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Committee for Orphan Medicinal Products

## Public summary of opinion on orphan designation

Adeno-associated viral vector serotype 2 containing the human *CHM* gene encoding human Rab escort protein 1 for the treatment of choroideremia

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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 4 June 2014, orphan designation (EU/03/14/1278) was granted by the European Commission to Alan Boyd Consultants Ltd, United Kingdom, for adeno-associated viral vector serotype 2 containing the human *CHM* gene encoding human Rab escort protein 1 for the treatment of choroideremia.

### What is choroideremia?

Choroideremia is a hereditary disease of the eye that leads to progressive loss of sight. The disease mostly affects males. In patients with choroideremia, cells in the retina (the light-sensitive surface at the back of the eye), the retinal pigment epithelium (the cell layer just outside the retina that nourishes retinal cells) and the choroid (a network of blood vessels located between the retina and the sclera, the "white of the eye") become damaged and eventually die.

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

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

At the time of designation, choroideremia affected less than 0.2 people in 10,000 per year in the European Union (EU). This was equivalent to fewer than 10,000 people per year\*, and is below the

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\*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 511,100,000 (Eurostat 2014).



ceiling for orphan designation. 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 to treat choroideremia. Vitamins, supplements and a healthy diet were often used to manage the disease. Patients were advised to use sunglasses in very sunny conditions. Patients with choroideremia usually received genetic counselling on the risks of passing the condition on to their children, and regular medical follow up.

### **How is this medicine expected to work?**

Choroideremia is caused by a defect in the *CHM* gene, which is responsible for the production of a protein called 'Rab escort protein 1'. This protein has a key role in the correct functioning of the cells of the eye.

This medicine is made of a virus that contains a normal copy of the *CHM* gene. It is expected that when injected into the patient's eye the virus will carry the *CHM* gene into the cells of the eye, enabling them to function normally and thereby helping to improve the patient's sight.

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?**

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 of the application for orphan designation, no clinical trials with the medicine in patients with choroideremia had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for choroideremia. Orphan designation has been granted in the United States for 'treatment of choroideremia due to mutations in the human choroideremia gene'.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 9 April 2014 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:

Alan Boyd Consultants Ltd  
Electra House  
Crewe Business Park, Crewe  
Cheshire CW1 6GL  
United Kingdom  
Tel. +44 (0)1270 270 010  
Fax +44 (0)1270 253 832  
E-mail: [info@bodyconsultants.com](mailto:info@bodyconsultants.com)

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	Adeno-associated viral vector serotype 2 containing the human CHM gene encoding human Rab escort protein 1	Treatment of choroideremia
Bulgarian	Адено-асоцииран вирусен вектор серотип 2, съдържащ човешкия CHM ген, кодиращ човешкия Rab escort протеин 1	Лечение на хороидеремия
Croatian	Adeno-povezani virusni vektor serotipa 2 koji sadrži ljudski gen CHM koji kodira za ljudski Rab prateći protein 1	Liječenje koroideremije
Czech	Adeno-asociovaný virový vektor sérotypu 2 obsahující lidský gen CHM kódující lidský Rab escort protein 1	Léčba choroidermie
Danish	Adeno-associeret viral vektor serotype 2 indeholdende det humane CHM gen, der koder for humant Rab ledsagelse protein 1	Behandling af choroideremi
Dutch	Adeno-geassocieerde virale vector serotype 2 die het humaan CHM-gen bevat dat codeert voor het Rab-escortproteïne 1	Behandeling van choroïderemie
Estonian	Adenoviirusega seotud viirusvektori serotüüp 2, mis sisaldab inimese Rab transportvalk-1 kodeerivat inimese CHM geeni	Koroidereemia ravi
Finnish	Adenovirukseen yhdistetty serotyyppi 2 virusvektori, joka sisältää Rab eskorttiproteiini 1:tä koodaavan ihmisen CMH-geenin	Koroideremian hoito
French	Vecteur dérivé de l'adénovirus de sérotype 2 qui contient le gène humain CHM codant la protéine 1 qui accompagne la protéine Rab humaine	Traitement de la choroïdémie
German	Adeno-assoziiertes virales Vektor 2, das das menschliche Gen CHM enthält, das für das Rab Begleitprotein 1 kodiert	Behandlung von Choroïderemie
Greek	Αδενο-σχετιζόμενος ιικός φορέας οροτύπου 2 που περιέχει το ανθρώπινο γονίδιο CHM, το οποίο κωδικοποιεί την ανθρώπινη πρωτεΐνη Rab 1	Θεραπεία για την έλλειψη του χοριοειδούς χιτώνα του οφθαλμού
Hungarian	Humán Rab escort protein-1-et kódoló humán CHM gént hordozó 2-es szerotípusú adeno-asszociált vírus vektor	Choroideraemia kezelésé
Italian	Vettore virale adenovirus-associato del serotipo 2 contenente il gene umano CHM che codifica la proteina Rab 1 umana	Trattamento della coroideremia

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

Language	Active ingredient	Indication
Latvian	Adeno-saistīts 2 serotipa virālais vektors , kas satur cilvēka CHM gēna kodējumu cilvēka Rab pavadošais proteīns 1	Horoiderēmijasc ārstēšana
Lithuanian	Adeno asocijuoto viruso vektoriaus 2 serotipas, turintis žmogaus CHM geną, koduojantį žmogaus Rab lydintį baltymą 1	Choroideremijos gydymas
Maltese	Vettor virali imnissel mill-adenovirus tas-serotip 2 li fih il-gene uman CHM li jikkodifika l-proteina tat-tip 1 li takkumpanja l-proteina Rab umana	Kura tal-korojderemja
Polish	Wektor adenowirusowy serotypu 2 zawierający ludzki gen CHM kodujący białko transportujące Rab 1	Leczenie choroideremi
Portuguese	Vector viral adeno-associado de serotipo 2 que contém o gene CHM humano que codifica a proteína acompanhante 1 de Rab humana	Tratamento de Coroidermia
Romanian	Vector viral adeno-asociat de serotip 2 conținând gena umană CHM care codifică proteinaînsoțitoare a RAB 1	Tratamentul coroideremiei
Slovak	Adeno-asociovaný vírusový vektor sérotypu 2, obsahujúci ľudský CHM gén, kódujúci ľudský Rab sprievodný proteín 1	Liečba choroiderémie
Slovenian	Adeno-pridružení virus serotipa 2, ki vsebuje človeški gen CHM, ki zapisuje človeški eskortni protein Rab 1	Zdravljenje horoideremije
Spanish	Vector viral adenoasociado del serotipo 2 que contiene el gen humano CHM y que codifica a la proteína humana acompañante de Rab 1	Tratamiento para coroidemia
Swedish	Adenoassocierad virusvektor serotyp 2 innehållande den mänskliga CHM-genen som kodar för mänskligt Rab escort protein 1	Behandling av koroideremi
Norwegian	Adenoassosiert viral vektor serotype 2 som inneholder det humane CHM genet som koder for humant Rab transportprotein 1	Behandling av koroideremi
Icelandic	Adenó-tengd veirufurja af sermisgerð 2 sem inniheldur manna CHM-gen sem tjáir manna Rab fylgdarprótein-1	Meðferð við æðahimnuhrönnun