3 May 2017
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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Sialic acid for treatment of GNE myopathy

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On 16 October 2015, orphan designation (EU/3/12/972) was granted by the European Commission to Ultragenyx UK Limited, United Kingdom, for sialic acid (also known as aceneuramic acid) for treatment of GNE myopathy.

The sponsorship was transferred to Ultragenyx Germany GmbH, Germany, in January 2017.

What is GNE myopathy?

GNE myopathy is an inherited muscle-wasting disease. Patients with the disease have a mutation (change) in a gene responsible for the GNE enzyme which is involved in the production of sialic acid. Lack of sialic acid is believed to be the cause of the wasting of muscles in people with this condition. The disease worsens over time as more muscles become affected, progressively reducing mobility and eventually confining the patient to a wheelchair.

GNE myopathy is a life-threatening and long-term debilitating condition due to progressive muscle wasting, which means that the patient becomes unable to live independently.

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

At the time of designation, GNE myopathy affected less than 0.1 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 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 submission, no satisfactory methods were authorised in the EU for the treatment of GNE myopathy. Patients received treatment to relieve their symptoms such as physiotherapy to improve muscle strength, and other physical supports to aid mobility.

*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).

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How is this medicine expected to work?

As a lack of sialic acid is believed to cause the muscle wasting in this condition, this medicine is expected to work by providing the missing sialic acid and thus helping to ensure normal muscle function.

The medicine is being developed as 'extended-release' tablets, designed to release the active substance over a prolonged period. This is important because sialic acid is quickly removed from the blood and a constant supply of sialic acid is needed to improve muscle function in this condition.

What is the stage of development of this medicine?

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

At the time of submission of the application for orphan designation, clinical trials with the medicine in patients with GNE myopathy were ongoing.

At the time of submission, the medicine was not authorised anywhere in the EU for GNE myopathy.

This medicine had been designed orphan on 5 March 2012 for the treatment of hereditary inclusion body myopathy. At the request of the sponsor and having assessed the additional data submitted, the COMP adopted a positive opinion on 3 September 2015 recommending the orphan condition be changed to GNE myopathy, the new preferred name for hereditary inclusion body myopathy.

Orphan designation had been granted in the United States for hereditary inclusion body myopathy.

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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.

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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.

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Translations of the active ingredient and indication in all official EU languages[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Sialic acid	Treatment of GNE myopathy
Bulgarian	Сиалова киселина	Лечение на GNE миопатия
Croatian	Sialatna kiselina	Liječenje GNE miopatije
Czech	Kyselina sialová	Léčba geneticky podmíněné myopatie
Danish	Sialinsyre	Behandling af GNE-myopati
Dutch	Siaalzuur	Behandeling van GNE-myopathie
Estonian	Siaalhape	GNE müopaatia ravi
Finnish	Siaalihappo	GNE-myopatian hoito
French	Acide sialique	Traitement de la myopathie liée à GNE
German	Sialinsäure	Behandlung der GNE-Myopathie
Greek	Σιαλικό οξύ	Θεραπεία της μυοπάθειας GNE
Hungarian	Szialinsav	GNE-myopathia kezelése
Italian	Acido sialico	Trattamento della miopatia GNE
Latvian	Siālskābe	GNE miopātijas ārstēšana
Lithuanian	Sialo rūgštis	GNE miopatijos gydymas
Maltese	Aċidu Sialiku	Kura tal-mijopatija GNE
Polish	Kwas sialowy	Leczenie miopatii spowodowanej mutacją w genie GNE
Portuguese	Ácido siálico	Tratamento da miopatia GNE
Romanian	Acid sialic	Tratamentul miopatiei GNE
Slovak	Kyselina sialová	Liečba GNE myopatie
Slovenian	Sialična kislina	Zdravljenje GNE miopatije
Spanish	Ácido siálico	Tratamiento de la miopatía GNE
Swedish	Sialinsyra	Behandling av GNE-myopati
Norwegian	Sialinsyre	Behandling av GNE-myopati
Icelandic	Síalínsýra	Meðferð við GNE vöðvakvilla

[1] At the time of designation