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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Lentiviral vector containing the human *ABCA4* gene for the treatment of Stargardt's disease

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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.	

On 2 February 2010, orphan designation (EU/3/09/720) was granted by the European Commission to Oxford BioMedica (UK) Ltd, United Kingdom, for lentiviral vector containing the human *ABCA4* gene for the treatment of Stargardt's disease.

The sponsorship was transferred to Sanofi-Aventis Recherche & Développement, France, in June 2014.

What is Stargardt's disease?

Stargardt's disease is a genetic disorder of the eye that leads to the gradual loss of sight. It is caused by abnormalities in a gene called *ABCA4*. The *ABCA4* gene is responsible for the production of a protein called ABCR, which controls the movement of substances into and out of 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 it leads to the patient's sight getting worse and eventually to blindness.

30 Churchill Place ● Canary Wharf ● London E14 5EU ● United Kingdom
Telephone +44 (0)20 3660 6000 **Facsimile** +44 (0)20 3660 5555
Send a question via our website www.ema.europa.eu/contact

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

At the time of designation, Stargardt's disease affected approximately 1 in 10,000 people in the European Union (EU). This was equivalent to a total of around 51,000 people*, and is below the threshold for orphan designation, which is 5 people in 10,000. This is based on the information provided by the sponsor and 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 Stargardt's disease. Patients with the disease were usually given physical aids such as sunglasses to reduce the rate of damage to the retina, or spectacles, magnifiers or telescopes to help them see during the early stages of the disease. Laser treatment can help to stabilise the loss of vision in some patients. Patients also often have genetic counselling (discussion of the risks of passing the condition on to children).

How is this medicine expected to work?

Lentiviral vector containing the human *ABCA4* gene is an advanced therapy medicine that belongs to the group called 'gene therapy products'. These are medicines that work by delivering genes into the body. The medicine 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. These cells can then start to produce normal ABCR protein to replace the defective protein. When it is injected into the eye, the medicine is expected to relieve the symptoms of the disease and prevent the loss of sight. The type of virus used in this medicine (lentivirus) is modified so that it does not cause disease in humans.

What is the stage of development of this medicine?

At the time of submission of the application for orphan designation, the evaluation of the effects of lentiviral vector containing the human *ABCA4* gene in experimental models was ongoing.

At the time of submission, no clinical trials with the designated product in patients with Stargardt's disease had been started.

At the time of submission, lentiviral vector containing the human *ABCA4* gene was not authorised anywhere in the EU for Stargardt's disease 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 3 December 2009 recommending the granting of this designation.

*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 506,300,000 (Eurostat 2010).

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 Community) 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:

Sanofi-Aventis Recherche & Développement
Part of the Sanofi-Aventis Groupe
France
Tel. +33 153 774 000
Fax +33 153 774 133
www.sanofi-aventis.com/contact/contact.asp

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	Lentiviral vector containing the human ABCA4 gene	Treatment of Stargardt's disease
Bulgarian	Лентивирусен вектор, съдържащ човешки ABCA4 ген	Лечение на Болест на Stargardt
Croatian	Lentivirusni vektor koji sadrži ljudski gen *ABCA4*	Liječenje Stargardtove bolesti
Czech	Lentivirový vektor obsahující lidský gen ABCA4	Léčba Stargardtovy choroby
Danish	Lentiviral vektor indeholdende det humane ABCA4 gen	Behandling af Stargardt sygdom
Dutch	Lentivirale vector welke humaan ABCA4 gen bevat	Behandeling van de ziekte van Stargardt
Estonian	Inimese ABCA4 geeni sisaldav lentiviraalne vektor	Stargardt'tõve ravi
Finnish	Lentivirusvektori, jossa on ihmisen ABCA4-geeni	Stargardtin taudin hoito
French	Vecteur lentiviral contenant le gène humain ABCA4	Traitement de la maladie de Stargardt
German	Lentiviraler Vektor, der das menschliche Gen ABCA4 enthält	Behandlung der Stargardt-Krankheit
Greek	Όχημα lenti-ιού που φέρει το ανθρώπινο γονίδιο ABCA4	Θεραπευτική αγωγή για την νόσο του Stargardt
Hungarian	Humán ABCA4 gént tartalmazó lentivirális vektor	Stargardt-kór kezelése
Italian	Vettore lentivirale contenente il gene umano ABCA4	Trattamento della malattia di Stargardt
Latvian	Cilvēka ABCA4 gēnu saturošs lentivīrusa vektors	Stargardta slimības ārstēšana
Lithuanian	Lentivirusinis vektorius, turintis žmogaus ABCA4 geną	Stargardt ligos gydymas
Maltese	Vettur lentivirali li fih il-ġene ABCA4 uman	Kura tal-marda ta' Stargardt
Polish	Wektor lentiwirusowy zawierający ludzki gen ABCA4	Leczenie choroby Stargardta
Portuguese	Vector lentiviral contendo o gene ABCA4 humano	Tratamento da doença de Stargardt
Romanian	Vector lentiviral conţinând gena umană ABCA4	Tratamentul bolii Stargardt
Slovak	Lentivírusový vektor obsahujúci ľudský gén ABCA4	Liečba Stargardtovej choroby
Slovenian	Lentivirusni vector s humanim genom ABCA4	Zdravljenje Stargardtjeve bolezni

[1] At the time of transfer of sponsorship

Language	Active ingredient	Indication
Spanish	Vector lentivírico que contiene el gen humano ABCA4	Tratamiento de la enfermedad de Stargardt
Swedish	Lentivirusvektor innehållande den humana ABCA4-genen	Behandling av Stargardts sjukdom
Norwegian	Lentiviral vektor som inneholder genet for human ABCA4	Behandling av Stargardts sykdom
Icelandic	Lentiveiru ferja sem inniheldur manna ABCA4 genið	Meðferð við Stargardts sjúkdómi