11 March 2015
EMA/COMP/97253/2014 Rev.1
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Adeno-associated viral vector serotype 8 containing the human *GUCY2D* gene for the treatment of Leber's congenital amaurosis

First publication	5 May 2014
Rev.1: sponsor's change of address	11 March 2015
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 26 March 2014, orphan designation (EU/3/14/1256) was granted by the European Commission to Fondazione Telethon, Italy, for adeno-associated viral vector serotype 8 containing the human *GUCY2D* 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 defects, 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 51,000 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.
At the time of designation, this represented a population of 512,900,000 (Eurostat 2014).

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

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 Leber's congenital amaurosis 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?

One form of Leber's congenital amaurosis is caused by a defect in the gene *GUCY2D*, which is responsible for the production of a protein called RetGC1. This protein has a key role in the correct functioning of the light-sensitive cells of the eye.

This medicine is made of a virus that contains a normal copy of the *GUCY2D* gene. When injected into the patient's eye, it is expected that the virus will carry the *GUCY2D* gene into the light-sensitive cells of the eye, enabling the cells to work 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?

The effects of this 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 6 February 2014 recommending the granting of this designation.

_____

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:

Fondazione Telethon
Via Varese 16
00185 Roma
Italy
Tel. +39 06 44 01 51
Fax +39 06 44 01 55 21
E-mail: info@telethon.it

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 8 containing the human *GUCY2D* gene	Treatment of Leber's congenital amaurosis
Bulgarian	аденосвързан вирусен вектор серотип 8, съдържащ човешки *GUCY2D* ген	Лечение на вродена амавроза на Лебер
Czech	Sérotyp 8 adeno asociovaný virový vektor, obsahující lidský gen *GUCY2D*	Léčba Leberovy vrozené slepoty
Croatian	Adeno-povezani virusni vektor serotip 8 koji sadrži ljudski gen *GUCY2D*	Liječenje Leberove kongenitalne amauroze
Danish	Adenoassocieret viral vektor serotype 8 indeholdende det humane gen *GUCY2D*	Behandling af Lebers kongenitte amaurose
Dutch	Adenogeassocieerde virale vector, serotype 8, welke het humane gen *GUCY2D* bevat	Behandeling van amaurosis congenita van Leber
Estonian	Adenoga assotsieeruv viirusvektori serotüüp 8, mis sisaldab inimese *GUCY2D* geeni	Leberi tüüpi pärilik amauroos (nägemisnärvi kõhetusest tingitud pimedus) ravii
Finnish	Adenoassosioitu virusvektori, serotyyppiä 8, joka sisältää ihmisen *GUCY2D* geenin	Leberin synnynnäisen amauroosin (sokeus) hoito
French	Vecteur viral adéno-associé de sérotype 8 contenant le gène humain *GUCY2D*	Traitement de l'amaurose congénitale de Leber
German	Das humane *GUCY2D* Gen beinhaltender, adeno-assoziierter viraler Vektor Serotyp 8	Behandlung der Leberschen Kongenitalen Amaurose
Greek	Αδενο-σχετιζόμενος ιικός φορέας ορότυπου 8 που περιέχει το ανθρώπινο γονίδιο *GUCY2D*	Θεραπεία της συγγενους αμαύρωσης του Leber
Hungarian	Humán *GUCY2D* gént tartalmazó 8-es szerotípusú adeno-asszociált vírus vektor	Leber-féle hereditaer opticus atrophia kezelése
Italian	Vettore virale adeno-associato del serotipo 8 contenente il gene umano *GUCY2D*	Trattamento dell'amaurosi congenita di Leber
Latvian	Adeno-saistītā virālā vektora 8 serotips, kas satur cilvēka *GUCY2D* gēnu	Iedzimta Lēbera akluma ārstēšana
Lithuanian	Adeno-asocijuotas viruso vektoriaus 8 serotipas, pernešantis žmogaus *GUCY2D* geną	Įgimtos Lėberio amaurozės gydymas

[1] At the time of designation

Language	Active ingredient	Indication
Maltese	Vettur imnissel mill-adenovirus tas-serotip 8 li fih il-ġene uman *GUCY2D*	Kura ta' l-amawrożi konġenitali ta' Leber
Polish	Wektor adenowirusowy serotypu 8 zawierający ludzki gen *GUCY2D*	Leczenie wrodzonej ślepoty Lebera
Portuguese	Vector viral adeno-associado de serotipo 8 contendo o gene humano *GUCY2D*	Tratamento do Amaurose Congénita de Leber
Romanian	Vector viral adeno-asociat de serotip 8 conţinând gena umană *GUCY2D*	Tratamentul amaurozei congenitale Leber
Slovak	Adeno-asociovaný vírusový vektor sérotypu 8 obsahujúci l'udský gén *GUCY2D*	Liečba Leberovej vrodenej amaurózy
Slovenian	Adenovirusni pridruženi virusni vektor serotipa 8, ki vsebuje človeški gen *GUCY2D*	Zdravljenje Leberjeve vrojene amavroze
Spanish	Vector viral adenoasociado del serotipo 8 que conteniene el gene humano *GUCY2D*	Tratamiento de la amaurosis congénita de Leber
Swedish	Adenoassocierad virusvektor serotyp 8, innehållande den mänskliga genen *GUCY2D*	Behandling av Lebers kongenitala amauros
Norwegian	Adenoassosiert virusvektor serotype 8 som inneholder det humane genet *GUCY2D*	Behandling av Lebers kongenitte amaurose
Icelandic	Adenotengd veiruferja af sermisgerð8 sem inniheldur manna *GUCY2D* gen	Meðferð á Leber meðfæddri blindu