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EMA/COMP/171945/2011 Rev. 2
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

9-*cis*-Retinyl acetate for the treatment of Leber's congenital amaurosis

On 13 May 2011, orphan designation (EU/3/11/861) was granted by the European Commission to ORS Oxford Ltd, United Kingdom, for 9-*cis*-retinyl acetate for the treatment of Leber's congenital amaurosis. The sponsorship was transferred to QLT Ophthalmics (UK), Ltd, United Kingdom, in November 2011.

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 was estimated to affect less than 1 in 10,000 people in the European Union (EU)*. This is 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).

What treatments are available?

At the time of submission of the application for orphan 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 children and regular medical follow up.

*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 506,300,000 (Eurostat 2011).

How is this medicine expected to work?

11-*cis*-Retinal is a substance derived from vitamin A that the specialised light-sensitive photoreceptor cells in the retina need, to be able to translate light into nerve impulses. In Leber's congenital amaurosis, the retina does not produce or regenerate enough of 11-*cis*-retinal and the photoreceptor cells become depleted and damaged.

9-*cis*-Retinyl acetate is expected to be taken by mouth and converted into 9-*cis*-retinal in the retina. 9-*cis*-Retinal is similar to the deficient 11-*cis*-retinal and is expected to replace it, thus enabling the photoreceptor cells to function properly.

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 9-*cis*-retinyl acetate in experimental models was ongoing.

At the time of submission of the application for orphan designation, clinical trials with 9-*cis*-retinyl acetate in patients with Leber's congenital amaurosis were ongoing.

At the time of submission, 9-*cis*-retinyl acetate 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 9 February 2011 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:

QLT Ophthalmics (UK), Ltd
c/o Hackwood Secretaries Limited
One Silk Street
London EC2Y 8HQ
United Kingdom
tel: +44 (0)20 7456 2000
fax: +44 (0)20 7456 2222
e-mail: emma.ash@linklaters.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 languages¹, Norwegian and Icelandic

Language	Active ingredient	Indication
English	9- <i>cis</i> -Retinyl acetate	Treatment of Leber's congenital amaurosis
Bulgarian	9-цис-рeтинил ацетат	Лечение на вродена амавроза на Лебер
Czech	9- <i>cis</i> -retinylacetát	Léčba Leberovy vrozené slepoty
Danish	9- <i>cis</i> -retinylacetat	Behandling af Lebers kongenitte amaurose
Dutch	9- <i>cis</i> -retinylacetaat	Behandeling van amaurosis congenita van Leber
Estonian	9- <i>cis</i> -retinülatsetaat	Leberi tüüpi pärilik amauroos (nägemisnärvi kõhetusest tingitud pimedus) ravii
Finnish	9- <i>cis</i> -retinyyliasetaatti	Leberin synnynnäisen amauroosin (sokeus) hoito
French	9 <i>cis</i> rétinyl acetate	Traitement de l'amaurose congénitale de Leber
German	9- <i>cis</i> -Retinylacetat	Behandlung der Leberschen Kongenitalen Amaurose
Greek	9- <i>cis</i> -οξεική ρετινόλη	Θεραπεία τής συγγενούς αμαύρωσης του Leber
Hungarian	9- <i>cisz</i> -retinil-acetát	Leber-féle hereditaer opticus atrophia kezelése
Italian	9- <i>cis</i> -retinil acetato	Trattamento dell'amaurosi congenita di Leber
Latvian	9- <i>cis</i> -retinilacetāts	Iedzimta Lēbera akluma ārstēšana
Lithuanian	9- <i>cis</i> -retinilacetatas	Įgimtos Lėberio amaurozės gydymas
Maltese	9- <i>cis</i> -retinyl acetate	Kura ta' l-amawroži konġenitali ta' Leber
Polish	Octan 9- <i>cis</i> -retinylu	Leczenie wrodzonej ślepoty Lebera
Portuguese	Acetato de 9- <i>cis</i> -retinil	Tratamento do Amaurose Congénita de Leber
Romanian	9- <i>cis</i> -retinilacetat	Tratamentul amaurozei congenitale Leber
Slovak	9- <i>cis</i> -retinylacetát	Liečba Leberovej vrodenej amaurózy
Slovenian	9- <i>cis</i> -retinil acetat	Zdravljenje Leberjeve vrojene amavroze
Spanish	9- <i>cis</i> -retinil acetato	Tratamiento de la amaurosis congénita de Leber
Swedish	9- <i>cis</i> -retinylacetat	Behandling av Lebers kongenitala amauros
Norwegian	9- <i>cis</i> -retinylacetat	Behandling av Lebers kongenitte amaurose
Icelandic	9- <i>cis</i> -retinýlasetat	Meðferð á Leber meðfæddri blindu

¹ At the time of designation