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Public summary of opinion on orphan designation

Adenovirus associated viral vector serotype 2/8 containing the human *CNGA3* gene for the treatment of achromatopsia

On 31 July 2018, orphan designation (EU/3/18/2042) was granted by the European Commission to MeiraGTx UK II Limited, United Kingdom, for adenovirus associated viral vector serotype 2/8 containing the human *CNGA3* gene for the treatment of achromatopsia.

What is achromatopsia?

Achromatopsia is an inherited disease of the eye that leads to reduced visual acuity (how well a person can see), colour blindness, severe photophobia (eye discomfort in bright or even normal light) and nystagmus (fast involuntary eye movements).

Achromatopsia affects the retina, the light-sensitive surface at the back of the eye. In patients with achromatopsia, retina cells called 'cone photoreceptors' do not work normally. These cells are needed to see in bright light and for seeing colours.

Achromatopsia is often caused by mutations (changes) in the *CNGA3* gene. It is a long-term debilitating disease because it affects how well a person can see, which may limit everyday activities.

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

At the time of designation, achromatopsia affected approximately 0.3 in 10,000 people in the European Union (EU). This was equivalent to a total of around 16,000 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 achromatopsia. Patients with the condition were given glasses with red-coloured lenses to reduce discomfort from bright light, as well as glasses to help with reading and seeing at long distance.

[*]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 517,400,000 (Eurostat 2018).

30 Churchill Place ● Canary Wharf ● London E14 5EU ● United Kingdom
Telephone +44 (0)20 3660 6000 **Facsimile** +44 (0)20 3660 5555
Send a question via our website www.ema.europa.eu/contact

How is this medicine expected to work?

In some patients with achromatopsia, a protein involved in the normal functioning of cone photoreceptors, called the CNGA3 protein, does not work properly due to mutations (changes) in the gene responsible for producing it.

This medicine is made up of a virus that contains normal copies of the *CNGA3* gene. When injected into the patient's eyes, it is expected that the virus will carry the *CNGA3* gene into photoreceptor cells, so that a working CNGA3 protein can be produced. This is expected to enable cone photoreceptors to work properly, thereby improving the symptoms of the disease.

The 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 in patients with achromatopsia had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for achromatopsia 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 21 June 2018 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	Adeno-associated viral vector serotype 2/8 containing the human *CNGA3* gene	Treatment of achromatopsia
Bulgarian	Адено-свързан вирусен вектор серотип 2/8, съдържащ човешкия *CNGA3* ген	Лечение на ахроматопсия
Croatian	Adeno vezani virusni vektor serotipa 2/8 koji sadrži ljudski gen *CNGA3*	Liječenje akromatopsije
Czech	Adeno-asociovaný virový vektor sérotypu 2/8, který obsahuje lidský gen *CNGA3*	Léčba achromatopsie
Danish	Adenoassocieret virusvektor serotype 2/8 som indeholdender det humane *CNGA3*-gen	Behandling af akromatopsi
Dutch	Adeno-geassocieerde virale vector serotype 2/8 die het humane *CNGA3* gen bevat	Behandeling van achromatopsie
Estonian	Inimese *CNGA3* geeni sisaldav adeno-assotsieerunud viirusvektori serotüüp 2/8	Akromatopsia ravi
Finnish	Adenoassosioitu serotyypin 2/8 virusvektori, joka sisältää ihmisen *CNGA3* geenin	Akromatopsian hoito
French	Vecteur viral adéno-associé du sérotype 2/8 qui contient le gène *CNGA3* humain	Traitement de l'achromatopsie
German	Adeno-assoziierter viraler Vektor Serotyp 2/8, der das humane *CNGA3* Gen enthält	Behandlung von Achromatopsie
Greek	Αδενο-σχετιζόμενος ιικός φορέας οροτύπου 2/8 που περιέχει το ανθρώπινο γονίδιο *CNGA3*	Θεραπεία της αχρωματοψίας
Hungarian	Adeno-asszociált virus vektor szerotípus 2/8, mely a humán *CNGA3* gént tartalmazza	Achromatopsia kezelése
Italian	Vettore virale ricombinante adeno-associato del sierotipo 2/8, contenente il gene *CNGA3* umano	Trattamento della acromatopsia
Latvian	Adeno-asociētā vīrusa vektora 2./8. serotips, kas satur cilvēka *CNGA3* gēnu	Ahromatopsijas ārstēšana
Lithuanian	Adenoasocijuoto viruso vektoriaus 2/8 serotipas, turintis žmogaus *CNGA3* geną	Achromatopsijos gydymas
Maltese	Vettur virali tas-serotip 2/8 assoċjat ma' adeno li fih il-ġene *CNGA3* uman	Trattament tal-akromatopsija
Polish	Wektor wirusowy związany z adenowirusami serotypu 2/8, zawierający ludzki gen *CNGA3*	Leczenie achromatopsji
Portuguese	Vetor viral adeno-associado de serotipo 2/8 contendo o gene humano *CNGA3*	Tratamento de acromatopsia
Romanian	Vector viral adeno-asociat de serotip 2/8 ce conţine gena umană *CNGA3*	Tratamentul acromatopsiei
Slovak	Adeno-asociovaný vírusový vektor sérotypu 2/8, ktorý obsahuje ľudský gén *CNGA3*	Liečba achromatopsie

[1] At the time of designation

Language	Active ingredient	Indication
Slovenian	Adenoasocirani virusni vektor serotipa 2/8, ki vsebuje človeški *CNGA3* gen	Zdravljenje akromatopsije
Spanish	Vector viral adenoasociado recombinante serotipo 2/8 que contiene el gen *CNGA3* humano	Tratamiento de la acromatopsia
Swedish	Adeno-associerad virusvektor 2/8 som innehåller den humana *CNGA3*-genen	Behandling av akromatopsi
Norwegian	Adenoassosiert virusvektor serotype 2/8 som inneholder det humane *CNGA3*-genet	Behandling av akromatopsi
Icelandic	Adeno-tengd veiru ferja af sermisgerð 2/8 sem inniheldur manna *CNGA3* gen	Meðferð á allitblindu