10 June 2014
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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Kifunensine for the treatment of gamma sarcoglycanopathy

First publication	13 October 2011
Rev.1: administrative update	18 February 2013
Rev.2: withdrawal from the Community Register	10 June 2014

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.

Please note that this product was withdrawn from the Community Register of designated Orphan Medicinal Products in May 2014 on request of the Sponsor.

On 27 September 2011, orphan designation (EU/3/11/908) was granted by the European Commission to Généthon, France, for kifunensine for the treatment of gamma sarcoglycanopathy.

What is gamma sarcoglycanopathy?

Sarcoglycanopathies belong to a group of inherited diseases called the limb-girdle muscular dystrophies, which cause progressive muscle wasting and weakness. Symptoms usually appear in childhood. Sarcoglycanopathies are caused by mutations in the genes for certain proteins called sarcoglycans, which are important for muscle fibres and are needed for muscles to work properly. Each type of sarcoglycanopathy is genetically different but people have similar symptoms. Gamma sarcoglycanopathy is caused by a fault in the gene for gamma sarcoglycan.

Gamma sarcoglycanopathy is a chronically debilitating and life-threatening condition as it causes progressive muscle wasting associated with reduced life expectancy due to heart and breathing problems.

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

At the time of designation, gamma sarcoglycanopathy affected less than 0.02 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 1,000 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 application, no satisfactory methods for the treatment of gamma sarcoglycanopathy had been authorised in the European Union. Treatment of patients with the disease mainly involved physiotherapy as supportive treatment, but also tendon-lengthening surgery and fusion of the spine to minimise painful deformity.

How is this medicine expected to work?

Kifunensine is expected to block the action of an enzyme called alpha mannosidase, which is involved in the natural process of breaking down defective sarcoglycans in the human body. By blocking the action of this enzyme, it is expected that defective sarcoglycan will not be degraded but can be repaired and used again by the muscle.

What is the stage of development of this medicine?

At the time of submission of the application for orphan designation, the evaluation of the effects of kifunensine in experimental models was ongoing.

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

At the time of submission, kifunensine was not authorised anywhere in the EU for gamma sarcoglycanopathy 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 15 July 2011 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 507,700,000 (Eurostat 2011).

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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http://www.genethon.fr/en/contacts-en/

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	Kifunensine	Treatment of gamma-sarcoglycanopathy
Bulgarian	Кифунензин	Лечение на гама-саркогликанопатия
Czech	Kifunensin	Léčba gamma-sarkoglykanopatie
Danish	Kifunensin	Behandling af gamma-sarcoglycanopati
Dutch	Kifunensine	Behandeling van gamma-sarcoglycanopathie
Estonian	Kifunensiin	Gamma-sarkoglükanopaatia ravi
Finnish	Kifunensiini	Gamma-sarkoglukanopatian hoito
French	Kifunensine	Traitement de la gamma-sarcoglycanopathie
German	Kifunensin	Behandlung der gamma-Sarkoglykanopathie
Greek	Κιφουνενσίνη	Θεραπεία της γάμμα-σαρκοπολυσακχαριδοπάθειας
Hungarian	Kifunenzin	Gamma-sarcoglycanopathia kezelése
Italian	Kifunensine	Trattamento della gamma-sarcoglicanopatia
Latvian	Kifunenzīns	Gamma-sarkoglikanopātijas ārstēšana
Lithuanian	Kifunenzinas	Gama sarkoglikanopatijos gydymas
Maltese	Kifunensine	Kura tal-gamma-sarkoglikanopatija
Polish	Kifunensyna	Leczenie gamma-sarkoglikanopatii
Portuguese	Kifunensina	Tratamento da gama-sarcoglicanopatia
Romanian	Kifunensină	Tratamentul gamma-sarcoglicanopatiei
Slovak	Kifunenzín	Liečba gama-sarkoglykanopatie
Slovenian	Kifunenzin	Zdravljenje gama-sarkoglikanopatije
Spanish	Kifunensina	Tratamiento de la gamma-sarcoglicanopatía
Swedish	Kifunensin	Behandling av gamma-sarcoglycanopati
Norwegian	Kifunensin	Behandling av gamma-sarkoglykanopati
Icelandic	Kífúnensín	Meðferð við gamma-sarkóglýkankvilla

[1] At the time of designation