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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

N-butyldeoxygalactonojirimycin for the treatment of Fabry 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 9 August 2012, orphan designation (EU/3/12/1033) was granted by the European Commission to Actelion Registration Limited, United Kingdom, for N-butyldeoxygalactonojirimycin for the treatment of Fabry disease.

What is Fabry disease?

Fabry disease is an inherited disease that is caused by the lack of an enzyme called alpha-galactosidase A. This enzyme is contained in lysosomes (part of the body's cells that break down nutrients and other materials) where it breaks down and removes globotriaosylceramide (Gb3, a complex molecule of sugars and a fatty substance stored in the body). When this enzyme is lacking, large amounts of Gb3 build up in certain tissues, such as the kidney, heart, nervous system and skin. The progressive build-up of Gb3 causes symptoms including pain in the hands and feet, lesions on the skin and clouding of the front part of the eye, and can lead to complications such as kidney and heart problems and neurological complications affecting the brain.

Fabry disease is a long-term debilitating disease due to recurrent episodes of severe pain not responding to analgesics and life-threatening due to kidney, heart and brain complications.

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

At the time of designation, Fabry disease affected less than 2.3 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 117,000 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, Fabrazyme (agalsidase beta) and Replagal (agalsidase alfa) were authorised in the EU to treat Fabry disease.

The sponsor has provided sufficient information to show that N-butyldeoxygalactonojirimycin might be of significant benefit for patients with Fabry disease because early studies in experimental models suggest that it works in a different way to existing treatments, which may represent an alternative for patients who do not respond or are not eligible for current 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?

N-butyldeoxygalactonojirimycin is a type of sugar expected to block an enzyme involved in the production of Gb3, thereby preventing build-up of Gb3 in patients who lack the enzyme to break Gb3 down. This is called 'substrate reduction therapy' and it differs from other therapies that aim to replace the missing enzyme (enzyme replacement therapies).

What is the stage of development of this medicine?

The effects of N-butyldeoxygalactonojirimycin have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials in patients with Fabry disease had been started.

At the time of submission, N-butyldeoxygalactonojirimycin was not authorised anywhere in the EU for Fabry disease 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 11 July 2012 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.

*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.
At the time of designation, this represented a population of 509,000,000 (Eurostat 2012).

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	N-Butyldeoxygalactonojirimycin	Treatment of Fabry disease
Bulgarian	N-Бутилдеоксигалактоножиримицин	Лечение на болест на Fabry
Czech	N-Butyldeoxygalaktonojirimycin	Léčba Fabryho choroby
Danish	N-Butyldeoxygalactonojirimycin	Behandling af Fabrys sygdom
Dutch	N-Butyldeoxygalactonojirimycine	Behandeling van de ziekte van Fabry
Estonian	N-butüüldeoksügalaktonojirimütsiin	Fabry tõve ravi
Finnish	N-butyylideoksigalaktonijirimysiini	Fabryn taudin hoito
French	N-butyl-désoxygalactonojirimycine	Traitement de la maladie de Fabry
German	N-Butyldeoxygalactonojirimycin	Behandlung des Fabry-Syndroms
Greek	Ν-βουτυλοδεοξυγαλακτονοτζιριμυκίνη	Αγωγή κατά της νόσου του Fabry
Hungarian	N-butil-dezoxi-galaktonojirimicin	Fabry betegség kezelése
Italian	N-Butildeossigalattonogirimicina	Trattamento della malattia di Fabry
Latvian	N-butil-deoksi-galaktonojirimicīns	Fabrī slimības ārstēšana
Lithuanian	N-butildeoksigalaktonojirimicinas	Fabry ligos gydymas
Maltese	N-Butyldeoxygalactonojirimycin	Kura tal-marda ta' Fabry
Polish	N-Butylodeoksygalaktonojirymycyna	Leczenie choroby Fabry'ego
Portuguese	N-Butil-desoxigalactonojirimicina	Tratamento da doença de Fabry
Romanian	N-Butildeoxigalactonojirimicina	Tratamentul bolii Fabry
Slovak	N-butyldeoxygalaktonojirimycín	Liečba Fabryho choroby
Slovenian	N-butildeoksigalaktonojirimicin	Zdravljenje Fabryjeve bolezni
Spanish	N-butildesoxigalactonojirimicina	Tratamiento de la enfermedad de Fabry
Swedish	N-butyldeoxigalaktonojorimycin	Behandling av Fabrys sjukdom
Norwegian	N-butyldeoksygalaktonojirimycin	Behandling av Fabrys sykdom
Icelandic	N-bútýldeoxýgalaktónójírímýcín	Meðferð Fabry-sjúkdóms

[1] At the time of designation