

22 June 2015
EMA/COMP/280663/2015
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Trehalose for the treatment of oculopharyngeal muscular dystrophy

On 21 May 2015, orphan designation (EU/3/15/1496) was granted by the European Commission to Dr Ulrich Granzer, Germany, for trehalose for the treatment of oculopharyngeal muscular dystrophy.

What is oculopharyngeal muscular dystrophy?

Oculopharyngeal muscular dystrophy is a hereditary condition marked by weakness of the muscles around the eyes and throat, leading to symptoms such as drooping eyelids and difficulty swallowing. As muscle weakness progresses, weakness in other parts of the body may also develop, particularly in the arms near the shoulder and in the upper legs and hips. Patients usually present with this condition in their sixties or seventies.

Oculopharyngeal muscular dystrophy is debilitating in the long term because of its symptoms such as drooping eyelids and difficulty swallowing, and because it spreads to other parts of the body affecting mobility.

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

At the time of designation, oculopharyngeal muscular dystrophy 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, no satisfactory methods were authorised in the EU for treating oculopharyngeal muscular dystrophy. Patients were mainly treated with surgery to correct drooping eyelids and improve swallowing.

^{*}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 512,900,000 (Eurostat 2015).

How is this medicine expected to work?

Trehalose is a sugar molecule known to stabilise certain proteins. It is expected to work by preventing the clumping together of a protein known as PABPN1, which occurs in muscles of patients with oculopharyngeal muscular dystrophy and is linked to the muscle weakness.

By preventing the clumping together of these proteins in the muscles, trehalose is expected to help reduce the muscle weakness, and thereby relieve symptoms of the condition.

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 oculopharyngeal muscular dystrophy had been started in Europe.

At the time of submission, trehalose was not authorised anywhere in the EU for oculopharyngeal muscular dystrophy. Orphan designation had been granted in the United States for the condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 16 April 2015 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

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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 languages¹, Norwegian and Icelandic

Language	Active ingredient	Indication
English	Trehalose	Treatment of oculopharyngeal muscular dystrophy
Bulgarian	Трехалоза	Лечение на окулофарингеална мускулна дистрофия
Croatian	Trehaloza	Liječenje okuloфарингеалne mišićne distrofije
Czech	Trehalosa	Léčba pacientů s oculopharyngeal muskulární dystrofií
Danish	Trehalose	Behandling af oculopharyngeal muskeldystrofi
Dutch	Trehalose	Behandeling van okuloфарингеale spierdystrofie
Estonian	Trehaloos	Okulofarüngeaalse lihasdüstroofia ravi
Finnish	Trehaloosi	Okulofaryngeaalisien lihasdystrofian hoito
French	Tréhalose	Traitement de la dystrophie musculaire oculopharyngée
German	Trehalose	Behandlung der okuloфарингеalen Muskeldystrophie
Greek	Τρεαλόζη	Θεραπεία της οφθαλμοφαρυγγικής μυϊκής δυστροφίας
Hungarian	Trehalóz	Oculopharyngealis izomsorvadás kezelése
Italian	Trealosio	Trattamento della distrofia muscolare okuloфарингеa
Latvian	Trehaloze	Okulofaringeālās muskuļu distrofijas ārstēšana
Lithuanian	Trehalozė	Akių – ryklės (okulofaringinės) raumenų distrofijos gydymas
Maltese	Trehalose	Kura tad-distrofija muskolari okuloфарингил
Polish	Trehaloza	Leczenie dystrofii mięśniowej oczno-gardzielowej
Portuguese	Trealosa	Tratamento da distrofia muscular okuloфарингеa
Romanian	Trehaloză	Tratamentul distrofiei musculare okuloфаринgiene
Slovak	Trehalóza	Liečba okuloфарингеalnej muskulárnej dystrofie
Slovenian	Trehaloza	Zdravljenje okuloфарингеalne mišične distrofije
Spanish	Trehalosa	Tratamiento de la distrofia muscular okuloфарингеa
Swedish	Trehalos	Behandling av okuloфарингеal muskeldystrofi
Norwegian	Trehalose	Behandling av okuloфарингеal muskeldystrofi
Icelandic	Trehalósi	Meðferð augn-og kok vöðvarýrnunar

¹ At the time of designation