European Medicines Agency
Pre-authorisation Evaluation of Medicines for Human Use

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COMMITTEE FOR ORPHAN MEDICINAL PRODUCTS

PUBLIC SUMMARY OF POSITIVE OPINION FOR ORPHAN DESIGNATION OF adeno-associated viral vector containing modified U7 snRNA gene for the treatment of Duchenne muscular dystrophy

On 27 July 2005, orphan designation (EU/3/05/297) was granted by the European Commission to Généthon, France, for adeno-associated viral vector containing modified U7 snRNA gene for the treatment of Duchenne muscular dystrophy.

What is Duchenne muscular dystrophy?
Duchenne muscular dystrophy is an inherited genetic disease with onset usually before the age of 6. It is characterised by symmetrical progressive diminishing and weakness of the muscles, first at the height of the pelvis and legs, later on also the muscles of the chest and arms are involved. Genes located on structures present in each cell of the body (the chromosomes), carry the genetic information that determines the characteristics of each individual. In humans, the so-called X and Y-chromosomes determine the sex, but carry also other genetic information. Duchenne muscular dystrophy is caused by an abnormality of a gene located on the X chromosome. This gene is responsible for the production of a protein, dystrophin, in the muscle cells. This means that patients suffering from this condition do not produce the dystrophin protein or produce a non-functional dystrophin. As boys, contrary to girls, only have one single copy of chromosome X, thus one single copy of dystrophin gene, they have much higher pobabilities of suffering from Duchenne muscular distrophy.
Duchenne muscular dystrophy is chronically debilitating and life-threatening.

What are the methods of treatment available?
At the time of submission of the application for orphan designation, no satisfactory method had been authorised in the European Union for treatment of the condition. Treatment of patients with Duchenne muscular dystrophy primarily involves physiotherapy as supportive treatments.

What is the estimated number of patients affected by the condition*?
According to the information provided by the sponsor, Duchenne muscular dystrophy was considered to affect about 23,000 persons in the European Union.

How is this medicinal product expected to act?
Adeno-associated viral vector containing modified U7 snRNA gene is a medicinal product, which in theory might overcome the abnormality in the production of dystrophin. The product would allow skipping the abnormal parts of the gene responsible for the lack of functional dystrophin production. Thus it could enable the production of shorter versions of the dystrophin protein, but still capable of ensuring the same functions as the normal full-length dystrophin.

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What is the stage of development of this medicinal product?
The evaluation of the effects of adeno-associated viral vector containing modified U7 snRNA gene in experimental models is ongoing.

At the time of submission of the application for orphan designation, no clinical trials in patients with Duchenne muscular dystrophy were initiated.

Adeno-associated viral vector containing modified U7 snRNA gene was not marketed anywhere worldwide for Duchenne muscular dystrophy, at the time of submission.

According to Regulation (EC) No 141/2000 of 16 December 1999, the Committee for Orphan Medicinal Products (COMP) adopted on 15 June 2005 a positive opinion recommending the grant of the above-mentioned designation.

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Opinions on orphan medicinal products designations are based on the following cumulative criteria: (i) the seriousness of the condition, (ii) the existence or not of alternative methods of diagnosis, prevention or treatment and (iii) either the rarity of the condition (considered to affect not more than five in ten thousand persons in the Community) or the insufficient return of development investments.

Designated orphan medicinal products are still investigational products which were considered for designation on the basis of potential activity. An orphan designation is not a marketing authorisation. As a consequence, demonstration of the quality, safety and efficacy will be necessary before this product can be granted a marketing authorisation.

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*Disclaimer: For the purpose of the designation, the number of patients affected by the condition is estimated and assessed based on data from the European Union (EU 25), Norway, Iceland and Lichtenstein. This represents a population of 459,700,000 (Eurostat 2004). This estimate is based on available information and calculations presented by the sponsor at the time of the application.

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Translations of the active ingredient and indication in all EU languages and Norwegian and Icelandic

Language	Active Ingredient	Indication
English	Adeno-associated viral vector containing a modified U7-snRNA gene	Treatment of Duchenne muscular dystrophy
Czech	Adeno asociovaný virový vektor obsahující modifikovaný gen U7 snRNA	Léčba pacientů s Duchennovou muskulární dystrofií
Danish	Adenoassocieret viral vektor indeholdende et modificeret U7 snRNA-gen	Behandling af Duchenne muskeldystrofi
Dutch	Adenogeassocieerde virale vector met een gemodificeerd gen U7 snRNA	Behandeling van Duchenne spierdystrofie
Estonian	Adenoga assotsieeruv viirusvektor, mis sisaldab muudetud U7 snRNA geeni	Duchenne'i lihasdüstroofia ravi
Finnish	Adenoassosioitu virusvektori, joka sisältää muunnetun U7 snRNA-geenin	Duchennen lihasdystrofian hoito
French	Vecteur viral adéno-associé contenant un gène U7 snRNA modifié	Traitement de la dystrophie musculaire de Duchenne
German	Adeno-assoziierte virale Vektoren, welche ein modifiziertes U7-snRNA Gen enthalten	Behandlung von Duchenne-Muskeldystrophie
Greek	Αδενοσύνδετος φορέας ιού περιέχων τροποποιημένο γονίδιο U7 snRNA	Θεραπεία της μυϊκής δυστροφίας Duchenne
Hungarian	Modósított U7-snRNA gént hordozó adeno-társított vírus vektor	Duchenne dystrophia a kezelése
Italian	Vettori virali associati all'adenovirus contenenti la versione modificata del gene U7-snRNA	Trattamento di distrofia muscolare di tipo Duchenne
Latvian	Adeno-saistīts vīrusu vektors, kas satur modificētu U7 snRNA gēnu	Dušēna muskuļu distrofijas ārstēšana
Lithuanian	Adeno-asocijuoto viruso vektorius su modifikuotu U7 sn RNR genu	Duchenne (Diušeno) raumenų distrofijos gydymas
Polish	Wektor sprzężony z adenowirusem zawierający zmodyfikowany gen U7 snRNA	Leczenie zaniku mięśni typu Duchenne'a
Portuguese	Vector viral adeno-associado contendo um gene U7 snRNA	Tratamento da distrofia muscular de Duchenne

	modificado	
Slovak	Adeno-asociovaný vírusový vektor obsahujúci modifikovaný gén U7 snRNA	Liečba Duchenneovej muskulárnej dystrofie
Slovenian	Adenovirusni pridruženi virusni vektor z modificiranim genom U7 snRNA	Zdravljenje Duchennove mišične distrofije
Spanish	Vector viral adenoasociado que contiene un gen U7 snRNA modificado	Tratamiento de la distrofia muscular de Duchenne
Swedish	Adenoassocierad virusvektor innehållande en modifierad U7 snRNA-gen	Behandling av Duchennes muskeldystrofi (DMD)
Norwegian	Adenoassosiert virusvektor som inneholder et modifisert U7 snRNA-gen	Behandling av Duchennes muskeldystrofi
Icelandic	Adenotengd veiruferja sem inniheldur breytt U7 snRNA gen	Meðferð á Duchenne vöðvarýrnun