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Public summary of opinion on orphan designation

Venglustat for the treatment of GM2 gangliosidosis

On 21 August 2020, orphan designation EU/3/20/2325 was granted by the European Commission to Genzyme Europe B.V., Netherlands, for venglustat for the treatment of GM2 gangliosidosis.

What is GM2 gangliosidosis?

GM2 gangliosidosis is an inherited disorder that causes progressive damage to the nerve cells in the brain and spinal cord.

Patients with this condition have mutations (defects) in at least one of 3 genes (HEXA, HEXB, and GM2A) that are responsible for the production of proteins that break down substances called GM2 gangliosides. Because of these defects, GM2 gangliosides build up in the body, particularly in the brain and spinal cord. Signs and symptoms include muscle weakness and problems with walking, intellectual disability, difficulty speaking, seizures (fits), and loss of sight and hearing.

GM2 gangliosidosis is a debilitating and life-threatening disease. The most severe form of the disease starts in early infancy and can lead to death in childhood.

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

At the time of designation, GM2 gangliosidosis affected approximately 0.4 in 10,000 people in the European Union (EU). This was equivalent to a total of around 21,000 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, no satisfactory methods were authorised in the EU to treat GM2 gangliosidosis. Treatment of patients was mainly supportive and included physical therapy and medicines to manage seizures.

*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, Iceland, Liechtenstein, Norway and the United Kingdom. This represents a population of 519,200,000 (Eurostat 2020).

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How is this medicine expected to work?

Venglustat is expected to block one of the steps in the formation of GM2 ganglioside, reducing the production and build up of this substance in the body. This is expected to improve the symptoms of GM2 gangliosidosis and help patients live longer.

What is the stage of development of this medicine?

The effects of venglustat 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 GM2 gangliosidosis had been started.

At the time of submission, venglustat was not authorised anywhere in the EU for the treatment of GM2 gangliosidosis or designated as an orphan medicinal product elsewhere for this condition.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 16 July 2020, 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

Contact details of the current sponsor for this orphan designation can be found on EMA website.

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	Venglustat	Treatment of GM2 gangliosidosis
Bulgarian	Венглустат	Лечение на GM2 ганглиозидоза
Croatian	Venglustat	Liječenje GM2 gangliozidoze
Czech	Venglustat	Léčba GM2 gangliosidozy
Danish	Venglustat	Behandling af GM2 gangliosidosis
Dutch	Venglustat	Behandeling vanGM2 gangliosidose
Estonian	Venglustaat	GM2 gangliosidoosi ravi
Finnish	Venglustaatti	GM2-gangliosidoosin hoito
French	Venglustat	Traitement de la gangliosidose à GM2
German	Venglustat	Behandlung der GM2 Gangliosidose
Greek	Βενγλουστάτη	Θεραπεία της γαγγλιοσίδοσης GM2
Hungarian	Venglusztát	GM2 gangliozidózis kezelése
Italian	Venglustat	Trattamento della gangliosidosi GM2
Latvian	Venglustats	GM2 gangliozidozes ārstēšana
Lithuanian	Venglustatas	GM2 gangliozidozės gydymas
Maltese	Venglustat	Kura ta’ ganglijosidoži GM2
Polish	Wenglustat	Leczenie gangliozydozy GM2
Portuguese	Venglustate	Tratamento da gangliosidose GM2
Romanian	Venglustat	Tratamentul gangliozidozei GM2
Slovak	Venglustat	Liečba GM2 gangliozidózy
Slovenian	Venglustat	Zdravljenje GM2 gangliozidoze
Spanish	Venglustat	Tratamiento de Gangliosidosis GM2
Swedish	Venglustat	Behandling av GM2-gangliosidos
Norwegian	Venglustat	Behandling av GM2 gangliosidose
Icelandic	Venglustat	Meðferð á GM2 ganglíósídósis

[1] At the time of designation