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EMA/COMP/269979/2015
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Triheptanoin for the treatment of glucose transporter type-1 deficiency syndrome

On 21 May 2015, orphan designation (EU/3/15/1495) was granted by the European Commission to Pharma Gateway AB, Sweden, for triheptanoin for the treatment of glucose transporter type-1 deficiency syndrome.

What is glucose transporter type-1 deficiency syndrome?

Glucose transporter type-1 deficiency syndrome is an inherited disorder that affects brain function. Patients with this disorder lack 'glucose transporter type-1', a protein responsible for the transport of glucose (sugar) from the blood into the brain.

Glucose is the main source of energy for the brain, and shortage of glucose leads to impairment of brain function and growth. Most children with the condition present with seizures (fits) in the first few months of life, although some children can develop seizures later on or have other neurological symptoms.

Glucose transporter type-1 deficiency is a long-term debilitating disease that leads to seizures, mental disabilities and movement disorders.

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

At the time of designation, glucose transporter type-1 deficiency syndrome 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).

*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 512,900,000 (Eurostat 2015).

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What treatments are available?

At the time of designation, no satisfactory methods were authorised in the EU for the treatment of glucose transporter type-1 deficiency syndrome. Patients were usually managed with measures such as a high-fat diet and antiepileptic medicines to treat seizures (although most patients did not respond to these medicines).

How is this medicine expected to work?

The medicine is made of fatty substances, called fatty acids, which are broken down in the liver into smaller substances called ketones.

Ketones can serve as an alternative energy source in the body and, as they do not rely on glucose transporter type-1 to enter the brain, the brain can use them in place of glucose. By meeting the brain's energy requirements with this alternative energy source, the medicine is expected to improve the symptoms of glucose transporter type-1 deficiency syndrome.

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 the medicine in patients with glucose transporter type-1 deficiency syndrome were ongoing.

At the time of submission, the medicine was not authorised anywhere in the EU for glucose transporter type-1 deficiency syndrome. 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 16 April 2015 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:

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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	Triheptanoin	Treatment of glucose transporter type-1 deficiency syndrome
Bulgarian	Трихептаноин	лечение на синдрома на дефицит на глюкозния транспортер от тип 1
Croatian	Triheptanoin	Liječenje sindroma deficijencije prijenosnika glukoze tip 1
Czech	Triheptanoin	Léčba deficitu glukózového přenašeče typu 1
Danish	Triheptanoin	Behandling af glukosetransporter type 1-mangelsyndrom
Dutch	Triheptanoin	Behandeling van glucosetransporter type 1-deficiëntiesyndroom
Estonian	Triheptanoiin	Glükoositransporter 1 puudulikkuse sündroomi ravi
Finnish	Triheptanoiini	Tyypin I glukoosinkuljettajan puutosoireyhtymän hoito
French	Triheptanoïne	Traitement du syndrome du déficit en transporteur de glucose de type 1
German	Triheptanoin	Behandlung des Glucosetransporter-Typ-1 Mangel-Syndroms
Greek	Τριεπτανοΐνη	Θεραπεία του συνδρόμου ανεπάρκειας του μεταφορέα γλυκόζης τύπου 1
Hungarian	Triheptanoin	A glükóz transzporter 1 hiány szindróma kezelésére
Italian	Trieptanoina	Trattamento della sindrome da deficit del trasportatore di glucosio di tipo 1
Latvian	Triheptanoīns	Glikozes 1. tipa transportētāja deficīta sindroma ārstēšana
Lithuanian	Triheptanoinas	Gliukozės pernašos baltymo stokos sindromo gydymas (*GLUT1* stoka)
Maltese	Triheptanoin	Kura tas-sindrome ta' nuqqas tat-trasportatur tal-glukożju tat-tip 1
Polish	Triheptanoina	Leczenie zespołu niedoboru transportera glukozy typu 1
Portuguese	Tri-heptanoína	Tratamento da síndrome de deficiência do transportador de glicose tipo 1
Romanian	Triheptanoin	Tratamentul sindromului de deficienţă a transportorului de glucoză de tip 1
Slovak	Triheptanoín	Liečba syndrómu z deficitu transportéra glukózy typu-1
Slovenian	Triheptanoin	Zdravljenje sindroma pomanjkanja glukoznega prenašalca tipa-1
Spanish	Triheptanoína	Tratamiento del síndrome de deficiencia del transportador de la glucosa tipo 1
Swedish	Triheptanoin	Behandling av glukostransportprotein typ 1-brist
Norwegian	Triheptanoin	Behandling av glukosetransportør type-1 mangelsyndrom
Icelandic	Tríheptanóín	Meðferð á heilkenni glúkósaferjuskorts af gerð 1

[1] At the time of designation