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# Public summary of opinion on orphan designation

## Trehalose for the treatment of neuronal ceroid lipofuscinosis

On 19 October 2020, orphan designation EU/3/20/2347 was granted by the European Commission to Theranexus S.A.S., France, for trehalose for the treatment of neuronal ceroid lipofuscinosis.

### What is neuronal ceroid lipofuscinosis?

Neuronal ceroid lipofuscinosis is a group of inherited diseases where deposits known as lipofuscins made of fats and proteins build up in the brain and other parts of the body, such as the eye, causing nerve damage. Symptoms of the disease include loss of vision, 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 0.1 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 designation, the medicine Brineura was authorised in the EU for the treatment of type 2 neuronal ceroid lipofuscinosis. 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. Laboratory studies indicate that it can reduce the death of nerve cells in type 3 neuronal ceroid lipofuscinosis, for which there are no authorised treatments.

*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).

**Official address** Domenico Scarlattilaan 6 ● 1083 HS Amsterdam ● The Netherlands
**Address for visits and deliveries** Refer to www.ema.europa.eu/how-to-find-us
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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?

Trehalose is a simple sugar consisting of two molecules of glucose. Based on results from laboratory studies, trehalose has the potential to slow the worsening of the disease by reducing the build-up of fats and proteins and relieving inflammation of the nerves. Trehalose is expected to be used together with miglustat, another medicine which has been granted orphan designation for the treatment of neuronal ceroid lipofuscinosis.

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

The effects of trehalose have been evaluated in experimental models.

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

At the time of submission, trehalose was not authorised anywhere in the EU for the treatment of neuronal ceroid lipofuscinosis 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 10 September 2020, 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

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.