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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Adenovirus-associated viral vector serotype 2 containing the human *RPE65* gene for the treatment of retinitis pigmentosa

On 28 July 2015, orphan designation (EU/3/15/1518) was granted by the European Commission to Alan Boyd Consultants Ltd, United Kingdom, for adenovirus-associated viral vector serotype 2 containing the human *RPE65* gene for the treatment of retinitis pigmentosa.

What is 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 a long-term debilitating disease because it causes the patient's sight to get worse, eventually leading to blindness.

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

At the time of designation, retinitis pigmentosa affected less than 3.7 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 190,000 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 retinitis pigmentosa. Patients with the condition were given sunglasses to slow down the damage to the retina, genetic counselling (discussion of the risks of passing the condition on to children) and general support.

[*]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 28), Norway, Iceland and Liechtenstein. This represents a population of 512,900,000 (Eurostat 2015).

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How is this medicine expected to work?

One form of retinitis pigmentosa is caused by mutations (changes) in the *RPE65* gene which is responsible for the production of an enzyme, called all-*trans* retinyl isomerase, that is necessary for the normal functioning of retinal cells. In patients with this form of the disease this enzyme is lacking.

The medicine consists of a virus that contains normal copies of the *RPE65* gene. When injected into the eye, under the retina, it is expected that the virus carries the *RPE65* gene into the retinal cells, enabling them to produce the missing enzyme. This is then expected to help the cells in the retina to function better, reducing symptoms of the condition.

The type of virus used in this medicine (adeno-associated virus) does not cause disease in humans.

What is the stage of development of this medicine?

The effects of the medicine have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with the medicine in patients with retinitis pigmentosa were ongoing.

At the time of submission, the medicine was not authorised anywhere in the EU for retinitis pigmentosa. Orphan designation had been granted in the United States for treatment of retinitis pigmentosa due to autosomal recessive *RPE65* gene mutations.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 18 June 2015 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

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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 - CHECK
English	Adenovirus-associated viral vector serotype 2 containing the human *RPE65* gene	Treatment of retinitis pigmentosa
Bulgarian	Аденовирусно-асоцииран вирусен вектор серотип 2, съдържащ човешкия ген *RPE65*	Лечение на пигментен ретинит
Croatian	Adeno-povezani virusni vektor serotipa 2 koji sadrži ljudski gen *RPE65*	Liječenje retinitisa pigmentoze
Czech	Adeno-asociovaný virus sérotypu 2 obsahující lidský gen *RPE65*	Léčba pigmentosní retinitidy
Danish	Adenovirus associeret viral vektor serotype 2 indeholdende det humane gen *RPE65*	Behandling af retinitis pigmentosa
Dutch	Adenovirus geassocieerde virale vector, serotype 2, welke het humane gen *RPE65* bevat	Behandeling van retinitis pigmentosa
Estonian	Adenoviirusega seotud viirusvektor serotüüp 2, mis sisaldab inimese *RPE65* geeni	Pigmentoosse võrkkestapõletiku ravi
Finnish	Serotyypin 2 adenovirusvektori, jossa on ihmisen *RPE65*-geeni	Verkkokalvorappeuman hoito
French	Vecteur viral adéno-associé de type 2 contenant le gène humain *RPE65*	Traitement de la rétinite pigmentaire
German	Adenovirus-assoziierter viraler Vektor Serotyp 2, der das humane *RPE65* Gen enthält	Behandlung der Retinopathia Pigmentosa
Greek	Ιικός φορέας σχετιζομενος με αδενοϊό ορότυπου 2 που περιέχει το ανθρώπινο γονίδιο *RPE65*	Θεραπεία της μελαγχρωστικής αμφιβληστροειδοπάθειας
Hungarian	Humán *RPE65* gént tartalmazó 2-es szerotípusú adenovírus vektor	Retinitis pigmentosa kezelése
Italian	Vettore virale adenovirus-associato del serotipo 2 contenente il gene umano *RPE65*	Trattamento della retinite pigmentosa
Latvian	Adenovīrusa saistīts 2. seroptipa vīrusa vektors, kas satur cilvēka *RPE65* gēnu	*Retinitis pigmentosa* ārstēšana
Lithuanian	Adeno-asocijuoto viruso vektoriaus 2 serotipas, turintis žmogaus *RPE65* geną	Pigmentinio retinito gydymas
Maltese	Vektor virali assoċjat ma' l-adenovirus tas-serotip 2 li għandu l-ġene uman *RPE65*	Kura tar-retinite pigmentuża
Polish	Wektor adenowirusowy serotyp 2 zawierający ludzki gen *RPE65*	Leczenie retinopatii barwnikowej
Portuguese	Vector viral adeno-associado de serotipo 2 contendo o gene humano *RPE65*	Tratamento da retinite pigmentosa
Romanian	Vector viral adeno-asociat de serotip 2 ce conţine gena umană *RPE65*	Tratamentul retinitei pigmentare
Slovak	Vírusový vektor spojený s adenovírusom sérotyp 2 obsahujúci ľudský gén *RPE65*	Liečba retinitis pigmentosa

[1] At the time of designation

Language	Active ingredient	Indication - CHECK
Slovenian	Adenovirusom sorodni virusni vektor serotipa 2, ki vsebuje človeški gen *RPE65*	Zdravljenje pigmentozne retinopatije
Spanish	Vector vírico adenoasociado del serotipo 2 que conteniene el gen humano *RPE65*	Tratamiento de la retinosis pigmentaria
Swedish	Adenoassocierad virusvektor av serotyp 2, innehållande den humana *RPE65* genen	Behandling av retinitis pigmentosa
Norwegian	Adenoassosiert virusvektor serotype 2 som inneholder det humane genet *RPE65*	Behandling av retinitis pigmentosa
Icelandic	Adenóveiru tengd veiruferja af sermisgerð 2 sem inniheldur manna RPE65 gen	Meðferð á retinitis pigmentosa