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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant human lysosomal acid lipase for the treatment of lysosomal acid lipase deficiency

First publication	18 January 2011
Rev.1: transfer of sponsorship	21 September 2011
Rev.2: sponsor's change of address	10 March 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 17 December 2010, orphan designation (EU/3/10/827) was granted by the European Commission to HungaroTrial Ltd, Hungary, for recombinant human lysosomal acid lipase for the treatment of lysosomal acid lipase deficiency.

The sponsorship was transferred to Synageva BioPharma Ltd, United Kingdom, in August 2011.

What is lysosomal acid lipase deficiency?

Lysosomal acid lipase deficiency is an inherited disease caused by the lack of one of the enzymes needed to break down fats within cells. In the absence of this enzyme, called lysosomal acid lipase, fats accumulate in the body's cells and tissues, causing symptoms such as growth failure, enlarged liver, diarrhoea and malabsorption (when nutrients from food are not easily absorbed during digestion).

Lysosomal acid lipase deficiency is a severe and life-threatening disease which, in its most severe form, is usually fatal in the first year of life.

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

At the time of designation, lysosomal acid lipase deficiency affected less than 0.2 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 10,000 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, no satisfactory methods were authorised in the EU for the treatment of lysosomal acid lipase deficiency. Patients were advised to follow a diet low in fats. In some cases, haematopoietic (blood) stem cell transplantation had been used, but with modest results. This is a complex procedure where the patient receives stem cells from a matched donor to help restore the bone marrow.

How is this medicine expected to work?

Recombinant human lysosomal acid lipase is an 'enzyme replacement therapy' that is expected to work by replacing the missing enzyme in lysosomal acid lipase deficiency, helping to break down fats and stopping them building up in the body's cells.

Recombinant human lysosomal acid lipase is produced by a method known as 'recombinant DNA technology': it is extracted from the eggs of hens that have been given genes that make them able to produce an exact copy of the human enzyme in their eggs.

What is the stage of development of this medicine?

The effects of recombinant human lysosomal acid lipase 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 lysosomal acid lipase deficiency had been started.

At the time of submission, recombinant human lysosomal acid lipase was not authorised anywhere in the EU for the treatment of lysosomal acid lipase deficiency. Orphan designation of the medicine had been granted in the United States of America for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 7 October 2010 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 506,300,000 (Eurostat 2010).

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 lysosomal acid lipase	Treatment of lysosomal acid lipase deficiency
Bulgarian	Рекомбинантна човешка лизозомна кисела липаза	Лечение на недостиг на лизозомна кисела липаза
Czech	Rekombinantní lidská kyselá (lysozomální) lipáza	Léčba deficitu kyselé (lysozomální) lipázy
Danish	Rekombinant human lysosomal sur lipase	Behandling af lysosomal sur lipase mangel
Dutch	Recombinant humaan lysosomaal lipase zuur	Behandeling van lysosomaal zuur lipase deficiëntie
Estonian	Rekombinantne inimese lüsosomaalse happe lipaas	Lüsosomaalse happe lipaasi puudulikkuse ravi
Finnish	Rekombinantti ihmisen lysosomaalinen hapan lipaasi	Lysosomaalisen happaman lipaasin puutoksen hoito
French	Lipase acide lysosomale recombinante humaine	Traitement des déficits en lipase acide lysosomale
German	Rekombinante humane lysosomale saure Lipase	Behandlung von lysosomaler saurer Lipasedefizienz
Greek	Ανασυνδυασμένη ανθρώπινη λυσοσωματική οξινη λιπάσης	Θεραπεία ανεπάρκειας λυσοσωμικης οξινης λιπάσης.
Hungarian	Rekombináns humán lizoszómális savi lipáz	A lizoszómális savi lipáz defektus kezelése
Italian	Lipasi acida lisosomiale umana ricombinante	Trattamento delle carenze di lipasi acida lisosomiale
Latvian	Rekombinantā cilvēka lizosomu skābes lipāze	Lizosomu skābes lipāzes deficīta ārstēšana
Lithuanian	Rekombinantinė žmogaus lizosomų rūgštinė lipazė	Lizosomų rūgštinės lipazės stokos gydymas
Maltese	Lysosomal acid lipase uman rikombinanti	Kura ta` nuqqas ta` lysosomal acid lipase
Polish	Rekombinowana ludzka lizosomalna lipaza kwaśna.	Leczenie niedoboru lizosomalnej lipazy kwaśnej.
Portuguese	Lipase ácida lisosómica humana recombinante	Tratamento da deficiência de Lipase de ácido lisossomal
Romanian	Lipază acidă lizozomală umană recombinantă	Tratarea deficienţei lipazei acide lizozomale
Slovak	Rekombinantná ľudská lyzozómová kyslá lipáza	Liečba deficitu lyzozómovej kyslej lipázy
Slovenian	Rekombinantna človeška lizosomska kisla lipaza	Zdravljenje pomankanja lizosomske kisle lipaze
Spanish	Lipasa lisosomal ácida recombinante humana	Tratamiento de la deficiencia de la lipasa lisosomal ácida.
Swedish	rekombinant humant lysosomalt syrligt lipas	Behandling av lysosomalt syrligt lipas brist

[1] At the time of designation

Language	Active ingredient	Indication
Norwegian	Rekombinant human lysosomal sur lipase	Behandling av lysosomal sur lipasemangel
Icelandic	Raðbrigða manna leysiagnasýru lípasi (lysosomal acid lipase)	Meðferð við skorti á leysiagnasýrulípasa