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EMA/270550/2017

## Public summary of opinion on orphan designation

### Ursodeoxycholic acid for the treatment of Niemann-Pick disease

On 22 May 2017, orphan designation (EU/3/17/1878) was granted by the European Commission to IntraBio Ltd, United Kingdom, for ursodeoxycholic acid for the treatment of Niemann-Pick disease.

#### What is Niemann-Pick disease?

Niemann-Pick disease is a group of genetic disorders belonging to the larger family of metabolic disorders called 'lysosomal storage diseases', in which fats build up within lysosomes (parts of the body's cells that break down nutrients and other materials). This affects the way cells work and causes them to die, affecting normal functioning of tissues and organs, including the liver, spleen and brain.

There are three types of Niemann-Pick disease (A, B and C), with different genetic causes and different symptoms, which may include behavioural problems, learning disabilities and difficulty moving and speaking.

Niemann-Pick disease is seriously debilitating and life threatening since the build-up of fatty substances can cause brain damage and swelling of organs such as the spleen and liver.

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

At the time of designation, Niemann-Pick disease affected approximately 0.1 in 10,000 people in the European Union (EU). This was equivalent to a total of 5,000 people<sup>\*</sup>, 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, Zavesca (miglustat) was authorised in the EU to treat Niemann-Pick disease, type C. Zavesca was used to treat the neurological symptoms of the disease (symptoms affecting the brain and nerves).

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<sup>\*</sup>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).

The sponsor has provided sufficient information to show that ursodeoxycholic acid might be of significant benefit for patients with Niemann-Pick disease because early data in patients indicated that the medicine may improve liver function. 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?**

Ursodeoxycholic acid is known to reduce the content of cholesterol in the liver, but the exact mechanism through which it may work in Niemann-Pick disease is not clearly understood. Preliminary data show it may improve liver function as well as neurological symptoms (affecting the brain).

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

The effects of ursodeoxycholic acid have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with ursodeoxycholic acid in patients with Niemann-Pick disease were ongoing.

At the time of submission, ursodeoxycholic acid was authorised in the EU for the treatment of gallstones and liver diseases such as primary biliary cirrhosis.

At the time of submission, ursodeoxycholic acid was not authorised anywhere in the EU for Niemann-Pick disease 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 11 April 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<sup>1</sup>, Norwegian and Icelandic

Language	Active ingredient	Indication
English	Ursodeoxycholic acid	Treatment of Niemann-Pick disease
Bulgarian	Урзодезоксихолева киселина	Лечение на болест на Нийман-Пик
Croatian	Ursodeoksiholna kiselina	Liječenje Niemann-Pickove bolesti
Czech	Kyselina ursodeoxycholová	Léčba Niemann-Pickovy choroby
Danish	Ursodeoxycholsyre	Behandling af Niemann-Picks sygdom
Dutch	Ursodeoxycholzuur	Behandeling van de ziekte van Niemann-Pick
Estonian	Ursodeoksükoolhape	Niemann-Pick'i tõve ravi
Finnish	Ursodeoksikoolihappo	Niemann-Pick'n taudin hoito
French	Acide ursodésoxycholique	Traitement de la maladie de Niemann-Pick
German	Ursodeoxycholsäure	Zur Behandlung des Morbus Niemann-Pick
Greek	Ουρσοδεοξυχολικό οξύ	Θεραπεία της Νόσου Niemann-Pick
Hungarian	Urzodezoxikólsav	Niemann-Pick betegség kezelése
Italian	Acido ursodesossicolico	Trattamento della Malattia di Niemann-Pick
Latvian	Ursodeoksiholskābe	Nīmaņa-Pika slimības ārstēšanai
Lithuanian	Ursodeoksicholio rūgštis	Niemann-Picko ligos gydymas
Maltese	Aċidu ursodeossikoliku	Kura tal-marda ta' Niemann-Pick
Polish	Kwas ursodeoksycholowy	Leczenie choroby Niemann-Picka
Portuguese	Ácido ursodesoxicólico	Tratamento da Doença de Niemann-Pick
Romanian	Acid ursodeoxicolic	Tratamentul bolii Niemann-Pick
Slovak	Kyselina ursodeoxycholová	Liečba Niemannovej-Pickovej choroby
Slovenian	Ursodeoksiholna kislina	Zdravljenje Niemann-Pickove bolezni
Spanish	Ácido ursodesoxicólico	Tratamiento de la Enfermedad de Niemann-Pick
Swedish	Ursodeoxicholsyra	Behandling av Niemann-Picks sjukdom
Norwegian	Ursodeoksykolsyre	Behandling av Niemann-Picks sykdom
Icelandic	Úrsódeoxýkolsýra	Til meðferðar á Niemann-Pick sjúkdómi

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