



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Adeno-associated viral vector containing DNA encoding an RNAi targeting rhodopsin / adeno-associated viral vector containing a rhodopsin gene for the treatment of rhodopsin-linked retinitis pigmentosa

On 17 December 2010, orphan designation (EU/3/10/817) was granted by the European Commission to Genable Technologies Ltd, Ireland, for adeno-associated viral vector containing DNA encoding an RNAi targeting rhodopsin / adeno-associated viral vector containing a rhodopsin gene for the treatment of rhodopsin-linked retinitis pigmentosa.

What is rhodopsin-linked retinitis pigmentosa?

Retinitis pigmentosa is a group of hereditary diseases of the eye that lead to progressive loss of sight. In patients with retinitis pigmentosa, cells in the retina (the light-sensitive surface at the back of the eye) become damaged and eventually die. Retinitis pigmentosa is seen in a variety of diseases, including diseases that affect the production of rhodopsin, a light-sensitive protein found in the retina. Patients with rhodopsin-linked retinitis pigmentosa have a mutation (fault) in the gene for this protein.

Rhodopsin-linked retinitis pigmentosa is a long-term debilitating disease because it causes the patient's sight to deteriorate, eventually leading to blindness.

What is the estimated number of patients?

At the time of designation, rhodopsin-linked retinitis pigmentosa affected approximately 0.4 in 10,000 people in the European Union (EU)*. This is equivalent to a total of around 20,000 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 designation, no satisfactory methods were authorised in the EU for treating rhodopsin-linked retinitis pigmentosa. Patients with the condition were given genetic counselling (discussion of

*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. This represents a population of 506,500,000 (Eurostat 2010).



the risks of passing the condition on to children), treatment for complications such as cataract (clouding of the lens) and general support with the social and psychological impact of blindness.

How is this medicine expected to work?

The medicine is a 'gene therapy product' that works by delivering genes into the body. The medicine is made of two viruses. One virus contains a piece of DNA that blocks both copies of the patient's rhodopsin gene, the normal copy and the faulty rhodopsin gene, and the other one contains a normal copy of the rhodopsin gene that cannot be blocked. When the medicine is injected into the eye, the viruses are expected to carry the genetic material into the cells of the eye so that only normal rhodopsin is produced.

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?

At the time of submission of the application for orphan designation, the evaluation of the effects of the medicine in experimental models was ongoing.

At the time of submission, no clinical trials with the medicine in patients with rhodopsin-linked retinitis pigmentosa had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for rhodopsin-linked retinitis pigmentosa 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 7 October 2010 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 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:

Genable Technologies Ltd
Smurfit Institute of Genetics
Trinity College Dublin
Dublin 2
Ireland
Telephone: +353 1 896 3695
Telefax: +353 1 679 8558
E-mail: info@genable.ie

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 languages¹, Norwegian and Icelandic

Language	Active ingredient	Indication
English	Adeno-associated viral vector containing DNA encoding an RNAi targeting rhodopsin / adeno-associated viral vector containing a rhodopsin gene	Treatment of rhodopsin-linked retinitis pigmentosa
Bulgarian	Адено-асоцииран вирусен вектор, съдържащ ДНК кодиран и иРНК прицелен родопсин/ адено-асоцииран вирусен вектор, съдържащ родопсинов ген	Лечение на родопсин-свързан ретинитис пигментоза.
Czech	Adeno-asociovaný virový vector obsahující DNA kódující RNAi cílenou proti rhodopsinu/ Adeno-asociovaný virový vector obsahující gen pro rhodopsin	Léčba rhodopsin-související retinitis pigmentóza
Danish	Adeno-associeret viral vektor som indeholder DNA kodende for RNAi målrettet rhodopsin / adeno-associeret viral vektor indeholdende et rhodopsin gen	Behandling af rhodopsin koblet retinitis pigmentosa
Dutch	Adeno-geassocieerde virale vector dewelke DNA bevat coderend voor een RNAi gericht op rhodopsine/ adeno- geassocieerde virale vector dewelke een rhodopsine gen bevat	Behandeling van rhodopsine-geassocieerde retinitis pigmentosa
Estonian	Adenoviirusega seotud viirusvektor, mis sisaldab DNA, mis kodeerib rodopsiini RNAi ja adenoviirusega seotud viirusvektor, mis sisaldab rodopsiini geeni	Rodopsiiniga seotud <i>retinitis pigmentosa</i> ravi
Finnish	Adenoassosioitu virusvektori, joka sisältää RNAi:ta koodaavaa DNA:ta rodopsiinia kohti/adenoassosioitu virusvektori, joka sisältää rodopsiinigeenin	Rodopsiiniin-liittyvä retinitis pigmentosa
French	Vecteur viral adénovirus-associé contenant l'ADN codant un ARN interférent ciblant la rhodopsine / Vecteur viral adénovirus-associé contenant un gène de la rhodopsine	Traitement de la retinite pigmentaire liée à la rhodopsine
German	Adeno-assoziiertes viraler Vektor, der für eine gegen Rhodopsin gerichtete RNAi kodiert / Adeno-assoziiertes viraler Vektor, der für ein Rhodopsin Gen kodiert	Behandlung einer Rhodopsin-assoziierten Retinitis pigmentosa
Greek	Ίκός φορέας σχετιζόμενος με αδενοϊό που περιέχει DNA που κωδικοποιεί για ένα RNAi έναντι της ροδοψίνης/Ίκός φορέας σχετιζόμενος με αδενοϊό που περιέχει ένα γονίδιο ροδοψίνης.	Θεραπεία της μελαχρωστικής αμφιβληστροειδοπάθειας που σχετίζεται με τη ροδοψίνη

