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Committee for Orphan Medicinal Products

## Public summary of opinion on orphan designation

Adenovirus associated viral vector serotype 4 containing the human *RPE65* gene for the treatment of retinitis pigmentosa

First publication	1 July 2008
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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 14 November 2007, orphan designation (EU/3/07/486) was granted by the European Commission to Centre Hospitalier Universitaire de Nantes, France, for adenovirus associated viral vector serotype 4 containing the human *RPE65* gene for treatment of retinitis pigmentosa.

The sponsorship was transferred to HORAMA SAS, France, in May 2015.

### What is retinitis pigmentosa?

Retinitis pigmentosa is a genetic disorder, characterised by progressive loss of sight. Approximately 2% of patients affected by retinitis pigmentosa have a defective *RPE65* gene, and thus do not produce the *RPE65* protein in the cells in the eye. *RPE65* protein normally helps the function of photoreceptors that are the light receiving cells in the eye needed for the eyesight. Retinitis pigmentosa is chronically debilitating, due to progressive loss of vision.

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

At the time of designation, retinitis pigmentosa affected approximately 2.9 in 10,000 people in the European Union (EU). This was equivalent to a total of around 145,000 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).

\*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 500,300,000 (Eurostat 2007).



## What treatments are available?

At the time of submission of the application for orphan designation, no satisfactory method had been authorised in the European Union for the treatment of the condition. Treatment of patients with retinitis pigmentosa primarily involved genetic counselling, and general support such as information and regular medical follow up.

## How is this medicine expected to work?

Adenovirus associated viral vector serotype 4 containing the human *RPE65* gene is a medicinal product which uses a virus to carry the gene necessary for the production of the protein *RPE65*. A virus is a small organism capable of introducing genetic material in cells. By doing this the virus will introduce the gene in the cells where the gene for protein *RPE65* is lacking and then the cells infected by the virus will start producing the protein. The type of virus (adeno-associated virus) used in this medicinal product is modified to avoid causing disease in humans.

## What is the stage of development of this medicine?

The evaluation of the effects of adenovirus associated viral vector serotype 4 containing the human *RPE65* gene in experimental models is ongoing.

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

Adenovirus associated viral vector serotype 4 containing the human *RPE65* gene was not authorised anywhere in the world for retinitis pigmentosa 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 12 September 2007 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.

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:

HORAMA SAS  
9 rue de l'Eperon  
75006 Paris  
France  
Tel. +33 6 22 73 26 34  
<http://www.horama.fr/contact/>

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<sup>1</sup>, Norwegian and Icelandic

Language	Active Ingredient	Indication
English	Adenovirus associated viral vector serotype 4 containing the human RPE65 gene	Treatment of retinitis pigmentosa
Bulgarian	Адено-асоцииран вирусен вектор серотип 4, съдържащ човешкия ген RPE65	Лечение на пигментен ретинит
Croatian	Adeno-povezani virusni vektor serotipa 4 koji sadrži ljudski gen RPE65	Liječenje Leberove kongenitalne amauroze
Czech	Sérotyp 4 virového vektoru spojeného s adenovirem, obsahující lidský gen RPE65	Léčba pigmentosní retinitidy
Danish	Adenovirus associeret viral vektor serotype 4 indeholdende det humane gen RPE65	Behandling af retinitis pigmentosa
Dutch	Adenovirus geassocieerde virale vector, serotype 4, welke het humane gen RPE65 bevat	Behandeling van retinitis pigmentosa
Estonian	Adeniivirusega ühendatud viirusvektori serotüüp 4, mis sisaldab inimese RPE65 geeni	Pigmentoosse võrkkestapõletiku ravi.
Finnish	Adenovirukseen liittyvä, serotyyppiä 4 oleva virustartunnanvälittäjä, joka sisältää ihmisen RPE65 geenin	Hoito verkkokalvorappeumaan
French	Vecteur viral adénovirus-associé de sérotype 4 contenant le gène humain RPE65	Traitement de la rétinite pigmentaire
German	Das humane RPE65 Gen beinhaltender, adenovirus-assoziiertes viraler Vektor Serotyp 4	Behandlung der Retinopathia Pigmentosa
Greek	Ιογενής φορέας συνδεόμενος με αδενοϊό, ορότυπου 4 που περιέχει το ανθρώπινο γονίδιο RPE65	Αγωγή κατά της μελαγχρωστικής αμφιβληστροειδοπάθειας
Hungarian	Humán RPE65 gént tartalmazó 4-es szerotípusú adenovírus vektor	Retinitis pigmentosa kezelése
Italian	Vettore virale adenovirus-associato del serotipo 4 contenente il gene umano RPE65	Trattamento della retinite pigmentosa
Latvian	Adenovīrusa Saistītā Virālā vektora 4. serotips, kas satur cilvēka RPE65 gēnu	Retinitis pigmentosa ārstēšana
Lithuanian	Adeno - asocijuotas virusinio vektoriaus 4 serotipas, pernešantis žmogaus RPE65 geną	Pigmentinio retinito gydymas
Maltese	Vektor virali assoċjat ma' l-adenovirus tas-serotip 4 li għandu l-gene uman RPE65	Kura tar-retinite pigmentuża
Polish	serotyp 4 wektora wirusowego związanego z adenowirusem, zawierający ludzki gen RPE65	Leczenie retinopatii barwnikowej
Portuguese	Vector viral associado ao adenovirus de serotipo 4 contendo o gene humano RPE65	Tratamento de retinite pigmentosa

<sup>1</sup> At the time of designation

Language	Active Ingredient	Indication
Romanian	Vector viral asociat adenovirusului de serotip 4 conținând gena umană RPE65	Tratamentul retinitei pigmentare
Slovak	Adenovírusový pridružený vírusový vektorový sérotyp 4 obsahujúci ľudský gén RPE65	Liečba retinitis pigmentosa
Slovenian	Adenovirusom sorodni virusni vektor serotipa 4, ki vsebuje človeški gen RPE65	Zdravljenje pigmentozne retinopatije
Spanish	Vector vírico asociado al adenovirus del serotipo 4 que contiene el gene humano RPE65	Tratamiento de retinosis pigmentaria
Swedish	Adenoassocierat virus, viral vektor serotyp 4, innehållande den mänskliga genen RPE65	Behandling av retinitis pigmentosa
Norwegian	Adenoassosiert virusvektor serotype 4 som inneholder det humane genet RPE65	Behandling av retinitis pigmentosa
Icelandic	Adenóveirutengd (af flokki 4) veirufurja sem inniheldur RPE65 gen úr manni	Meðferð áh retinitis pigmentosa