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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Adeno-associated viral vector containing the human alpha sarcoglycan gene for the treatment of alpha sarcoglycanopathy

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Disclaimer Please note that revisions to the Public Summary of Opinion are purely administrative updates. Therefore, the scientific content of the document reflects the outcome of the Committee for Orphan Medicinal Products (COMP) at the time of designation and is not updated after first publication.	

Please note that this product was withdrawn from the Community Register of designated Orphan Medicinal Products in May 2014 on request of the Sponsor.

On 7 November 2008, orphan designation (EU/3/08/573) was granted by the European Commission to Généthon, France, for adeno-associated viral vector containing the human alpha sarcoglycan gene for the treatment of alpha sarcoglycanopathy.

What is alpha sarcoglycanopathy?

Alpha sarcoglycanopathy is an inherited disease, which causes progressive muscle weakness. It usually appears in childhood. Patients with alpha sarcoglycanopathy cannot produce a protein called alpha sarcoglycan in their muscles. This protein is needed for the muscles to work properly. The disease is due to an abnormal gene that is found on chromosome 17. For a patient to develop this disease, he or she needs to have inherited one copy of the abnormal gene from each parent. This is called 'autosomal recessive' transmission.

Alpha sarcoglycanopathy is a chronically debilitating and life-threatening disease.

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

At the time of designation, alpha sarcoglycanopathy 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).

What treatments are available?

At the time of submission of the application for orphan designation, no satisfactory methods for the treatment of alpha sarcoglycanopathy had been authorised in the European Union. Treatment of patients with the disease mainly involves physiotherapy as supportive treatment, but also tendon-lengthening surgery and fusion of the spine to minimise painful deformity.

How is this medicine expected to work?

This medicinal product is made up of a virus that contains a copy of the human alpha sarcoglycan gene. The virus is used to carry the gene into the muscles of the patient. The gene allows the muscles to produce the alpha sarcoglycan protein, which replaces the missing protein, relieving the symptoms of the disease and halting the progressive muscles weakness. The type of virus used in this medicine ('adeno-associated virus') is modified so that it does not cause disease in humans.

What is the stage of development of this medicine?

The evaluation of the effects of adeno-associated viral vector containing the human alpha sarcoglycan gene in experimental models is ongoing.

At the time of submission of the application for orphan designation, no clinical trials in patients with alpha sarcoglycanopathy had been initiated.

Adeno-associated viral vector containing the human alpha sarcoglycan gene was not marketed anywhere worldwide for alpha sarcoglycanopathy or designated as orphan medicinal product elsewhere for this condition, at the time of submission.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 10 September 2008 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.

*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 502,800,000 (Eurostat 2008).

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 the human alpha-sarcoglycan gene	Treatment of alpha-sarcoglycanopathy
Bulgarian	Аденоасоцииран вирусен вектор, съдържащ човешкия алфа-саркогликанов ген	Лечение на алфа-саркогликанопатия
Czech	Adenovirový vektor s lidským genem pro alfa-sarkoglykan	Léčba alfa -sarkoglykanopatie
Danish	Adenoassocieret viral vektor indeholdende det humane alfa-sarcoglycan-gen	Behandling af alfa-sarcoglycanopati
Dutch	Adeno-geassocieerde virale vector welke het humaan alfa-sarcoglycaan-gen bevat.	Behandeling van alfa-sarcoglycanopathie
Estonian	Adenoga assotsieeruv viirusvektor, mis sisaldab inimese alfa-sarkoglükaani geeni	Alfa-sarkoglükanopaatia ravi
Finnish	Adenoassosioitu virusvektori, joka sisältää ihmisen alfa-sarkoglukaani –geenin	Alfa-sarkoglukanopatian hoito
French	Vecteur viral adéno-associé contenant le gène humain de l'alpha-sarcoglycane	Traitement de l'alpha-sarcoglycanopathie
German	Adeno-assoziierter viraler Vektor, der das human alpha-sarkoglykan-Gen enthält	Behandlung der Alpha-Sarkoglykanopathie
Greek	Αδενοσύνδετος φορέας ιού περιέχων το γονίδιο του ανθρώπινου α-σαρκοπολυσακχαριδίου	Θεραπεία της α-σαρκοπολυσακχαριδοπάθειας
Hungarian	Emberi alfa-szarkoglikán gént hordozó adenovirus vektor	Alfa-sarcoglycanopathia kezelése
Italian	Vettore derivato da adenovirus contenente il gene dell'alfa-sarcoglicano umano	Trattamento della alfa-sarcoglicanopatia
Latvian	Adeno-saistīts vīrusu vektors, kas satur cilvēka alfa-sarkoglikāna gēnu	Alfa-sarkoglikanopātijas ārstēšana
Lithuanian	Adeno-asocijuotas viruso vektorius, pernešantis žmogaus alfa-sarkoglikano geną	Alfa sarkoglikanopatijos gydymas
Maltese	Vettur imnissel mill-adenovirus li fih il-ġene alfa-sarkoglikan uman	Kura ta' l-alfa-sarkoglikanopatija
Polish	Wektor rekombinowanego wirusa sprzężonego z adenowirusem zawierający ludzki gen alfa-sarkoglikanu	Leczenie alfa-sarkoglikanopatii
Portuguese	Vector viral adeno-associado contendo o gene humano alfa-sarcoglicano	Tratamento da alfa-sarcoglicanopatia

[1] At the time of designation

Language	Active Ingredient	Indication
Romanian	Vector viral adeno-asociat conţinând gena umană pentru alfa-sarcoglican	Tratamentul alfa-sarcoglicanopatiei
Slovak	Adeno-asociovaný vírusový vektor obsahujúci ľudský gén alfa-sarkoglykan	Liečba alfa-sarkoglykanopatie
Slovenian	Adenovirusom podobni virusni vektor s humanim genom za alfa sarkoglikan	Zdravljenje alfa-sarkoglikanopatije
Spanish	Vector viral adenoasociado que contiene el gen humano del alfa-sarcoglicano	Tratamiento de la alfa-sarcoglicanopatía
Swedish	Adenoassocierad virusvektor som innehåller den humana alfa-sarcoglycangenen	Behandling av alfa-sarcoglycanopati
Norwegian	Adenoassosiert virusvektor som inneholder humant alfa-sarkoglykangen	Behandling av alfa-sarkoglykanopati
Icelandic	Adenóveirutengd ferja sem inniheldur manna alfa-sarkóglýkan gen	Meðferð við alfa-sarkóglýkankvilla