¹ At the time of designation

Hungarian	Rodopszinra irányuló iRNS-t kódoló DNS-t tartalmazó adeno-asszociált vírus vektor / rodopszin-gént tartalmazó adeno-asszociált vírus vektor	Rodopszinhosz kapcsolódó retinitis pigmentosa kezelése
Italian	Vettore virale adeno-associato contenente DNA codificante un RNAi che silenzia il gene della Rodopsina/vettore virale adeno-associato contenente un gene della rodopsina	Trattamento della retinite pigmentosa associata alla rodopsina
Latvian	Adeno-asociētais vīrusa vektors, kas satur DNS kodētu RNS mērkētu uz rodopsīn/adeno asociēto rodopsīna gēnu saturošu vīrusa vektoru	Rodopsīna saistītā retinitis pigmentosa ārstēšana
Lithuanian	Adeno-susijusio viruso vektorius su DNR koduojančia rodopsino RNR interferencija/ adeno-susijusio viruso vektorius su rodopsino genu	Pigmentinio retinito susijusio su rodopsinu gydymas
Maltese	Vettur imnissel mill-adenovirus li fih DNA li jikkodifika RNAi immirat għar-rodopsina / vettur imnissel mill-adenovirus li fih ġene għar-rodopsina	Kura tar-retinite pigmentosa assoċjata mar-rodopsina
Polish	Wektor adenowirusowy zawierający DNA kodujący RNAi dla rodopsyny / Wektor adenowirusowy zawierający gen rodopsyny	Leczenie retinopatii barwnikowej spowodowanej rodopsyną
Portuguese	Vector viral adeno-associado contendo ADN para a expressão de um ARNi anti rodopsina / Vector viral adeno-associado contendo o gene da rodopsina	Tratamento da Retinite pigmentar ligada à rodopsina
Romanian	Vector viral adeno-asociat conținând ADN codant pentru ARNi ce are drept țintă rodopsina/ vector viral adeno-asociat ce conține o genă pentru rodopsină	Tratamentul retinitei pigmentare rodopsin-asociată
Slovak	Adenóvírusový vektor obsahujúci DNA kódujúcu RNAi pre rodopsín / adenóvírusový vektor obsahujúci gén pre rodopsín	Liečba pigmentovej retinitídy spojenej s rodopsínom
Slovenian	Adenovirusni vector, ki vsebuje kodirano DNA za RNA rodopsina/adenovirusni vector , ki vsebuje gen za rodopsin	Zdravljenje z rodopsinom povezanega pigmentoznega retinitisa
Spanish	Vector viral adeno asociado conteniendo ADN que codifica un iARN para rodopsina/vector viral adeno asociado conteniendo un gen de rodopsina	Tratamiento de la renitis pigmentosa ligada a rodopsina
Swedish	Adenoassocierad virusvektor innehållande DNA som kodar för RNAi mot rodopsin/adenoassocierad virusvektor innehållande en rodopsin gen	Rodopsin-relaterad retinitis pigmentosa
Norwegian	Adenoassosiert viral vektor som inneholder DNA kodende for RNAi mot rhodopsin / adenoassosiert viral vektor som inneholder et gen for rhodopsin	Behandling av rodopsin-forbundet retinitis pimentosa
Icelandic	Adenó-tengd veirufurja sem inniheldur DNA sem umritar RNAi sem beinist gegn rhódopsín/adenó-tengdri veirufurju sem inniheldur rhódopsín gen.	Meðferð á rhódopsín-tengdri retinitis pigmentosa