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Public summary of opinion on orphan designation

20-hydroxyecdysone for treatment of Duchenne muscular dystrophy

On 27 June 2018, orphan designation (EU/3/18/2030) was granted by the European Commission to Biophytis, France, for 20-hydroxyecdysone for treatment of Duchenne muscular dystrophy.

What is Duchenne muscular dystrophy?

Duchenne muscular dystrophy (DMD) is a genetic disease that gradually causes weakness and atrophy (wasting) of muscles. It mainly affects boys, and usually starts before the age of six years. The muscle weakness usually starts in the hips and legs, before affecting the arms, chest and the heart. Patients with DMD lack normal dystrophin, a protein found in muscles. Because this protein helps to protect muscles from injury as muscles contract and relax, in patients with DMD the muscles become weaker and eventually stop working.

DMD causes long-term disability and is life threatening because of its effects on the heart and the respiratory muscles (muscles that are used to breathe). The disease usually leads to death in early adulthood.

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

At the time of designation, DMD affected less than 0.5 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 26,000 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 Translarna (ataluren) was authorised in the EU for the treatment of a small group of patients with DMD caused by a particular type of mutation (change), called a nonsense mutation, in the dystrophin gene. Patients also received supportive treatment such as physiotherapy.

*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 517,400,000 (Eurostat 2018).

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The sponsor has provided sufficient information to show that 20-hydroxyecdysone might be of significant benefit for patients with DMD because laboratory data indicate that the product may have positive effects in a wider patient population than with the authorised treatment.

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?

20-hydroxyecdysone is thought to activate a receptor (target) on muscle cells called MAS. This in turn results in a reduced production of a protein called myostatin, which blocks muscle growth. By reducing myostatin, the medicine is thought to reduce muscle wasting in patients with DMD.

What is the stage of development of this medicine?

The effects of 20-hydroxyecdysone have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with 20-hydroxyecdysone in patients with DMD had been started.

At the time of submission, 20-hydroxyecdysone was not authorised anywhere in the EU for DMD 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 24 May 2018 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[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	20-hydroxyecdysone	Treatment of Duchenne muscular dystrophy
Bulgarian	20-хидроксиекдизон	Лечение на мускулна дистрофия на Duchenne
Croatian	20-hidroksiekdison	Liječenje Duchenneove mišićne distrofije
Czech	20-hydroxyekdyson	Léčba pacientů s Duchennovou muskulární dystrofií
Danish	20-hydroxyecdysone	Behandling af Duchenne muskeldystrofi
Dutch	20-hydroxyecdysone	Behandeling van Duchenne spierdystrofie
Estonian	20-hüdroksüekdüsoon	Duchenne'i lihasdüstroofia ravi
Finnish	20-hydroksiekdysoni	Duchennen lihasdystrofian hoito
French	20-hydroxyecdysone	Traitement de la dystrophie musculaire de Duchenne
German	20-Hydroxyecdysone	Behandlung der Duchenne-Muskeldystrophie
Greek	20-υδροξυεκδυσόνη	Θεραπεία της μυϊκής δυστροφίας Duchenne
Hungarian	20-hidroxyekdizon	Duchenne dystrophia kezelése
Italian	20-idrossiecdisone	Trattamento della distrofia muscolare di tipo Duchenne
Latvian	20-hidroksiekdizons	Dišēna muskuļu distrofijas ārstēšana
Lithuanian	20-hidroksiekdisonas	Duchenne (Diušeno) raumenų distrofijos gydymas
Maltese	20-idrossiekdison	Kura tad-distrofija muskolari tat-tip Duchenne
Polish	20-hydroxyekdyson	Leczenie zaniku mięśni typu Duchenne'a
Portuguese	20-hidroxiecdisona	Tratamento da distrofia muscular de Duchenne
Romanian	20-hidroxiecdisonă	Tratamentul distrofiei musculare Duchenne
Slovak	20-hydroxyekdyzón	Liečba Duchennovej muskulárnej dystrofie
Slovenian	20-hidroksiekdizon	Zdravljenje Duchennove mišične distrofije
Spanish	20-hidroxiecdison	Tratamiento de la distrofia muscular de Duchenne
Swedish	20-hydroxyecdyson	Behandling av Duchennes muskeldystrofi
Norwegian	20-hydroksyekdyson	Behandling av Duchennes muskeldystrofi
Icelandic	20-hydroxyecdyson	Meðferð á Duchenne vöðvarýrnun

[1] At the time of designation