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Public summary of opinion on orphan designation

Adeno-associated virus serotype rh74 containing the human micro-dystrophin gene for the treatment of Duchenne muscular dystrophy

On 28 February 2020, orphan designation EU/3/20/2250 was granted by the European Commission to Sarepta Therapeutics Ireland Limited, Ireland, for adeno-associated virus serotype rh74 containing the human micro-dystrophin gene (also known as SRP-9001) for the treatment of Duchenne muscular dystrophy.

What is Duchenne muscular dystrophy?

Duchenne muscular dystrophy is a genetic disease that causes increasing weakness and atrophy (wasting) of muscles. It mainly affects boys, and usually starts before the age of 6 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 the disease the muscles become progressively weaker and eventually stop working.

Duchenne muscular dystrophy 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, Duchenne muscular dystrophy 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 Duchenne muscular dystrophy caused by a particular type

*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, Norway, Iceland and Liechtenstein. This represents a population of 519,200,000 (Eurostat 2020).

Official address Domenico Scarlattilaan 6 ● 1083 HS Amsterdam ● The Netherlands
Address for visits and deliveries Refer to www.ema.europa.eu/how-to-find-us
Send us a question Go to www.ema.europa.eu/contact **Telephone** +31 (0)88 781 6000 An agency of the European Union

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of mutation (change), 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 Duchenne muscular dystrophy because early studies showed that the condition improved in children treated with the medicine. Also, it has the potential to work in patients with all forms of the dystrophin gene mutation. By contrast, Translarna is for use in patients with only one form of 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?

The medicine is made of a virus that contains genetic material for producing a shortened, but working, form of dystrophin. The medicine is designed to introduce the genetic material into muscles and the heart. A single injection is expected to enable the patient to produce a working form of dystrophin and so slow down progression of the disease.

The type of 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, clinical trials with the medicine in patients with Duchenne muscular dystrophy were ongoing.

At the time of submission, the medicine was not authorised anywhere in the EU for the treatment of Duchenne muscular dystrophy. Orphan designation of the medicine had been granted in the United States for Duchenne muscular dystrophy.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 22 January 2020, 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.

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 virus serotype rh74 containing the human micro-dystrophin gene	Treatment of Duchenne muscular dystrophy
Bulgarian	Адено-свързан вирус серотип rh74, съдържащ гена на човешкия микро-дистрофин	Лечение на мускулна дистрофия на Duchenne
Croatian	Adeno-povezani virus serotipa rh74 koji sadrži ljudski gen mikrodistrofin	Liječenje Duchenneove mišićne distrofije
Czech	Adeno-asociovaný virus sérotypu rh74 obsahující lidský gen pro mikrodystrofin	Léčba pacientů s Duchennovou muskulární dystrofií
Danish	Adeno-associeret virus serotype rh74 indeholdende det humane mikro-dystrofin-gen	Behandling af Duchenne muskeldystrofi
Dutch	Adeno-geassocieerd virus serotype rh74 welke het humane micro-dystrofine-gen bevat	Behandeling van Duchenne spierdystrofie
Estonian	Inimese mikrodüstrofiini geeni sisaldav adeno-assotsieerunud viiruse serotüüp rh74	Duchenne'i lihasdüstroofia ravi
Finnish	Adeno-assosioitunut serotyypin rh74 virus, joka sisältää ihmisen mikrodystrofiinigeenin	Duchennen lihasdystrofian hoito
French	Virus adéno-associé de sérotype rh74 contenant le gène de la micro-dystrophine humaine	Traitement de la dystrophie musculaire de Duchenne
German	Adeno-assoziierter Virus vom Serotyp rh74, das das humane Micro-Dystrophin Gen trägt	Behandlung der Duchenne-Muskeldystrophie
Greek	Αδενο-σχετιζόμενος ιός οροτύπου rh74 που περιέχει το ανθρώπινο γονίδιο μικροδυστροφίνης	Θεραπεία της μυϊκής δυστροφίας Duchenne
Hungarian	Humán mikro-disztrofin gént tartalmazó adeno-asszociált vírus rh74 szerotípusa	Duchenne dystrophia kezelése
Italian	Virus adeno-associato di serotipo rh74 contenente il gene della micro-distrofina umana	Trattamento della distrofia muscolare di tipo Duchenne
Latvian	Cilvēka mikrodistrofīna gēnu saturošs adenoasociētais vīrusa serotips rh74	Dišēna muskuļu distrofijas ārstēšana
Lithuanian	Su adeno virusu susijęs serotipas rh74, turintis žmogaus mikrodistrofino geną	Duchenne (Diušeno) raumenų distrofijos gydymas

[1] At the time of designation

Language	Active ingredient	Indication
Maltese	Serotip ta' virus rh74 adeno-assoċjat li fih il-ġene tal-bniedem mikro-distrofin	Kura tad-distrofija muskolari tat-tip Duchenne
Polish	Wirus zależny od adenowirusów o serotypie rh74 zawierający gen ludzkiej mikrodystrofiny	Leczenie zaniku mięśni typu Duchenne'a
Portuguese	Vírus adeno-associado de serotipo rh74 contendo o gene da microdistrofina humana	Tratamento da distrofia muscular de Duchenne
Romanian	Virus adeno-asociat de serotip rh74, ce conţine gena micro-distrofinei umane	Tratamentul distrofiei musculare Duchenne
Slovak	Adeno-asociovaný vírus sérotypu rh74 obsahujúci gén ľudského mikrodystrofínu	Liečba Duchennovej muskulárnej dystrofie
Slovenian	Serotip adenovirusa rh74 s človeškim mikrodistrofinskim genom	Zdravljenje Duchennove mišične distrofije
Spanish	Virus adenoasociado serotipo rh74 que contiene el gen humano de la microdistrofina	Tratamiento de la distrofia muscular de Duchenne
Swedish	Adeno-associerad virusserotyp rh74 som innehåller den humana mikrodystrofingenen	Behandling av Duchennes muskeldystrofi
Norwegian	Adenoassosiert virus serotype rh74 som inneholder genet for humant mikrodystrofin	Behandling av Duchennes muskeldystrofi
Icelandic	Adenótengd veira af sermisgerð rh74 sem inniheldur manna míkró-dystrófín genið	Meðferð á Duchenne vöðvarýrnun