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EMA/COMP/513707/2016
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant human acid alpha-glucosidase conjugated with mannose-6-phosphate analogues for the treatment of glycogen storage disease type II (Pompe's disease)

On 29 August 2016, orphan designation (EU/3/16/1726) was granted by the European Commission to NanoMedSyn, France, for recombinant human acid alpha-glucosidase conjugated with mannose-6-phosphate analogues 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 simpler 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 15,000 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).

[*]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 513,700,000 (Eurostat 2016).

30 Churchill Place ● Canary Wharf ● London E14 5EU ● United Kingdom
Telephone +44 (0)20 3660 6000 **Facsimile** +44 (0)20 3660 5555
Send a question via our website www.ema.europa.eu/contact

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.

The sponsor has provided sufficient information to show that the medicine might be of significant benefit for patients with glycogen storage disease type II. This is because early studies in the laboratory suggest that it can improve muscle function and regeneration better than existing therapies. 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 a version of the GAA enzyme. It has been modified by the addition of a compound to improve the uptake of GAA by muscle cells and to increase entry into the lysosomes, where it replaces the missing enzyme.

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 of the application for orphan designation, no clinical trials with the medicine in patients with glycogen storage disease type II had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for glycogen storage disease type II or designated as an orphan medicinal product elsewhere for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 13 July 2016 recommending the granting of this designation.

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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 conjugated with mannose-6-phosphate analogues	Treatment of glycogen storage disease type II (Pompe's disease)
Bulgarian	Рекомбинантна човешка кисела алфа-глюкозидаза, свързана с аналози на маноза-6-фосфат	Лечение на тип 2 гликогеноза (Болест на Помпе)
Croatian	Rekombinantna ljudska kisela alfa-glukozidaza, konjugirana s analogom manoza-6-fosfata	Liječenje bolesti taloženja glikogena tip II (Pompeova bolest)
Czech	Rekombinantní humánní kyselina alfa-glukozidová konjugovaná s analogem manóza -6 fosfátu	Léba glykogenózy typu II (Pompeho choroba)
Danish	Rekombinant human syre alfa-glucosidase konjugeret med mannose-6-phosphat analoger	Behandling af glycogenose type II (Pompes sygdom)
Dutch	Recombinante humane zure alpha-glucosidase geconjugeerd aan mannose-6-phosphaatanalogen	Behandeling van de glycogeenstapelingsziekte type II (Pompe-ziekte)
Estonian	Rekombinantne inimese happeline alfa-glükosidaas konjugeeritud mannoos-6-fosfaadi analoogidega	2.tüüpi glükogenoosi (Pompe tõve) ravi
Finnish	Rekombinantti humaani hapan alfa-glukosidaasi, joka on konjugoitu mannoosi-6-fosfaatin analogeilla	Tyyppi II glykogenoosin (Pompen tauti) hoito
French	Alpha-glucosidase acide recombinante humaine conjuguée aux analogues du mannose-6-phosphate	Traitement de la glycogénose de type II (maladie de Pompe)
German	Rekombinante humane Acid-Alpha-Glucosidase konjugiert an Mannose-6-Phosphat-Analogien	Behandlung der Glykogenspeicherkrankheit Typ II (Pompe-Krankheit)
Greek	Ανασυνδυασμένο ανθρώπινο οξύ της α- γλυκοσιδάσης συζευγμένο με ανάλογο της -6 -φωσφορική μαννόζης	Θεραπεία της Γλυκογόνωσης τύπου II (Νόσος του Pompe)
Hungarian	Mannóz-6-foszfát analógokkal konjugált rekombináns humán savas alfa-glukozidáz	II-es típusú glikogéntárolási betegség (Pompe-kór) kezelése
Italian	Alfa glucosidase acida ricombinante umana coniugata con analoghi di mannoso-6-fosfato	Trattamento della glicogenosi, tipo II (malattia di Pompe)
Latvian	Rekombinanta cilvēka skābā alfa-glukozidāze, kas konjugēta ar mannozes-6-fosfāta analogiem	Glikogēna uzkrāsanas II tipa traucējumu (Pompe slimība) ārstēšana
Lithuanian	Rekombinantinė žmogaus rūgštinė alfa-gliukozidazė, sujungta su manozės-6-fosfato analogais	II tipo glikogenozės (Pompe ligos) gydymas
Maltese	Aċidu alfa-glucosidase uman rikombinanti kkonjugat ma' analogi ta' mannose-6-phosphate	Kura tal-glikoġenożi tat-tip II (marda ta' Pompe)
Polish	Rekombinowana ludzka kwaśna alfa-glukozydaza połączona z analogami mannozo-6-fosforanowymi	Leczenie choroby spichrzania glikogenu typu II (choroby Pompego)

[1] At the time of designation

Language	Active ingredient	Indication
Portuguese	Alfa-glucosidase ácida humana recombinante conjugada com análogos de manose-6-fosfato	Tratamento da glicogenose de tipo II (Doença de Pompe)
Romanian	Alfa-glucozidază acidă recombinantă umană conjugate cu analogi ai manozei-6-fosfat	Tratamentul glicogenozei tip II (boala Pompe)
Slovak	Rekombinantná ľudská alfa-glukozidáza konjugovaná s analógmi manóza-6-fosfátu	Liečba glykogenózy typ II (Pompeho choroba)
Slovenian	Rekombinantna humana kisla alfa-gukozigaza konjugirana z analogi manoza-6-fosfata	Zdravljenje glikogenoze tipa II (Pompejeva bolezen)
Spanish	Alfa-glucosidasa acida umana recombinante conjugada con analagos de manosa-6-fosfato	Tratamiento de la enfermedad de almacenamiento del glucógeno tipo II (enfermedad de Pompe)
Swedish	Rekombinant human alfaglukosidassyra konjugerad med mannos-6-fosfat analoger.	Behandling av glykogen upplagringssjukdom typ II (Pompes sjukdom)
Norwegian	Rekombinant human sur alfa-glucosidase konjugert med mannose-6-fosfat analoger	Behandling av glykogenose type II (Pompes sykdom)
Icelandic	Raðbrigða manna súr alfa-glúkódídasisamtengdurmeð mannose-6-fósfat samstæðum	Meðferð á glýkógenupphleðslu sjúkómi af gerð II (Pompes sjúkdómur)