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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant adeno-associated viral vector containing the human *CNGB3* gene for the treatment of achromatopsia caused by mutations in the *CNGB3* gene

On 8 February 2013 orphan designation (EU/3/13/1099) was granted by the European Commission to TMC Pharma Services Ltd, United Kingdom, for recombinant adeno-associated viral vector containing the human *CNGB3* gene for the treatment of achromatopsia caused by mutations in the *CNGB3* gene.

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 (increased sensitivity to light) and nystagmus (fast involuntary eye movements). In patients with achromatopsia, cells in the retina (the light-sensitive surface at the back of the eye) called 'cone photoreceptors', which provide vision in bright light including colour vision, do not function normally.

Achromatopsia is often caused by mutations (defects) 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 7,600 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. This represents a population of 509,000,000 (Eurostat 2013).

7 Westferry Circus ● Canary Wharf ● London E14 4HB ● United Kingdom
Telephone +44 (0)20 7418 8400 **Facsimile** +44 (0)20 7523 7040
E-mail info@ema.europa.eu **Website** www.ema.europa.eu
An agency of the European Union

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 sunglasses 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, is lacking due to defects in the gene responsible for producing it. The 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 the 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 caused by mutations in the *CNGB3* gene 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. Orphan designation of the medicine had been granted in the United States of America for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 9 January 2013 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:

TMC Pharma Services Ltd
Lodge Farm Barn
Elvetham Park Estate
Fleet Road
Hartley Wintney
Hampshire RG27 8AS
United Kingdom
Telephone: +44 125 2842 255
Telefax: +44 125 2842 277
E-mail: info@tmcpharma.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[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Recombinant adeno-associated viral vector containing the human *CNGB3* gene	Treatment of achromatopsia caused by mutations in the *CNGB3* gene
Bulgarian	Рекомбинантен адено-асоцииран вирусен вектор, съдържащ човешкия ген *CNGB3*	Лечение на ахроматопсия, предизвикана от мутации в *CNGB3* гена
Czech	Rekombinantní adeno-asociovaný virový vektor obsahující lidský gen *CNGB3*	Léčba achromatopsie způsobené mutacemi v genu *CNGB3*
Danish	Rekombinant adeno-associeret viral vektor som indeholder det humane *CNGB3*-gen	Behandling af akromatopsi, forårsaget af mutationer i *CNGB3*-genet
Dutch	Recombinant adeno-geassocieerde virale vector welke het humaan *CNGB3* gen	Behandeling van door mutaties in het *CNGB3*-gen veroorzaakte achromatopsie
Estonian	Rekombinantne adenoviirusega seotud viirusvektor, mis sisaldab inimese *CNGB3* geeni	Mutatsioonidest geenis *CNGB3* põhjustatud akromatopsia ravi
Finnish	Rekombinantti adeno-pohjainen virusvektori, joka sisältää ihmisen *CNGB3*-geenin	*CNGB3*-geenin mutaation aiheuttaman akromatopsian hoito
French	Vecteur viral recombinant adeno-associe contenant le gène humain *CNGB3*	Traitement de l'achromatopsie causée par des mutations du gène *CNGB3*
German	Rekombinanter Adeno-assoziierter viraler Vektor, der das menschliche Gen *CNGB3 enthält*	Behandlung von Achromatopsie infolge von Mutationen des *CNGB3* -Gens
Greek	Ανασυνδυασμένοςιϊκός φορέας σχετιζόμενος με αδενοϊό που περιέχει το ανθρώπινο γονίδιο *CNGB3*	Θεραπεία της αχρωματοψίας που προκαλείται από μεταλλαγές στο γονίδιο *CNGB3*
Hungarian	Humán *CNGB3* gént tartalmazó rekombináns adeno vitussal asszociált virális vektor	*CNGB3* gén mutációk által okozott achromatopsia kezelése
Italian	Vettore virale ricombinante adeno-associato contenente il gene della *CNGB3* umana	Trattamento della acromatopsia causata da mutazioni del gene *CNGB3*
Latvian	Rekombinants adenoasociētā vīrusa vektors, kas satur cilvēka *CNGB3* gēnu	*CNGB3* gēna mutācijas izraisītas ahromatopsijas ārstēšana
Lithuanian	Rekombinantinis adeno-asocijuoto viruso vektorius, pernešantis žmogaus *CNGB3* geną	Achromatopsijos, sąlygotos *CNGB3* geno mutacijų, gydymas
Maltese	Vettur virali rikombinanti adeno-assoċjat li fih il-ġene *CNGB3* uman	Kura tal-akromatopsija kkawżata minn mutazzjonijiet fil-ġene tas-*CNGB3*

[1] At the time of designation

Language	Active ingredient	Indication
Polish	Rekombinowany wektor adenowirusowy zawierający ludzki gen *CNGB3*	Leczenie ślepoty barw wywołanej mutacjami genu *CNGB3*
Portuguese	Vetor recombinante viral adeno-associado, contendo o um gene humano *CNGB3*	Tratamento de acromatopsia causada pelas mutações no gene *CNGB3*
Romanian	Vector viral adeno-asociat recombinant, care conţine gena umană care codifică *CNGB3*	Tratamentul acromatopsiei produse de mutaţii ale genei *CNGB3*
Slovak	Rekombinantný adeno-asociovaný vírusový vektor obsahujúci ľudský gén *CNGB3*	Liečba achromatopsie spôsobenej mutáciami v géne *CNGB3*
Slovenian	Rekombinantni adenovirusni vektor z genom za humanim genom *CNGB3*	Zdravljenje akromatopsije, ki jo povzročijo mutacije gena *CNGB3*
Spanish	Vector viral adenoasociado recombinante que contiene el gen humano de la *CNGB3*	Tratamiento de la acromatopsia causada por mutaciones en el gen *CNGB3*
Swedish	Rekombinant adeno-associerad viral vektor som innehåller den humana *CNGB3*-genen	Behandling av akromatopsi orsakad av mutationer i *CNGB3*-genen
Norwegian	Rekombinant adenoassosiert virusvektor som inneholder det humane *CNGB3*-genet	Behandling av akromatopsi forårsaket av mutasjoner i *CNGB3*-genet
Icelandic	Raðbrigða adenótengd veiru ferja sem inniheldur manna *CNGB3* gen.	Meðferð á allitblindu af völdum stökkbreytinga í *CNCB3* geninu