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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Ataluren for the treatment of aniridia

On 9 October 2015, orphan designation (EU/3/15/1561) was granted by the European Commission to PTC Therapeutics International Limited, Ireland, for ataluren for the treatment of aniridia.

What is aniridia?

Aniridia is a rare inborn disorder in which the tissue that makes up the iris (the coloured part of the eye) is underdeveloped or completely missing. This means that the eye is not able to adjust to differing levels of light. Aniridia affects both eyes and causes poor vision and increased sensitivity to light. The disorder may be associated with other changes to other parts of the eye, either from birth or developing over time. Other senses may also be affected, including reduced sense of smell and taste, and hearing difficulties.

Aniridia is almost always caused by a mutation (change) in a gene named *PAX6*, which is involved in the development of the eye and other tissues. Patients may also have other genetic problems.

Aniridia is a long-term debilitating condition due to progressive loss of sight.

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

At the time of designation, aniridia affected approximately 0.2 in 10,000 people in the European Union (EU). This was equivalent to a total of around 10,000 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 method had been authorised in the EU to treat aniridia. Treatment of patients with aniridia primarily involved supportive treatments such as spectacles, tinted lenses to reduce damage to the retina, surgery for cataracts (a complication of aniridia) and measures to reduce increased pressure in the eye.

[*]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 512,900,000 (Eurostat 2015).

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How is this medicine expected to work?

The *PAX6* gene is responsible for producing a protein required for normal development of eye tissues, including the iris. Some patients with aniridia have a change in the *PAX6* gene (a 'nonsense mutation') that leads to production of the protein being stopped short before it is complete. Ataluren works in these patients by enabling the protein-making apparatus in cells to move past the mutated gene, allowing the cells to produce a functional protein.

What is the stage of development of this medicine?

The effects of ataluren have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with ataluren in patients with aniridia had been started.

Ataluren has been authorised as Translarna in the EU for the treatment of Duchenne muscular dystrophy due to a nonsense mutation.

At the time of submission, the medicine was not authorised anywhere in the EU for aniridia. Orphan designation for ataluren has been granted in the United States for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 3 September 2015 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

For details of the current sponsor of the orphan designation please refer to the information on the main web page of this Public Summary of Opinion.

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	Ataluren	Treatment of aniridia
Bulgarian	Аталурен	Лечение на аниридия
Croatian	Ataluren	Liječenje aniridije
Czech	Ataluren	Léčba aniridie
Danish	Ataluren	Behandling af aniridi
Dutch	Ataluren	Behandeling van aniridie
Estonian	Ataluren	Aniriidia ravi
Finnish	Atalureeni	Aniridian hoito
French	Ataluren	Traitement de l'aniridie
German	Ataluren	Behandlung von Aniridie
Greek	Ατσλουρένη	Θεραπεία ανιριδίας
Hungarian	Ataluren	Aniridia kezelése
Italian	Ataluren	Trattamento dell'aniridia
Latvian	Atalurēns	Anirīdijas ārstēšana
Lithuanian	Atalurenas	Aniridijos gydymas
Maltese	Ataluren	Kura tal-aniridja
Polish	Ataluren	Leczenie aniridii
Portuguese	Atalurene	Tratamento da aniridia
Romanian	Ataluren	Tratamentul aniridiei
Slovak	Ataluren	Liečba anirídie
Slovenian	Ataluren	Zdravljenje aniridije
Spanish	Ataluren	Tratamiento de la aniridia
Swedish	Ataluren	Behandling av aniridi
Norwegian	Ataluren	Behandling av aniridi
Icelandic	Atalúren	Meðferð við lithimnuleysi

[1] At the time of designation