7 November 2016
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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant adeno-associated viral vector encoding a human micro-dystrophin gene under the control of a muscle specific promoter for the treatment of Duchenne muscular dystrophy

On 14 October 2016, orphan designation (EU/3/16/1759) was granted by the European Commission to Pharma Gateway AB, Sweden, for recombinant adeno-associated viral vector encoding a human micro-dystrophin gene under the control of a muscle specific promoter (also known as SGT-001) for the 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 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).

*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, the medicine Translarna (ataluren) was authorised in the EU for the treatment of a small group of patients with DMD due to a particular type of mutation, called a nonsense mutation, in the dystrophin gene. Patients also received supportive treatment such as physiotherapy.

The sponsor has provided sufficient information to show that the medicine might be of significant benefit for patients with DMD because data in experimental models show that it can potentially be used in a wider group of patients than the currently 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?

The medicine consists of a virus that contains a copy of the microdystrophin gene. This gene produces 'microdystrophin', a protein that works like dystrophin but is smaller. When given by injection to the patient, it is expected that the virus will carry the gene into the muscle cells, enabling them to produce the microdystrophin protein. This is then expected to reduce muscle wasting, slowing down progression of the condition.

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

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, 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. Orphan designation of the medicine has been granted in United States. for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 8 September 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	Recombinant adeno-associated viral vector encoding a human micro-dystrophin gene under the control of a muscle specific promoter	Treatment of Duchenne muscular dystrophy
Bulgarian	Рекомбинантен адено-свързан вирусен вектор, кодиращ човешкия микро-дистрофин ген под контрола на мускулно-специфичен промотор	Лечение на мускулна дистрофия на Duchenne
Croatian	Rekombinantni adeno-povezani virusni vektor koji kodira ljudski gen mikrodistrofin pod kontrolom za mišić specifičnog promotora	Liječenje Duchenneove mišićne distrofije
Czech	Rekombinantní adeno-asociovaný virový vektor kódující lidský mikro-dystrofin gen pod kontrolou svalově specifického promotoru	Léčba pacientů s Duchennovou muskulární dystrofií
Danish	Rekombinant adenoassocieret viral vektor, der koder for et humant mikrodystrofin gen under kontrol af en muskelspecifik promotor	Behandling af Duchenne muskeldystrofi
Dutch	Recombinant adeno-geassocieerde virale vector coderend voor een humaan microdystrofine gen onder controle van een spierspecifieke promoter	Behandeling van Duchenne spierdystrofie
Estonian	Lihasespetsiifilise promootori kontrolli all olev inimese mikrodüstrofiini geeni kodeeriv rekombinantne adenoviirusega seotud viirusvektor	Duchenne'i lihasdüstroofia ravi
Finnish	Rekombinantti adeno- assosioitu virusvektori, joka koodaa ihmisen mikrodystrofiinia geeni lihasspesifisen promoottorin säätelemänä	Duchennen lihasdystrofian hoito
French	Vecteur viral recombinant adéno-associé d'un gène codant pour une microdystrophine humaine sous le contrôle d'un promoteur musculaire spécifique	Traitement de la dystrophie musculaire de Duchenne
German	Rekombinanter adeno-assoziierter viraler Vektor, der für ein humanes Mikrodystrophin Gen unter der Kontrolle eines muskelspezifischen Promoters kodiert	Behandlung der Duchenne-Muskeldystrophie
Greek	Ανασυνδυασμένος αδενο-σχετιζόμενος ιϊκός φορέας που κωδικοποιεί ένα ανθρώπινο γονίδιο μικροδυστροφίνης υπό τον έλεγχο ενός ειδικού μυϊκού εκκινητή	Θεραπεία της μυϊκής δυστροφίας Duchenne
Hungarian	Izom-specifikus promoterrel kontrollált humán mikro-disztrofin génjét kódoló, rekombináns adeno-asszociált vírus vektor	Duchenne dystrophia kezelése
Italian	Vettore virale ricombinante adeno-associato codificante un gene per la microdistrofina umana sotto il controllo di un promotore muscolo-specifico	Trattamento della distrofia muscolare di tipo Duchenne
Latvian	Rekombinants adeno-asociētais vīrusa vektors, kas kodē cilvēka mikrodistrofīna gēnu, kuru kontrolē muskuļu specifiskais promoters	Dišēna muskuļu distrofijas ārstēšana

[1] At the time of designation

Language	Active ingredient	Indication
Lithuanian	Rekombinantinis su adeno virusu susijęs vektorius, koduojantis žmogaus mikrodistrofino geną, kontroliuojamą raumenims specifinio promotoriaus	Duchenne (Diušeno) raumenų distrofijos gydymas
Maltese	Vettur rikombinanti mnissel mill-adenovirus li jikkodifika il-ġene micro-dystrophin uman taħt il-kontroll ta' promotur speċifiku għal muskolu	Kura tad-distrofija muskolari tat-tip Duchenne
Polish	Rekombinowany, związany z adenowirusami, wirusowy wektor kodujący ludzki gen mikrodystrofiny pod kontrolą promotora swoistego dla mięśni	Leczenie zaniku mięśni typu Duchenne'a
Portuguese	Vetor viral recombinante adeno-associado que codifica um gene da microdistrofina humana sob o controlo de um promotor específico do músculo.	Tratamento da distrofia muscular de Duchenne
Romanian	Vector viral adeno-asociat recombinant care codifică o genă a micro-distrofinei umane sub controlul unui promotor muscular specific	Tratamentul distrofiei musculare Duchenne
Slovak	Rekombinantný adeno-asociovaný vírusový vektor kódujúci ľudský gén pre mikrodystrofín kontrolovaný svalovo špecifickými promotérmi	Liečba Duchennovej muskulárnej dystrofie
Slovenian	Rekombinantni adenovirusom pridruženi virusni vektor, kodiran za humani mikro-distrofin pod nadzorom za mišico specifičnega promotorja	Zdravljenje Duchennove mišične distrofije
Spanish	Vector vírico adenosaociado recombinante que codifica un gene de microdistrofina humana bajo el control de un promotor específico de músculo	Tratamiento de la distrofia muscular de Duchenne
Swedish	Rekombinant adenoassocierad viral vektor som kodar för en human mikrodystrofingen under kontroll av en muskelspecifik promotor	Behandling av Duchennes muskeldystrofi
Norwegian	Rekombinant adenoassosiert viral vektor som koder for et humant mikrodystrofingen under kontroll av en muskelspesifikk promotor	Behandling av Duchennes muskeldystrofi
Icelandic	Raðbrigða adenótengd veiruferja sem kóðar fyrir manna míkródystrófín geni sem stjórnað er af vöðvasérstæku stýriefni í vöðvum	Meðferð á Duchenne vöðvarýrnun