20 April 2015
EMA/COMP/32699/2014 Rev.1
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant human acid ceramidase for the treatment of Farber disease

First publication	31 March 2014
Rev.1: transfer of sponsorship	20 April 2015

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 19 February 2014, orphan designation (EU/3/14/1243) was granted by the European Commission to QOL Therapeutics UK Ltd, United Kingdom, for recombinant human acid ceramidase for the treatment of Farber disease.

The sponsorship was transferred to Plexcera Therapeutics EU Limited, Ireland, in April 2015.

What is Farber disease?

Farber disease is an inherited disease that affects the metabolism of lipids (the breakdown and use of fats in the body). Patients with the disease lack an enzyme called 'acid ceramidase', which is essential for the breakdown of fats. In the absence of this enzyme, fats accumulate abnormally in cells and tissues throughout the body, particularly around the joints. Farber disease usually manifests itself during infancy with three classic signs: swollen and painful joints, small lumps (nodules) under the skin and a hoarse voice or a weak cry. Affected individuals may also have difficulty breathing, an enlarged liver and spleen, and developmental delay.

Farber disease is a long-term debilitating condition which, in its most severe forms, can be life-threatening because of its effects on several organs.

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

At the time of designation, Farber disease affected less than 0.01 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 500 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, no satisfactory methods were authorised in the EU for the treatment of Farber disease.

How is this medicine expected to work?

This medicine is a copy of the enzyme acid ceramidase naturally produced by the body. It is expected to work by replacing the missing enzyme and therefore restoring the body's ability to break down and use fats.

The medicine is made by a method known as 'recombinant DNA technology': it is made by cells into which a gene (DNA) has been introduced that makes them able to produce human acid ceramidase.

What is the stage of development of this medicine?

The effects of recombinant human acid ceramidase have been evaluated in experimental models.

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

At the time of submission, recombinant human acid ceramidase was not authorised anywhere in the EU for Farber disease. Orphan designation of the medicine had been granted in the United States for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 9 January 2014 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 28), Norway, Iceland and Liechtenstein.
At the time of designation, this represented a population of 512,900,000 (Eurostat 2014).

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	Recombinant human acid ceramidase	Treatment of Farber disease
Bulgarian	Рекомбинантна, човешка кисела церамидаза	Лечение на болест на Фарбер
Czech	Rekombinantní lidská ceramidasová kyselina	Léčba Fabryho choroby
Croatian	Rekombinantna ljudska kisela ceramidaza	Liječenje Farberove bolesti
Danish	Rekombinant human ceramidase	Farber sygdom
Dutch	Recombinant humaan ceramidasezuur	Behandeling van ziekte van Farber
Estonian	Rekombinantne inimese happeline keramidaas	Farber'i haiguse ravi
Finnish	Rekombinanttitekniikalla tehty ihmisen hapan seramidaasi	Farberin taudin hoito
French	Céramidase acide humaine recombinante	Traitement de la maladie de Farber
German	Rekombinante menschliche saure Ceramidase	Behandlung der Farber-Krankheit
Greek	Ανασυνδυασμένη ανθρώπινη όξινη κεραμιδάση	Θεραπεία της νόσου Farber
Hungarian	Rekombináns human savanyú ceramidase	Farber betegség kezelése
Italian	Ceramidasi acida ricombinante umana	Trattamento della malattia di Farber
Latvian	Rekombinanta cilvēka skābā keramidāze	Farbera slimības ārstēšana
Lithuanian	Rekombinantinė žmogaus ceramidazės rūgštis	*Farber* ligos gydymas
Maltese	Aċidu ceramidase uman rikombinanti	Kura tal-marda ta' Farber
Polish	Rekombinowana ludzka kwaśna ceramidaza	Leczenie choroby Farbera
Portuguese	Ceramidase ácida humana recombinante	Tratamento da doença de Farber
Romanian	Ceramidază acidă umană recombinantă	Tratamentul bolii Farber
Slovak	Rekombinantná ľudská kyslá ceramidáza	Liečba Farberovej choroby
Slovenian	Rekombinantna humana kisla ceramidaza	Zdravljenje Farberjeve bolezni
Spanish	Ceramidase ácido humano recombinante	Tratmiento de la enfermedad de Farber
Swedish	Rekombinant mänskligt surt ceramidas	Behandling av Farbers sjukdom
Norwegian	Rekombinant human sur ceramidase	Behandling av Farbers sykdom
Icelandic	Raðbrigða manna súr ceremídasi	Meðferð við Farbers sjúkdómi

[1] At the time of designation