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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Adeno-associated viral vector containing modified U1 snRNA for the treatment of Duchenne muscular dystrophy

On 8 October 2009, orphan designation (EU/3/09/663) was granted by the European Commission to Amsterdam Molecular Therapeutics BV, the Netherlands, for adeno-associated viral vector containing modified U1 snRNA for the treatment of Duchenne muscular dystrophy.

In July 2012, Amsterdam Molecular Therapeutics BV changed name to uniQure biopharma B.V.

What is Duchenne muscular dystrophy?

Duchenne muscular dystrophy (DMD) is an inherited genetic disease characterised by progressive weakening of the muscles. It mainly affects boys, usually before the age of six years. The muscle weakness usually starts in the hips and legs, before reaching the chest, arms, and possibly the heart. DMD is caused by abnormalities in the gene that is responsible for the production of a protein called dystrophin. As a result, patients with DMD do not have enough of this protein. As dystrophin is an important component of muscle fibres, the muscles of patients with DMD cannot grow, so they become weak 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).

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

At the time of designation, DMD affected approximately 0.3 in 10,000 people in the European Union (EU). This was equivalent to a total of around 15,000 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 27), Norway, Iceland and Liechtenstein.
At the time of designation, this represented a population of 504,800,000 (Eurostat 2009).

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What treatments are available?

At the time of designation, there were no treatments available in the EU that could cure DMD. Physiotherapy is used to relieve symptoms and improve the patient's general condition. In addition, corticosteroids are used in an attempt to improve symptoms, although they are not specifically authorised for use in this disease.

How is this medicine expected to work?

In DMD, the lack of dystrophin is caused by the gene for dystrophin containing abnormal regions that affect the way the dystrophin protein is produced. Adeno-associated viral vector containing modified U1 snRNA is made up of the capsid (shell) of a virus containing genetic material that has been modified to enable cells to skip certain parts of their genes when they make proteins. The medicine is expected to be given to patients directly into the blood, so that the virus can carry the genetic material into the muscle cells, including the heart and respiratory muscles. This is expected to enable the cell to skip the abnormal parts of the dystrophin gene, so that the cell can produce a shorter version of the protein that is still able to work in the same way as normal dystrophin.

The type of virus used in this medicine (an adeno-associated virus) is modified so that it does not cause disease in humans.

What is the stage of development of this medicine?

The effects of adeno-associated viral vector containing modified U1 snRNA have been evaluated in experimental models.

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

At the time of submission, adeno-associated viral vector containing modified U1 snRNA 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 8 July 2009 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:

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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 containing modified U1 snRNA	Treatment of Duchenne muscular dystrophy
Bulgarian	Адено-асоцииран вирусен вектор съдържащ модифицирана U1 малка некодираща РНК (snRNA)	Лечение на мускулна дистрофия на Duchenne
Czech	Adeno asociovaný virový vektor obsahující modifikovaný gen U1 snRNA	Léčba pacientů s Duchennovou muskulární dystrofií
Danish	Adenoassocieret viral vektor indeholdende et modificeret U1 snRNA-gen	Behandling af Duchenne muskeldystrofi
Dutch	Adenogeassocieerde virale vector met een gemodificeerd gen U1 snRNA	Behandeling van Duchenne spierdystrofie
Estonian	Adenoga assotsieeruv viirusvektor, mis sisaldab muudetud U1 snRNA geeni	Duchenne'i lihasdüstroofia ravi
Finnish	Adenoassosioitu virusvektori, joka sisältää muunnetun U1 snRNA-geenin	Duchennen lihasdystrofian hoito
French	Vecteur viral adéno-associé contenant un gène U1 snRNA modifié	Traitement de la dystrophie musculaire de Duchenne
German	Adeno-assoziierte virale Vektoren, welche ein modifiziertes U1-snRNA Gen enthalten	Behandlung der Duchenne-Muskeldystrophie
Greek	Αδενοσύνδετος φορέας ιού περιέχων τροποποιημένο γονίδιο U1 snRNA	Θεραπεία της μυϊκής δυστροφίας Duchenne
Hungarian	Modósított U1-snRNA gént hordozó adeno-társított vírus vektor	Duchenne dystrophia kezelése
Italian	Vettori virali associati all'adenovirus contenenti la versione modificata del gene U1-snRNA	Trattamento di distrofia muscolare di tipo Duchenne
Latvian	Adeno-saistīts vīrusu vektors, kas satur modificētu U1 snRNA gēnu	Dišēna muskuļu distrofijas ārstēšana
Lithuanian	Adeno-asocijuoto viruso vektorius su modifikuotu U1 sn RNR genu	Duchenne (Diušeno) raumenų distrofijos gydymas
Maltese	Vettur imnissel mill-adenovirus li fih ġene U1 snRNA immodifikat	Kura tad-distrofija muskolari tat-tip Duchenne
Polish	Wektor sprzężony z adenowirusem zawierający zmodyfikowany gen U1 snRNA	Leczenie zaniku mięśni typu Duchenne'a
Portuguese	Vector viral adeno-associado contendo um gene U1 snRNA modificado	Tratamento da distrofia muscular de Duchenne
Romanian	Vector viral asociat adenovirusului conţinând gena modificată ARNsn U1	Tratamentul distrofiei musculare Duchenne
Slovak	Adeno-asociovaný vírusový vektor obsahujúci modifikovaný gén U1 snRNA	Liečba Duchennovej muskulárnej dystrofie

[1] At the time of designation

Language	Active ingredient	Indication
Slovenian	Adenovirusni pridruženi virusni vektor z modificiranim genom U1 snRNA	Zdravljenje Duchennove mišične distrofije
Spanish	Vector viral adenoasociado que contiene un gen U1 snRNA modificado	Tratamiento de la distrofia muscular de Duchenne
Swedish	Adenoassocierad virusvektor innehållande en modifierad U1 snRNA-gen	Behandling av Duchennes muskeldystrofi
Norwegian	Adenoassosiert virusvektor som inneholder et modifisert U1 snRNA-gen	Behandling av Duchennes muskeldystrofi
Icelandic	Adenotengd veiruferja sem inniheldur breytt U1 snRNA gen	Meðferð á Duchenne vöðvarýrnun