



EUROPEAN MEDICINES AGENCY
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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Adeno-associated viral vector serotype 5 containing the human *ABCA4* gene for the treatment of Stargardt's disease

First publication	26 October 2009
Rev.1: sponsor's change of address	16 February 2010
Rev.2: sponsor's change of address	26 November 2013
Rev.3: withdrawal from the Community Register	6 October 2014
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Please note that this product was withdrawn from the Community Register of designated Orphan Medicinal Products in June 2014 on request of the Sponsor.

On 6 February 2009, orphan designation (EU/3/08/609) was granted by the European Commission to Fondazione Telethon, Italy, for adeno-associated viral vector serotype 5 containing the human *ABCA4* gene for the treatment of Stargardt's disease.

What is Stargardt's disease?

Stargardt's disease is a genetic (hereditary) disorder of the eye that leads to progressive loss of sight. Stargardt's disease is caused by abnormalities in a gene called *ABCA4*. The *ABCA4* gene is responsible for the production of a protein called ABCR that regulates the transport of substances in and out of some cells in the retina (the light-sensitive surface at the back of the eye). In patients with Stargardt's disease, ABCR does not work properly. This causes deposits to build up inside the retina cells, which become damaged and eventually die.

Stargardt's disease is a long-term debilitating disease because the patient's sight becomes progressively worse and eventually leads to blindness.



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

At the time of designation, Stargardt's disease affected approximately 1.1 in 10,000 people in the European Union (EU). This was equivalent to a total of around 56,000 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 were authorised in the EU for the treatment of the condition. Patients with Stargardt's disease were mainly given genetic counselling (discussion of the risk of passing the condition on to children), and general support such as information and regular medical follow-up.

How is this medicine expected to work?

Adeno-associated viral vector serotype 5 containing the human *ABCA4* gene is made up of a virus that contains the human *ABCA4* gene. The virus is used to carry the *ABCA4* gene into the cells of the retina. By doing this, these cells are expected to start producing the ABCR protein, which will replace the defective protein. When it is injected into the eye, this may relieve the symptoms of the disease and prevent the loss of sight. 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 serotype 5 containing the human *ABCA4* gene have been evaluated in experimental models.

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

At the time of submission, adeno-associated viral vector serotype 5 containing the human *ABCA4* gene was not authorised anywhere in the world for Stargardt's disease or designated as 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 10 December 2008 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 European Union) 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 504,800,000 (Eurostat 2009).

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:

Fondazione Telethon
Via dei Magazzini Generali 18/20
00154 Roma
Italy
Tel. +39 06 44 01 51
Fax +39 06 44 20 20 32
E-mail: info@telethon.it

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, Norwegian and Icelandic

Language	Active Ingredient	Indication
English	Adeno-associated viral vector serotype 5 containing the human <i>ABCA4</i> gene	Treatment of Stargardt's disease
Bulgarian	Адено-свързан вирусен вектор серотип 5 съдържащ човешки <i>ABCA4</i> ген	Лечение на Болест на Stargardt
Czech	Sérotyp 5 adeno asociovaný virový vektor, obsahující lidský gen <i>ABCA4</i>	Léčba Stargardtovy choroby
Danish	Adenoassocieret viral vektor serotype 5 indeholdende det humane gen <i>ABCA4</i>	Behandling af Stargardt sygdom
Dutch	Adeno-geassocieerde virale vector, serotype 5, welke het humane gen <i>ABCA4</i> bevat	Behandeling van de ziekte van Stargardt
Estonian	Adenoga assotsieeruv viirusvektori serotüüp 5, mis sisaldab inimese <i>ABCA4</i> geeni	Stargardt'tõve ravi
Finnish	Serotyyppiä 5 oleva adenoassosioitu virusvektori, joka sisältää ihmisen <i>ABCA4</i> geenin	Stargardtin taudin hoito
French	Vecteur viral adéno-associé de sérotype 5 contenant le gène humain <i>ABCA4</i>	Traitement de la maladie de Stargardt
German	Das humane <i>ABCA4</i> Gen beinhaltender, adeno-assoziiierter viraler Vektor Serotyp 5	Behandlung der Stargardt-Krankheit
Greek	Αδενοσύνδετος φορέας ορότυπου 5 που περιέχει το ανθρώπινο γονίδιο <i>ABCA4</i>	Θεραπευτική αγωγή για την νόσο του Stargardt
Hungarian	Humán <i>ABCA4</i> gént tartalmazó 5-ös szerotípusú adeno-társított vírus vektor	Stargardt-kór kezelése
Italian	Vettore virale adeno-associato del serotipo 5 contenente il gene umano <i>ABCA4</i>	Trattamento della malattia di Stargardt
Latvian	Adeno-saistītā vīrusa vektora 5. serotips, kas satur cilvēka <i>ABCA4</i> gēnu	Stargardta slimības ārstēšana
Lithuanian	Adeno-asocijuotas viruso vektoriaus 5 serotipas, pernešntis žmogaus <i>ABCA4</i> geną	Stargardt ligos gydymas
Maltese	Vettur imnissel mill-adenovirus tas-serotip 5 li fih il-ġene uman <i>ABCA4</i>	Kura tal-marda ta' Stargardt
Polish	Wektor adenowirusowy serotyp 5 zawierający ludzki gen <i>ABCA4</i>	Leczenie choroby Stargardta
Portuguese	Vector viral adeno-associado de serotipo 5 contendo o gene humano <i>ABCA4</i>	Tratamento da doença de Stargardt
Romanian	Vector viral adeno-asociat de serotip 5 conținând gena umană <i>ABCA4</i>	Tratamentul bolii Stargardt
Slovak	Adeno-asociovaný vírusový vektorový sérotyp 5 obsahujúci ľudský gén <i>ABCA4</i>	Liečba Stargardtovej choroby

Language	Active Ingredient	Indication
Slovenian	Adenovirusni pridruženi virusni vektor serotipa 5, ki vsebuje človeški gen <i>ABCA4</i>	Zdravljenje Stargardtjeve bolezni
Spanish	Vector viral adenoasociado del serotipo 5 que contiene el gen humano <i>ABCA4</i>	Tratamiento de la enfermedad de Stargardt
Swedish	Adenoassocierad virusvektor serotyp 5, innehållande den mänskliga genen <i>ABCA4</i>	Behandling av Stargardts sjukdom
Norwegian	Adenoassosiert virusvektor serotype 5 som inneholder det humane genet <i>ABCA4</i>	Behandling av Stargardts sykdom
Icelandic	Adenotengd veirufurja af sermisflokki 5 sem inniheldur manna <i>ABCA4</i> gen	Meðferð við Stargardts sjúkdómi