

7 May 2018  
EMA/120664/2018

## Public summary of opinion on orphan designation

### Gemfibrozil for the treatment of neuronal ceroid lipofuscinosis

On 21 March 2018, orphan designation (EU/3/18/1993) was granted by the European Commission to Quintiles Ireland Limited, Ireland, for gemfibrozil for the treatment of neuronal ceroid lipofuscinosis.

In May 2018, Quintiles Ireland Limited changed name to IQVIA RDS Ireland Limited.

#### What is neuronal ceroid lipofuscinosis?

Neuronal ceroid lipofuscinosis is a group of inherited diseases where substances known as lipofuscins made of fats and proteins build up in the brain and other parts of the body, such as the eye. Symptoms of the disease include delayed speech, inability to coordinate muscle movements, fits, loss of vision and mental disability.

Neuronal ceroid lipofuscinosis is a debilitating and life-threatening condition that leads to death by early adulthood.

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

At the time of designation, neuronal ceroid lipofuscinosis affected approximately 0.5 in 10,000 people in the European Union (EU). This was equivalent to a total of around 26,000 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 designation, the medicine Brineura was authorised in the EU for the treatment of neuronal ceroid lipofuscinosis type 2. Brineura replaces the TPP1 protein missing in this form of the disease and it is given by infusion (drip) directly into the brain. The disease was also managed by treating its symptoms.

The sponsor has provided sufficient information to show that the medicine might be of significant benefit for patients with neuronal ceroid lipofuscinosis because laboratory data show improvements in

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\*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 517,400,000 (Eurostat 2018).

survival and movement skills. The medicine would be given by mouth, which is an advantage over the currently authorised medicine.

This assumption will need to be confirmed at the time of marketing authorisation, in order to maintain the orphan status.

## **How is this medicine expected to work?**

Gemfibrozil is expected to work in several ways, including by increasing production and activity of TPP1 (the protein lacking in patients with neuronal ceroid lipofuscinosis type 2), by increasing production of lysosomes (part of the body's cells that break down nutrients and other materials), and by preventing cell death and inflammation. These activities are together expected to improve the outcome of patients affected by the condition.

## **What is the stage of development of this medicine?**

The effects of gemfibrozil have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with gemfibrozil in patients with neuronal ceroid lipofuscinosis had been started.

Gemfibrozil is authorised in several EU countries to help lower the level of fats in the blood.

At the time of submission, gemfibrozil was not authorised anywhere in the EU for neuronal ceroid lipofuscinosis. Orphan designation of the medicine had been granted in the US for neuronal ceroid lipofuscinoses.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 15 February 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.

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<sup>1</sup>, Norwegian and Icelandic

Language	Active ingredient	Indication
English	Gemfibrozil	Treatment of neuronal ceroid lipofuscinosis
Bulgarian	Гемфиброзил	Лечение на невронална цериодна липофусциноза
Croatian	Gemfibrozil	Liječenje neuronske ceroidne lipofuscinoze
Czech	Gemfibrozil	Léčba neuronální ceroidní lipofuscinózy
Danish	Gemfibrozil	Behandling af neuronal ceroid lipofuscinose
Dutch	Gemfibrozil	Behandeling van neuronaal ceroid lipofuscinose
Estonian	Gemfibrosiil	Neuronaalse tseroidse lipofustsinoosi ravi
Finnish	Gemfibrotsiili	Neuronaalisen seroidilipofuskinoosin hoito
French	Gemfibrozil	Traitement de la céroïde-lipofuscinose neuronale
German	Gemfibrozil	Behandlung der neuronalen Ceroid-Lipofuszinose
Greek	Γεμφιμπροζίλη	Θεραπεία της Νευρωνικής Κηροειδούς Λιποφουσκίνωσης
Hungarian	Gemfibrozil	Neuronális ceroid lipofuscinosis kezelése
Italian	Gemfibrozil	Trattamento della ceroido lipofuscinosi neuronale
Latvian	Gemfibrozils	Neironu ceroīdās lipofuscinozes ārstēšana
Lithuanian	Gemfibrozilis	Neuronų ceroidinės lipofuscinozės gydymas
Maltese	Ġemfibrožil	Kura taċ-ċerojdu lipofuxinosi newronali
Polish	Gemfibrozyl	Leczenie ceroidolipofuscynozy neuronalnej
Portuguese	Gemfibrozil	Tratamento da lipofuscinose ceróide neuronal
Romanian	Gemfibrozil	Tratamentul lipofuscinozei ceroide neuronale
Slovak	Gemfibrozil	Liečba neuronálnej ceroidnej lipofuscinózy
Slovenian	Gemfibrozil	Zdravljenje nevronalne ceroidne lipofuscinoze
Spanish	Gemfibrozilo	Tratamiento de la lipofuscinosis neuronal ceróidea
Swedish	Gemfibrozil	Behandling av neuronal ceroidlipofuscinosis
Norwegian	Gemfibrozil	Behandling av nevronal ceroid lipofuscinose
Icelandic	Gemfibrózil	Meðferð við taugaceróíð lípófúskinósis

<sup>1</sup> At the time of designation