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Public summary of opinion on orphan designation

Acetylleucine for the treatment of GM2 gangliosidosis

On 12 December 2017, orphan designation (EU/3/17/1949) was granted by the European Commission to IntraBio Ltd, United Kingdom, for acetylleucine (also known as IBI1000) 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 lack an enzyme called beta-hexosaminidase A, which normally breaks down a substance called GM2 ganglioside. Without this enzyme, GM2 ganglioside builds up in the body, particularly in the brain and spinal cord, causing progressive nerve damage. Signs and symptoms include muscle weakness and problems with walking, intellectual disability, difficulty speaking, seizures (fits), 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 during childhood.

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

At the time of designation, GM2 gangliosidosis affected less than 0.5 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 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, 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.

*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 515,700,000 (Eurostat 2017).

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How is this medicine expected to work?

Acetylleucine has been authorised in some countries for the treatment of vertigo (a spinning sensation that affects balance). The way acetylleucine works in GM2 gangliosidosis is not clear, but it is thought to stabilise the nerve cells responsible for balance and for coordinating movement. This is expected to improve movement control and coordination in patients with 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, no clinical trials with the medicine in patients with GM2 gangliosidosis had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for GM2 gangliosidosis 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 31 October 2017 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[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Acetylleucine	Treatment of GM2 gangliosidosis
Bulgarian	Ацетиллевцин	Лечение на GM2 ганглиозидоза
Croatian	Acetilleucin	Liječenje GM2 gangliozidoze
Czech	Acetylleucin	Léčba GM2 gangliosidozy
Danish	Acetylleucine	Behandling af GM2 gangliosidosis
Dutch	Acetylleucine	Behandeling vanGM2 gangliosidose
Estonian	Atsetüülleutsiin	GM2 gangliosidoosi ravi
Finnish	Asetyylileusiini	GM2-gangliosidoosin hoito
French	Acétylleucine	Traitement de la gangliosidose à GM2
German	Acetylleucin	Behandlung der GM2 Gangliosidose
Greek	Ακετυλευκίνη	Θεραπεία της γαγγλιοσίδωσης GM2
Hungarian	Acetilleucin	GM2 gangliozidózis kezelése
Italian	Acetilleucina	Trattamento della gangliosidosi GM2
Latvian	Acetilleicīns	GM2 gangliozidozes ārstēšana
Lithuanian	Acetilleucinas	GM2 gangliozidozės gydymas
Maltese	Acetylleucine	Kura ta' ganglijosidoži GM2
Polish	Acetyloleucyna	Leczenie gangliozydozy GM2
Portuguese	Acetilleucina	Tratamento da gangliosidose GM2
Romanian	Acetilleucină	Tratamentul gangliozidozei GM2
Slovak	Acetylleucín	Liečba GM2 gangliozidózy
Slovenian	Acetillevcin	Zdravljenje GM2 gangliozidoze
Spanish	Acetileucina	Tratamiento de Gangliosidosis GM2
Swedish	Acetylleucin	Behandling av GM2-gangliosidos
Norwegian	Acetylleucin	Behandling av GM2 gangliosidose
Icelandic	Asetýlleucín	Meðferð á GM2 ganglíósídósis

[1] At the time of designation