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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Velaglucerase alfa for the treatment of Gaucher disease

On 6 June 2010, orphan designation (EU/3/10/752) was granted by the European Commission to Shire Pharmaceuticals Ireland Limited, Ireland, for velaglucerase alfa for the treatment of Gaucher disease.

What is Gaucher disease?

Gaucher disease is an inherited disorder that is caused by the lack of an enzyme called glucocerebrosidase. This enzyme normally breaks down a fatty waste product called glucocerebroside. Without the enzyme, glucocerebroside builds up in the body, typically in the liver, spleen and bone marrow. This causes a wide range of symptoms, including anaemia (low red blood cell counts), tiredness, easy bruising and a tendency to bleed, an enlarged spleen and liver, and bone pain and fractures.

Gaucher disease is a long-term, debilitating and life-threatening disease that is associated with a reduced life expectancy if left untreated.

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

At the time of designation, Gaucher disease affected approximately 0.3 in 10,000 people in the European Union (EU)*. This is equivalent to a total of around 15,000 people, and is below the threshold 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, two medicines, imiglucerase and miglustat, were authorised for the treatment of Gaucher disease in the EU. Imiglucerase is an 'enzyme replacement therapy' that works by replacing the missing enzyme. Miglustat blocks the production of glucocerebroside and is used in patients who cannot receive enzyme replacement therapy.

*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 506,500,000 (Eurostat 2010).

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The sponsor has provided sufficient information to show that velaglucerase alfa might be of significant benefit for patients with Gaucher disease because it may represent an alternative treatment to imiglucerase, should the long-term supply problems that are occurring with this medicine continue or happen again in the future. Velaglucerase alfa might also be less 'immunogenic' than imiglucerase. This means that it might be less likely to trigger the production of antibodies (proteins that are produced in response to a substance, which can reduce the effects of a treatment). These assumptions 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?

Velaglucerase alfa is an enzyme replacement therapy that is expected to work by replacing the missing enzyme in Gaucher disease, helping to break down glucocerebroside and stopping it building up in the body. Velaglucerase alfa is produced by a method known as 'recombinant DNA technology': it is made by human cells that have received a gene (DNA), which make them able to produce the enzyme.

What is the stage of development of this medicine?

The effects of velaglucerase alfa have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with velaglucerase alfa in patients with Gaucher disease were ongoing.

At the time of submission, velaglucerase alfa was authorised and had been granted orphan designation in the United States of America for Gaucher disease.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 3 March 2010 recommending the granting of this designation.

Update: velaglucerase alfa (Vpriv) was authorised in the EU on 26 August 2010 for for long-term enzyme replacement therapy (ERT) in patients with type 1 Gaucher disease.

More information on Vpriv 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

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	Velaglucerase alfa	Treatment of Gaucher disease
Bulgarian	Велаглуцераза алфа	Лечение на болест на Гоше
Czech	Velagluceráza alfa	Léčba Gaucherovy choroby
Danish	Velaglucerase alfa	Behandling af Gauchers sygdom
Dutch	Velaglucerase-alfa	Behandeling van de ziekte van Gaucher
Estonian	Alfa-velaglütseraas	Gaucher' tõve ravi
Finnish	Alfavelagluseraasi	Gaucherin taudin hoito
French	Vélaglucérase alfa	Traitement de la maladie de Gaucher
German	Velaglucerase Alfa	Behandlung der Gaucher-Krankheit
Greek	Βελαγλυκεράση άλφα	Θεραπευτική αγωγή για την νόσο του Gaucher
Hungarian	Velagluceráz-alfa	Gaucher-kór kezelése
Italian	Velaglucerasi alfa	Trattamento della malattia di Gaucher
Latvian	Alfa velaglicerāze	Gošē slimības ārstēšana
Lithuanian	Alfa velaglucerazė	Gošė ligos gydymas
Maltese	Velaglucerase alfa	Kura tal-marda ta' Gaucher
Polish	Welagluceraza alfa	Leczenie choroby Gaucher'a
Portuguese	Velaglucerase alfa	Tratamento da doença de Gaucher
Romanian	Velaglucerază alfa	Tratamentul bolii Gaucher
Slovak	Velagluceráza alfa	Liečba Gaucherovej choroby
Slovenian	Velagluceraza alfa	Zdravljenje Gaucherove bolezni
Spanish	Velaglucerasa alfa	Tratamiento de la enfermedad de Gaucher
Swedish	Velaglucerase alfa	Behandling av Gauchers sjukdom
Norwegian	Velaglucerase alfa	Behandling av Gauchers sykdom
Icelandic	Velaglúkerasi alfa	Meðferð á Gauchersveiki

[1] At the time of designation