22 January 2018
EMA/754024/2017

Public summary of opinion on orphan designation

Adenovirus associated viral vector serotype 8 containing the human *AIPL1* gene for the treatment of Leber's congenital amaurosis

On 12 December 2017, orphan designation (EU/3/17/1950) was granted by the European Commission to MeiraGTx UK II Limited, United Kingdom, for adenovirus associated viral vector serotype 8 containing the human *AIPL1* gene for the treatment of Leber's congenital amaurosis.

What is Leber's congenital amaurosis?

Leber's congenital amaurosis is an inherited disease characterised by loss of sight at birth or soon after birth. The disease is linked to a number of genetic mutations (changes), which affect the normal development of the light-sensitive cells in the eye.

Leber's congenital amaurosis is a long-term debilitating disease due to progressive loss of vision.

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

At the time of designation, Leber's congenital amaurosis affected less than 1 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 52,000 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 Leber's congenital amaurosis. Patients with the condition usually received regular medical follow up, vision aids and genetic counselling on the risks of passing the condition on to their children.

*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 515,700,000 (Eurostat 2017).

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How is this medicine expected to work?

This medicine has been designed to treat Leber's congenital amaurosis type 4. This form of the disease is caused by mutations in a gene called *AIPL1* which is involved in the production of an enzyme that is important for the correct functioning of the light-sensitive cells of the eye.

This medicine is made of a virus that contains normal copies of the *AIPL1* gene. When injected into the patient's eye, it is expected that the virus will carry the *AIPL1* gene into the light-sensitive cells of the eye, restoring normal cell function and thereby helping to improve the patient's sight and reduce other 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?

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

At the time of submission of the application for orphan designation, no clinical trials with the medicine in patients with Leber's congenital amaurosis had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for Leber's congenital amaurosis 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 31 October 2017 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 *AIPL1* gene	Treatment of Leber's congenital amaurosis
Bulgarian	Аденовирусно-асоцииран вирусен вектор серотип 8, съдържащ човешкия ген *AIPL1*	Лечение на вродена амавроза на Лебер
Croatian	Adenovirus svezan s virusnim vektorom serotipa 8 koji sadržava humani gen *AIPL1*	Liječenje Leberove kongenitalne amauroze
Czech	Adeno-asociovaný virus sérotypu 8 obsahující lidský gen *AIPL1*	Léčba Leberovy vrozené slepoty
Danish	Adeno-associeret viral vektor serotype 8 indeholdende det humane gen *AIPL1*	Behandling af Lebers kongenitte amaurose
Dutch	Adenovirus geassocieerde virale vector, serotype 8, welke het humane gen *AIPL1* bevat	Behandeling van amaurosis congenita van Leber
Estonian	Adenoviirusega seotud viirusvektor serotüüp 8, mis sisaldab inimese *AIPL1* geeni	Leberi tüüpi pärilik amauroos (nägemisnärvi kõhetusest tingitud pimedus) ravii
Finnish	Serotyypin 8 adenovirusvektori, jossa on ihmisen *AIPL1*-geeni	Leberin synnynnäisen amauroosin (sokeus) hoito
French	Vecteur viral adéno-associé de type 8 contenant le gène humain *AIPL1*	Traitement de l'amaurose congénitale de Leber
German	Adenovirus-assoziierter viraler Vektor Serotyp 8, der das humane *AIPL1* Gen enthält	Behandlung der Leberschen Kongenitalen Amaurose
Greek	Αδενο-σχετιζόμενος ιϊκός φορέας οροτύπου8 που περιέχει το ανθρώπινο γονίδιο *AIPL1*	Θεραπεία τής συγγενους αμαύρωσης του Leber
Hungarian	Humán *AIPL1* gént tartalmazó 8-as szerotípusú adenovírus vektor	Leber-féle hereditaer opticus atrophia kezelése
Italian	Vettore virale adenovirus-associato del serotipo 8 contenente il gene umano *AIPL1*	Trattamento dell'amaurosi congenita di Leber
Latvian	Adenovīrusa saistītā virālā vektora 8. serotips, kas satur cilvēka *AIPL1* gēnu	Iedzimta Lēbera akluma ārstēšana
Lithuanian	Adeno - asocijuoto viruso vektoriaus 8 serotipas, turintis žmogaus *AIPL1* geną	Įgimtos Lėberio amaurozės gydymas
Maltese	Vettur virali assoċjat mal-adenovirus tas-serotip 8 li għandu l-ġene tal-bniedem *AIPL1*	Kura ta' l-amawrożi konġenitali ta' Leber
Polish	Wektor wirusowy związany z adenowirusami serotypu 8 zawierający ludzki gen *AIPL1*	Leczenie wrodzonej ślepoty Lebera
Portuguese	Vetor viral adeno-associado de serotipo 8 contendo o gene humano *AIPL1*	Tratamento do Amaurose Congénita de Leber
Romanian	Vector viral adeno-asociat de serotip 8 ce conţine gena umană *AIPL1*	Tratamentul amaurozei congenitale Leber
Slovak	Adeno-asociovaný vírusový vektor sérotypu 8 obsahujúci ľudský gén *AIPL1*	Liečba Leberovej vrodenej amaurózy

[1] At the time of designation

Language	Active ingredient	Indication
Slovenian	Adenovirusom pridruženi virusni vektor serotipa 8, ki vsebuje človeški gen *AIPL1*	Zdravljenje Leberjeve vrojene amavroze
Spanish	Vector víral adenoasociado del serotipo 8 que conteniene el gen humano *AIPL1*	Tratamiento de la amaurosis congénita de Leber
Swedish	Adenoassocierad virusvektor av serotyp 8, innehållande den humana *AIPL1* genen	Behandling av Lebers kongenitala amauros
Norwegian	Adenoassosiert virusvektor serotype 8 som inneholder det humane genet *AIPL1*	Behandling av Lebers kongenitte amaurose
Icelandic	Adenóveiru tengd veiruferja af sermisgerð 8 sem inniheldur manna *AIPL1* gen	Meðferð á Leber meðfæddri blindu