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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Ataluren for treatment of Becker muscular dystrophy

First publication	19 July 2012
Rev.1: sponsor's change of address	13 September 2013
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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 4 July 2012, orphan designation (EU/3/12/1010) was granted by the European Commission to PTC Therapeutics Limited, United Kingdom, for ataluren for treatment of Becker muscular dystrophy.

What is Becker muscular dystrophy?

Becker muscular dystrophy is an inherited disease that gradually causes the muscles to become weaker. The muscle weakness usually starts in the hips and legs and may later on involve the chest and the heart. The disease mainly affects boys, and usually starts between the ages of 10 and 15.

Becker muscular dystrophy is caused by abnormalities in the gene responsible for the production of dystrophin, a protein that forms an important component of muscle fibres. As the patients do not have enough of the functional protein, the muscle fibres gradually break down leading to muscle weakness.

Becker muscular dystrophy causes long-term disability and is life threatening because of its effects on the heart and the respiratory muscles (muscles that are used to breathe).

What is the estimated number of patients?

At the time of designation, Becker muscular dystrophy 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

*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. At the time of designation, this represented a population of 509,000,000 (Eurostat 2012).



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 the treatment of Becker muscular dystrophy. Patients received supportive treatment to relieve symptoms and improve the patient's general condition. In addition, corticosteroids were used in an attempt to improve symptoms, although they were not authorised for use in this disease.

How is this medicine expected to work?

Becker muscular dystrophy is caused by a number of genetic abnormalities. Some of these involve the presence of defective sequences in certain DNA threads which stop the production of dystrophin, leading to a truncated dystrophin protein that does not function properly.

This medicine is expected to work by enabling the protein-making apparatus in cells to 'read through' the affected DNA threads, allowing the cells to produce fully functional dystrophin.

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, clinical trials with ataluren in patients with Becker muscular dystrophy were ongoing.

At the time of submission, ataluren was not authorised anywhere in the EU for Becker muscular dystrophy 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 11 May 2012 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:

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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	Ataluren	Treatment of Becker muscular dystrophy
Bulgarian	Аталурен	Лечение на мускулна дистрофия тип Бекер
Czech	Ataluren	Léčba Beckerovy svalové dystrofie
Danish	Ataluren	Behandling af Beckers muskeldystrofi
Dutch	Ataluren	Behandeling van Becker spierdystrofie
Estonian	ataluren	Beckeri lihasdüstroofia ravi
Finnish	Atalureeni	Beckerin lihasdystrofian hoito
French	Ataluren	Traitement de la dystrophie musculaire de Becker
German	Ataluren	Behandlung der Muskeldystrophie Typ Becker
Greek	Αταλουρένη	Θεραπεία της μυϊκής δυστροφίας του Becker
Hungarian	Ataluren	Becker-féle izomdisztrófia kezelése
Italian	Ataluren	Trattamento della distrofia muscolare di Becker
Latvian	Atalurēns	Bekera muskuļu distrofijas ārstēšanai
Lithuanian	Atalurenas	Bekerio raumenų distrofijos gydymas
Maltese	Ataluren	Kura tad-distrofija muskolari ta' Becker
Polish	Ataluren	Leczenie dystrofii mięśniowej Beckera
Portuguese	Atalurene	Tratamento da distrofia muscular de Becker
Romanian	Ataluren	Tratamentul distrofiei musculare Becker
Slovak	Ataluren	Liečba Beckerovej svalovej dsystrofie
Slovenian	Ataluren	Zdravljenje Beckerjeve mišične distrofije
Spanish	Ataluren	Tratamiento de la distrofia muscular de Becker
Swedish	Ataluren	Behandling av Beckers muskeldystrofi
Norwegian	Ataluren	Behandling av Beckers muskeldystrofi
Icelandic	Atalúren	Meðferð við Beckers vöðvakyrkingi

¹ At the time of designation