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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Adenovirus associated viral vector serotype 8 containing the human *CNGB3* gene for the treatment of achromatopsia caused by mutations in the *CNGB3* gene

On 11 November 2015, orphan designation (EU/3/15/1578) was granted by the European Commission for adenovirus associated viral vector serotype 8 containing the human *CNGB3* gene for the treatment of achromatopsia caused by mutations in the *CNGB3* gene.

An administrative transfer of sponsorship, to Athena Vision Ltd, United Kingdom, was finalised in January 2016.

What is achromatopsia caused by mutations in the *CNGB3* gene?

Achromatopsia is an inherited disease of the eye that leads to reduced visual acuity (how well a person can see), colour blindness, severe photophobia (increased sensitivity to light) and nystagmus (fast involuntary eye movements). Achromatopsia is a disorder of the retina (the light-sensitive surface at the back of the eye). In patients with achromatopsia, a type of retina cells called 'cone photoreceptors', which provide vision in bright light including colour vision, do not function normally.

Achromatopsia is often caused by mutations (changes) in the *CNGB3* gene. It is a long-term debilitating disease because it affects how well a person can see in bright light, which may limit everyday activities.

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

At the time of designation, achromatopsia caused by mutations in the *CNGB3* gene affected approximately 0.15 in 10,000 people in the European Union (EU). This was equivalent to a total of around 8,000 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 28), Norway, Iceland and Liechtenstein. This represents a population of 512,900,000 (Eurostat 2015).

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What treatments are available?

At the time of designation, no satisfactory methods were authorised in the EU for treating achromatopsia caused by mutations in the *CNGB3* gene. Patients with the condition were given glasses with red-coloured lenses to reduce light sensitivity, as well as low vision aids such as magnifiers for reading.

How is this medicine expected to work?

In this disease, a protein involved in the normal functioning of cone photoreceptors, called the CNGB3 protein, does not work properly due to defects in the gene responsible for producing it.

This medicine is made up of a virus that contains normal copies of the *CNGB3* gene. When injected into the patient's eyes, it is expected that the virus will carry the *CNGB3* gene into photoreceptor cells, so that a functional *CNGB3* protein can be produced. This is expected to enable cone photoreceptors to work properly, thereby preventing the symptoms of the disease.

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, no clinical trials with the medicine in patients with achromatopsia had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for achromatopsia caused by mutations in the *CNGB3* gene 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 8 October 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

Sponsor's contact details:

Contact details of the current sponsor for this orphan designation can be found on EMA website, on the medicine's rare disease designations page.

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	Adenovirus associated viral vector serotype 8 containing the human *CNGB3* gene	Treatment of achromatopsia caused by mutations in the *CNGB3*
Bulgarian	Аденовирусно-асоцииран вирусен вектор серотип 8, съдържащ човешкия ген *CNGB3*	Лечение на ахроматопсия, предизвикана от мутации в *CNGB3* гена
Croatian	Adeno-povezani virusni vektor serotipa 8 koji sadrži ljudski gen *CNGB3*	Liječenje akromatopsije uzrokovane mutacijama gena *CNGB3*
Czech	Adeno-asociovaný virus sérotypu 8 obsahující lidský gen *CNGB3*	Léčba achromatopsie způsobené mutacemi v genu *CNGB3*
Danish	Adenovirus associeret viral vektor serotype 8 indeholdende det humane gen *CNGB3*	Behandling af akromatopsi, forårsaget af mutationer i *CNGB3*-genet
Dutch	Adenovirus geassociëerde virale vector, serotype 8, welke het humane gen *CNGB3* bevat	Behandeling van door mutaties in het *CNGB3*-gen veroorzaakte achromatopsie
Estonian	Adenoviirusega seotud viirusvektor serotüüp 8, mis sisaldab inimese *CNGB3* geeni	Mutatsioonidest geenis *CNGB3* põhjustatud akromatopsia ravi
Finnish	Serotyypin 8 adenovirusvektori, jossa on ihmisen *CNGB3* -geeni	*CNGB3*-geenin mutaation aiheuttaman akromatopsian hoito
French	Vecteur viral adéno-associé de type 8 contenant le gène humain *CNGB3*	Traitement de l'achromatopsie causée par des mutations du gène *CNGB3*
German	Adenovirus-assoziierter viraler Vektor Serotyp 8, der das humane *CNGB3* Gen enthält	Behandlung von Achromatopsie infolge von Mutationen des *CNGB3*-Gens
Greek	Ιικός φορέας σχετιζομενος με αδενοϊό ορότυπου 8 που περιέχει το ανθρώπινο γονίδιο *CNGB3*	Θεραπεία της αχρωματοψίας που προκαλείται από μεταλλαγές στο γονίδιο *CNGB3*
Hungarian	Humán *CNGB3* gént tartalmazó 8-es szerotípusú adenovírus vektor	*CNGB3* gén mutációk által okozott achromatopsia kezelése
Italian	Vettore virale adenovirus-associato del serotipo 8 contenente il gene umano *CNGB3*	Trattamento della acromatopsia causata da mutazioni del gene *CNGB3*
Latvian	Adenovīrusa saistīts 8. seroptipa vīrusa vektors, kas satur cilvēka *CNGB3* gēnu	*CNGB3* gēna mutācijas izraisītas ahromatopsijas ārstēšana
Lithuanian	Adeno-asocijuoto viruso vektoriaus 8 serotipas, turintis žmogaus *CNGB3* geną	Achromatopsijos, sąlygotos *CNGB3* geno mutacijų, gydymas
Maltese	Vektor virali assoċjat ma' l-adenovirus tas-serotip 8 li għandu l-ġene uman *CNGB3*	Kura tal-akromatopsija kkawżata minn mutazzjonijiet fil-ġene tas-*CNGB3*
Polish	Wektor adenowirusowy serotyp 8 zawierający ludzki gen *CNGB3*	Leczenie ślepoty barw wywołanej mutacjami genu *CNGB3*
Portuguese	Vector viral adeno-associado de serotipo 8 contendo o gene humano *CNGB3*	Tratamento de acromatopsia causada pelas mutações no gene *CNGB3*

[1] At the time of designation

Language	Active ingredient	Indication
Romanian	Vector viral adeno-asociat de serotip 8 ce conţine gena umană *CNGB3*	Tratamentul acromatopsiei produse de mutaţii ale genei *CNGB3*
Slovak	Vírusový vektor spojený s adenovírusom sérotyp 8 obsahujúci ľudský gén *CNGB3*	Liečba achromatopsie spôsobenej mutáciami v géne *CNGB3*
Slovenian	Adenovirusom sorodni virusni vektor serotipa 8, ki vsebuje človeški gen *CNGB3*	Zdravljenje akromatopsije, ki jo povzročijo mutacije gena *CNGB3*
Spanish	Vector vírico adenoasociado del serotipo 8 que conteniene el gen humano *CNGB3*	Tratamiento de la acromatopsia causada por mutaciones en *CNGB3*
Swedish	Adenoassocierad virusvektor av serotyp 8, innehållande den humana *CNGB3* genen	Behandling av akromatopsi orsakad av mutationer i *CNGB3*-genen
Norwegian	Adenoassosiert virusvektor serotype 8 som inneholder det humane genet *CNGB3*	Behandling av akromatopsi forårsaket av mutasjoner i *CNGB3*-genet
Icelandic	Adenóveiru tengd veiruferja af sermisgerð 8 sem inniheldur manna *CNGB3* gen	Meðferð á allitblindu af völdum stökkbreytinga í *CNCB3* geninu