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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Adeno-associated viral vector serotype 9 containing the human mini-dystrophin gene for the treatment of Duchenne muscular dystrophy

On 29 August 2016, orphan designation (EU/3/16/1716) was granted by the European Commission to Advanced Biotherapeutics Consulting SARL, France, for adeno-associated viral vector serotype 9 containing the human mini-dystrophin gene for the treatment of Duchenne muscular dystrophy.

What is Duchenne muscular dystrophy?

Duchenne muscular dystrophy (DMD) is a serious genetic disease that gradually causes weakness and atrophy (wasting) of the muscles. It mainly affects boys, and is usually diagnosed 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 adolescence or early adulthood.

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

At the time of designation, DMD affected approximately 0.5 in 10,000 people in the European Union (EU). This was equivalent to a total of around 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).

*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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What treatments are available?

At the time of designation, Translarna (ataluren) was authorised in the EU to treat patients with DMD who have a specific type of mutation (change) called a nonsense mutation in their dystrophin gene. Patients also received supportive treatment such as physiotherapy.

The sponsor has provided sufficient information to show that this medicine might be of significant benefit for patients with DMD because laboratory studies indicate that it can restore muscle strength, it works in a different way to Translarna and it would not be limited to patients with a specific mutation. 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?

Duchenne muscular dystrophy is caused by mutations (changes) in the gene for the protein dystrophin, which lead to the production of a non-functional dystrophin. The medicine consists of a virus that contains a shortened, but functional version of the dystrophin gene. When given to the patient, it is expected that the virus will carry the short dystrophin gene into muscle cells, enabling them to produce a short, but functional version of the dystrophin protein. This is expected to help the muscle cells work better, improving symptoms of the condition.

The virus used in this medicine (adeno-associated viral vector) does not cause disease in humans.

What is the stage of development of this medicine?

At the time of submission of the application for orphan designation, the evaluation of the effects of the medicine in experimental models was ongoing.

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

At the time of submission, the medicine 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 13 July 2016 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	Adeno-associated viral vector serotype 9 containing the human mini-dystrophin gene	Treatment of Duchenne muscular dystrophy
Bulgarian	Аденосвързан вирусен вектор серотип 9, съдържащ човешкия мини дистрофин ген	Лечение на мускулна дистрофия на Duchenne
Croatian	Adeno-povezani virusni vektor serotipa 9 koji sadrži ljudski mini-distrofin gen	Liječenje Duchenneove mišićne distrofije
Czech	Sérotyp 9 adeno asociovaný virový vektor, obsahující lidský mini-dystrofinového gen	Léčba pacientů s Duchennovou muskulární dystrofií
Danish	Adenoassocieret viral vektor serotype 9 indeholdende det humane mini-dystrophin gen	Behandling af Duchenne muskeldystrofi
Dutch	Adenogeassocieerde virale vector, serotype 9, welke het humane mini-dystrofine gen bevat	Behandeling van Duchenne spierdystrofie
Estonian	Adenoviirusega seotud viirusvektori serotüüp 9, mis sisaldab inimese mini-düstrofiini geeni	Duchenne'i lihasdüstroofia ravi
Finnish	Adenoassosioitu virusvektori, serotyyppi 9, joka sisältää ihmisen mini-dystrofiinigeenin	Duchennen lihasdystrofian hoito
French	Vecteur viral adéno-associé de sérotype 9 contenant le gène humain de la mini-dystrophine	Traitement de la dystrophie musculaire de Duchenne
German	Das humane mini-Dystrophin Gen beinhaltender, adeno-assoziierter viraler Vektor Serotyp 9	Behandlung der Duchenne-Muskeldystrophie
Greek	Αδενο-σχετιζόμενος ιικός φορέας ορότυπου 9 που περιέχει το ανθρώπινο γονίδιο της mini-δυστροφίνης	Θεραπεία της μυϊκής δυστροφίας Duchenne
Hungarian	Humán mini disztrofin gént tartalmazó 9-es szerotípusú adeno-asszociált vírus vector	Duchenne dystrophia kezelése
Italian	Vettore virale adeno-associato del serotipo 9 contenente il gene umano della mini-distrofina	Trattamento della distrofia muscolare di tipo Duchenne
Latvian	Adeno-saistītā vīrālā vektora 9. serotips, kas satur cilvēka mini-distrofīna gēnu	Dišēna muskuļu distrofijas ārstēšana
Lithuanian	Adeno-asocijuoto viruso vektoriaus 9 serotipas, pernešantis žmogaus mini-distrofino geną	Duchenne (Diušeno) raumenų distrofijos gydymas
Maltese	Vettur imnissel mill-adenovirus tas-serotip 9 li fih il-ġene ta' mini-distrofina uman	Kura tad-distrofija muskolari tat-tip Duchenne
Polish	Wektor adenowirusowy serotypu 9 zawierający gen ludzkiej minidystrofiny	Leczenie zaniku mięśni typu Duchenne'a
Portuguese	Vetor viral adeno-associado de serotipo 9 contendo o gene humano da mini-distrofina	Tratamento da distrofia muscular de Duchenne
Romanian	Vector viral adeno-asociat de serotip 9 conţinând gena umană a mini-distrofinei	Tratamentul distrofiei musculare Duchenne
Slovak	Adeno-asociovaný vírusový vektor sérotypu 9 obsahujúci ľudský mini-dystrofínový gén	Liečba Duchennovej muskulárnej dystrofie

[1] At the time of designation

Language	Active ingredient	Indication
Slovenian	Adeno-pridruženi virusni vektor serotipa 9, ki vsebuje človeški gen mini distrofina	Zdravljenje Duchennove mišične distrofije
Spanish	Vector viral adenoasociado del serotipo 9 que conteniene el gene humano de la mini-distrofina	Tratamiento de la distrofia muscular de Duchenne
Swedish	Adenoassocierad virusvektor serotyp 9, innehållande den mänskliga mini-dystrofingenen	Behandling av Duchennes muskeldystrofi
Norwegian	Adenoassosiert virusvektor serotype 9 som inneholder det humane mini-dystrofingenet	Behandling av Duchennes muskeldystrofi
Icelandic	Adenótengd veiruferja af sermisgerð 9 sem inniheldur manna míni-dýstróphín gen	Meðferð á Duchenne vöðvarýrnun