (Pompe tõve) ravi
Finnish	Glykosylaatiosta riippumaton lysosomaaliin kohdentuvasti merkitty rekombinantti ihmisen hapan alfaglukosidaasi	Tyyppi II glykogenoosin (Pompen tauti)hoito
French	Alpha-glucosidase acide recombinante humaine marquée à la GILT	Traitement de la glycogénose de type II (maladie de Pompe)
German	'Glykosylierungsunabhängiges lysosomales targeting'-getaggte rekombinante humane lyosomale alpha-Glucosidase	Behandlung der Glykogenspeicherkrankheit Typ II (Pompe-Krankheit)
Greek	Ανασυνδυασμένη ανθρώπινη όξινη α-γλυκοσιδάση, σημασμένη για λυσοσωμιακή στόχευση ανεξάρτητη γλυκοζυλίωσης.	Θεραπεία της Γλυκογόνωσης τύπου II (Νόσος του Pompe)
Hungarian	Glikoziláció-független lizoszomális targeting-címkés rekombináns humán savas alfa-glükozidáz	II.-es típusú glycogentárolási betegség kezelése (Pompe-kór)
Italian	Alfa glucosidasi acida umana ricombinante GILT-marcata	Trattamento della glicogenosi, tipo II (malattia di Pompe)

[1] At the time of designation

Language	Active ingredient	Indication
Latvian	Glikolizēšanas neatkarīgā lizosomālā mērķa marķera rekombinēta cilvēka skābā alfa-glukozidāze	2 tipa glikogēna uzkrāšanās slimības (Pompes slimība) ārstēšana
Lithuanian	Nepriklausanti glikozilinimui lizosomų žymėtoji rekombinantinė rūgštinė žmogaus alfa gliukozidazė	II tipo glikogenozės (Pompe ligos) gydymas
Maltese	Aċidu alpha glucosidase rikombinanti uman immarkat permezz ta' proċess li jimmira l-lisosomi mingħajr il-bżonn ta' glikosilazzjoni	Kura tal-glikoġenożi tat-tip II (marda ta' Pompe)
Polish	Rekombinowana ludzka kwaśna alfa glukozydaza o wlaściwościach lizosomalnego pozycjonowania nizależnego od glikozylacji	Leczenie choroby spichrzeniowej glukogenu typu II (choroby Pompego)
Portuguese	Alfa-glucosidase ácida humana recombinante marcada com GILT (Glycosylation independent lysosomal targeting)	Tratamento da glicogenose de tipo II (Doença de Pompe)
Romanian	Alfa-glucozidază acidă umană recombinantă marcată prin tehnica ţintei lipozomale independente de glicozilare	Tratamentul glicogenozei de tip II (Boala lui Pompe)
Slovak	Rekombinantná ľudská kyslá α-glukozidáza označená pomocou lyzozomálneho zacielenia nezávislého od glykozylácie	Liečba glykogenózy typu II (Pompeho choroba)
Slovenian	Od glikozilacije neodvisna označena lizosomska človeška rekombinantna kisla alfa glukozidaza	Zdravljenje bolezni kopičenja glikogena tip II (Pompejeva bolezen)
Spanish	Alfa glucosidasa ácida humana recombinante fundida con un fragmento direccionador lisosomal independiente de la glucosilación	Tratamiento de la enfermedad de almacenamiento del glucógeno tipo II (enfermedad de Pompe)
Swedish	Glykosylerings-oberoende, lysosomalt-kopplat rekombinant humant surt alfaglukosidas	Behandling av glykogen upplagringssjukdom typ II (Pompes sjukdom)
Norwegian	Glykosylerings-uavhengig lysosomal målrettingsmerket rekombinant human syre-alfaglukosidase	Behandling av glykogenose type II (Pompes sykdom)
Icelandic	Sykrunar óháð lýsósómal miðað merkt manna raðbrigða alfa-sýruglúkósíðasa	Meðferð við glýkógen upphleðslusjúkdómi af gerð II (Pompe sjúkdómur)