

27 April 2016 EMA/COMP/172186/2016 Committee for Orphan Medicinal Products

## Public summary of opinion on orphan designation

N-acetyl-D-mannosamine monohydrate for the treatment of GNE myopathy

On 21 March 2016, orphan designation (EU/3/16/1635) was granted by the European Commission to Escala Therapeutics Ltd, United Kingdom, for N-acetyl-D-mannosamine monohydrate for the treatment of GNE myopathy.

#### What is GNE myopathy?

GNE myopathy is an inherited muscle-wasting disease. Patients with the disease have a mutation in a gene responsible for the GNE enzyme which is involved in the production of sialic acid. A lack of sialic acid is believed to be the cause of the wasting of muscles seen 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<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 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.



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<sup>&</sup>lt;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 513,700,000 (Eurostat 2016).

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#### How is this medicine expected to work?

The medicine consists of a sugar called N-acetyl-D-mannosamine monohydrate which is converted into sialic acid by the action of the GNE enzyme. In patients with GNE myopathy, GNE does not work properly and as a result not enough N-acetyl-D-mannosamine monohydrate is available for the production of sialic acid. Giving the medicine to patients with GNE myopathy is expected to increase the production of sialic acid. This is expected to allow the muscles to develop and function normally, relieving the symptoms of the condition.

#### 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, 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. Orphan designation of the medicine had been granted in the United States for hereditary inclusion body myopathy type 2 (another name for GNE myopathy).

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 18 February 2016 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

Sponsor's contact details:

Contact details of the current sponsor for this orphan designation can be found on EMA website, on the medicine's <u>rare disease designations page</u>.

For contact details of patients' organisations whose activities are targeted at rare diseases see:

- <u>Orphanet</u>, a database containing information on rare diseases, which includes a directory of patients' organisations registered in Europe;
- <u>European Organisation for Rare Diseases (EURORDIS)</u>, 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	N-acetyl-D-mannosamine monohydrate	Treatment of GNE myopathy
Bulgarian	N-ацетил-D-манозамин монохидрат	Лечение на GNE миопатия
Croatian	N-acetil-D-manozamin monohidrat	Liječenje GNE miopatije
Czech	N-acetyl-D-mannosamin monohydrát	Léčba geneticky podmíněné myopatie
Danish	N-acetyl-D-mannosamin monohydrat	Behandling af GNE-myopati
Dutch	N-acetyl-D-mannosaminemonohydraat	Behandeling van GNE-myopathie
Estonian	N-atsetüül-D-mannoosamiinmonohüdraat	GNE müopaatia ravi
Finnish	N-asetyyli-D-mannosamiini monohydraatti	GNE-myopatian hoito
French	N-acétyl-D-mannosamine monohydrate	Traitement de la myopathie liée à GNE
German	N-Acetyl-D-Mannosamin-Monohydrat	Behandlung der GNE-Myopathie
Greek	Ν-ακετυλ-D-μανοζαμίνη μονοϋδρική	Θεραπεία της μυοπάθειας GNE
Hungarian	N-acetil-D-mannózamin monohidrát	GNE-myopathia kezelése
Italian	N-acetil-D-mannosamina monoidrato	Trattamento della miopatia GNE
Latvian	N-acetil-D-mannozamīna monohidrāts	GNE miopātijas ārstēšana
Lithuanian	N-acetil-D-manozamino monohidratas	GNE miopatijos gydymas
Maltese	N-acetyl-D-mannosamine monohydrate	Kura tal-mijopatija GNE
Polish	N-acetylo-D-mannozamina jednowodna	Leczenie miopatii spowodowanej mutacją w genie GNE
Portuguese	N-acetil-D-manosamina mono hidratada	Tratamento da miopatia GNE
Romanian	N-acetil-D-manozamină monohidrat	Tratamentul miopatiei GNE
Slovak	N-acetyl-D-manozamín monohydrát	Liečba GNE myopatie
Slovenian	N-acetil-D-manozamin monohidrat	Zdravljenje GNE miopatije
Spanish	N-acetil-D-manosamina monohidratada	Tratamiento de la miopatía GNE
Swedish	N-acetyl-D-mannosaminmonohydrat	Behandling av GNE-myopati
Norwegian	N-acetyl-D-mannosamin-monohydrat	Behandling av GNE-myopati
Icelandic	N-asetýl-D-mannósamín einhýdrat	Meðferð við GNE vöðvakvilla

<sup>&</sup>lt;sup>1</sup> At the time of designation