5 September 2016
EMA/COMP/447479/2016
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Triheptanoin for the treatment of McArdle's disease

On 14 July 2016, orphan designation (EU/3/16/1710) was granted by the European Commission to Vall d'Hebron Institute of Research, Spain, for triheptanoin for the treatment of McArdle's disease.

What is McArdle's disease?

McArdle's disease (also known as glycogen storage disease type V) is an inherited disorder caused by the lack of an enzyme called myophosphorylase. This enzyme breaks down glycogen (a complex sugar) stored in the muscles into glucose (a simple sugar), which is used to generate energy. When this enzyme is lacking, less energy is made in the muscles, leading to decreased ability to exercise, muscle fatigue (loss of strength in muscles), contractures (permanent shortening and hardening of muscles), and sometimes myoglobinuria (presence of a protein from broken-down muscle in the urine, which can damage the kidneys).

McArdle's disease is a long-term debilitating disease because it reduces the ability to exercise, causes tiredness and stiffness, and shortens and hardens muscles. It is also life-threatening because it can cause sudden breakdown of muscle, which can cause kidney damage and lead to death.

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

At the time of designation, McArdle's disease affected approximately 0.1 in 10,000 people in the European Union (EU). This was equivalent to a total of around 5,000 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 submission of the application for orphan designation, no satisfactory method had been authorised in the European Union for the treatment of McArdle's disease.

[*]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. This represents a population of 513,700,000 (Eurostat 2016).

30 Churchill Place ● Canary Wharf ● London E14 5EU ● United Kingdom
Telephone +44 (0)20 3660 6000 **Facsimile** +44 (0)20 3660 5555
Send a question via our website www.ema.europa.eu/contact

How is this medicine expected to work?

Triheptanoin is an oil which is broken down in the liver into substances that can be used to generate energy without the need for myophosphorylase, the enzyme lacking in patients with McArdle's disease. By bypassing the need for myophosphorylase, this medicine is expected to increase energy generation in muscle and improve the symptoms of the disease.

What is the stage of development of this medicine?

The effects of the medicine have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with triheptanoin in patients with McArdle's disease were ongoing.

At the time of submission, triheptanoin was not authorised anywhere in the EU for McArdle's 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 16 June 2016 recommending the granting of this designation.

_____

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:

Contact details of the current sponsor for this orphan designation can be found on EMA website, on the medicine's rare disease designations page.

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	Triheptanoin	Treatment of McArdle's disease
Bulgarian	Трихептаноин	Лечение на болест на McArdle
Croatian	Triheptanoin	Liječenje McArdleove bolesti
Czech	Triheptanoin	Léčba Mcardlovi choroby
Danish	Triheptanoin	Behandling af McArdles sygdom
Dutch	Triheptanoin	Behandeling van de ziekte McArdle
Estonian	Triheptanoiin	McArdle haiguse ravi
Finnish	Triheptanoiini	McArdlen taudin hoito
French	Triheptanoïne	Traitement de la maladie de McArdle
German	Triheptanoin	Behandlung der McArdle-Krankheit
Greek	Τριεπτανοΐνη	Θεραπεία της νόσου McArdle
Hungarian	Triheptanoin	McArdle betegség kezelése
Italian	Trieptanoina	Trattamento della malattia di McArdle
Latvian	Triheptanoīns	*McArdle* slimības ārstēšana
Lithuanian	Triheptanoinas	*McArdle* ligos gydymas
Maltese	Triheptanoin	Kura tal-marda ta' McArdle
Polish	Triheptanoina	Leczenie choroby McArdle'a
Portuguese	Tri-heptanoína	Tratamento da doença de McArdle
Romanian	Triheptanoin	Tratamentul bolii McArdle
Slovak	Triheptanoín	Liečba ochorení McArdle
Slovenian	Triheptanoin	Zdravljenje McArdle bolezni
Spanish	Triheptanoína	Tratamiento de la enfermedad de McArdle
Swedish	Triheptanoin	Behandling av McArdle sjukdom
Norwegian	Triheptanoin	Behandling av McArdle sykdom
Icelandic	Tríheptanóín	Meðferð við McArdle sjúkdómi

[1] At the time of designation