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Public summary of opinion on orphan designation

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

On 21 March 2018, orphan designation (EU/3/18/2000) was granted by the European Commission to Amicus Therapeutics UK Ltd, United Kingdom, for recombinant human acid alpha-glucosidase (also known as ATB200) 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, also known as Pompe's disease, is an inherited disorder caused by the lack of an enzyme called acid alpha glucosidase (GAA). This enzyme is contained in lysosomes (part of the body's cells that break down nutrients and other materials). GAA breaks down glycogen (a complex sugar stored in the body) into glucose (a simple sugar). When this enzyme is lacking, large amounts of glycogen build up in the muscles, including the heart and diaphragm (the main breathing muscle under the lungs). The progressive build-up of glycogen causes a wide range of signs and symptoms, including heart problems, breathing difficulties and muscle weakness.

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

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

At the time of designation, glycogen storage disease type II affected approximately 0.3 in 10,000 people in the European Union (EU). This was equivalent to a total of around 16,000 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, Myozyme (alglucosidase alfa) 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 28), Norway, Iceland and Liechtenstein. This represents a population of 517,400,000 (Eurostat 2018).

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The sponsor has provided sufficient information to show that the medicine might be of significant benefit for patients with glycogen storage disease type II because early studies have found that the medicine improves muscle function better than the authorised 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 similar to the natural GAA enzyme. The medicine, which is given by injection into a vein, is intended to be used with another medicine called miglustat given by mouth. Miglustat prevents the medicine from breaking down in the blood and so more of the GAA enzyme is expected to get into the lysosomes.

The medicine is expected to replace the missing enzyme thus improving the symptoms of the disease.

What is the stage of development of this medicine?

The effects of the medicine have been evaluated in experimental models.

At the time of submission, clinical trials with the medicine in patients with glycogen storage disease type II were ongoing.

At the time of submission, the medicine was not authorised anywhere in the EU for glycogen storage disease type II. Orphan designation of the medicine had been granted in the United States for the condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 15 February 2018 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:

Contact details of the current sponsor for this orphan designation can be found on EMA website, on the medicine's rare disease designations page.

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 alpha-glucosidase	Treatment of glycogen storage disease type II (Pompe's disease)
Bulgarian	Рекомбинантна човешка киселa алфа-глюкозидаза	Лечение на тип 2 гликогеноза (Болест на Помпе)
Croatian	Rekombinantna ljudska kisela alfa-glukozidaza	Liječenje bolesti taloženja glikogena tip II (Pompeova bolest)
Czech	Lidská rekombinantní kyselá alfa-glukosidáza	Léba glykogenózy typu II (Pompeho choroba)
Danish	Rekombinant human syre alfaglucosidase	Behandling af glycogenose type II (Pompes sygdom)
Dutch	Recombinant humane zure alfa-glucosidase	Behandeling van de glycogeenstapelingsziekte type II (Pompe-ziekte)
Estonian	Rekombinantne inimese happeline alfa-glükosidaas	2.tüüpi glükogenoosi (Pompe tõve) ravi
Finnish	Rekombinantti ihmisen hapan alfaglukosidaasi	Tyyppi II glykogenoosin (Pompen tauti) hoito
French	Alpha-glucosidase acide recombinante humaine	Traitement de la glycogénose de type II (maladie de Pompe)
German	Rekombinante humane Saure Alpha-Glucosidase	Behandlung der Glykogenspeicherkrankheit Typ II (Pompe-Krankheit)
Greek	Ανασυνδυασμένη ανθρώπινη όξινη α-γλυκοσιδάση	Θεραπεία της Γλυκογόνωσης τύπου ΙΙ (Νόσος του Pompe)
Hungarian	Rekombináns emberi savas alfa-glükozidáz	II-es típusú glikogéntárolási betegség (Pompe-kór) kezelése
Italian	Alfa-glucosidasi acida umana ricombinante	Trattamento della glicogenosi, tipo II (malattia di Pompe)
Latvian	Rekombinanta cilvēka skābā alfa-glikozidāze	Glikogēna uzkrāšanas II tipa traucējumu (Pompe slimība) ārstēšana
Lithuanian	Rekombinantinė žmogaus rūgštinė alfa-glikozidazė	II tipo glikogenozės (Pompe ligos) gydymas
Maltese	Aċidu rikombinanti tal-bniedem alfa-glukosidaži	Kura tal-glikoġenożi tat-tip II (marda ta' Pompe)
Polish	Rekombinowana ludzka kwaśna alfa-glukozydaza	Leczenie choroby spichrzania glikogenu typu II (choroby Pompego)
Portuguese	Alfa-glicosidase ácida humana recombinante	Tratamento da glicogenose de tipo II (Doença de Pompe)
Romanian	Alfa-glucozidază acidă umană recombinantă	Tratamentul glicogenozei tip II (boala Pompe)
Slovak	Rekombinantná ľudská alfa-glukozidáza	Liečba glykogenózy typ II (Pompeho choroba)

[1] At the time of designation

Language	Active ingredient	Indication
Slovenian	Rekombinantna humana kisla alfa-glukozidaza	Zdravljenje glikogenoze tipa II (Pompejeva bolezen)
Spanish	Alfa-glucosidasa ácida humana recombinante	Tratamiento de la enfermedad de almacenamiento del glucógeno tipo II (enfermedad de Pompe)
Swedish	Humant rekombinant alfaglukosidas	Behandling av glykogen upplagringssjukdom typ II (Pompes sjukdom)
Norwegian	Rekombinant human sur alfaglukosidase	Behandling av glykogenose type II (Pompes sykdom)
Icelandic	Raðbrigða, manna sýru alfa glúkósídasa	Meðferð á glýkógenupphleðslu sjúkómi af gerð II (Pompes sjúkdómur)