15 March 2019
EMADOC-628903358-222

Public summary of opinion on orphan designation

Acetylleucine for treatment of ataxia telangiectasia

On 11 January 2019, orphan designation (EU/3/18/2124) was granted by the European Commission to IntraBio Limited, United Kingdom, for acetylleucine for treatment of ataxia telangiectasia.

What is ataxia telangiectasia?

Ataxia telangiectasia is an inherited disorder that affects parts of the brain, leading to an increasing inability to control movements (ataxia), speech problems and difficulty moving the eyes from side to side. The disorder also affects the immune system (the body's natural defences), increasing patients' risk of infections and certain blood cancers. Patients may also have telangiectasias, which are clusters of dilated blood vessels, which may occur in the white of the eyes.

Ataxia telangiectasia is a debilitating and life-threatening disease because of the worsening of symptoms over time and increased risk of infections and cancers.

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

At the time of designation, ataxia telangiectasia affected less than 0.25 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 13,000 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, there was no satisfactory treatment for ataxia telangiectasia authorised in the EU. Patients generally received speech therapy, physiotherapy and treatment for infections and other complications.

*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 517,400,000 (Eurostat 2018).

Official address Domenico Scarlattilaan 6 ● 1083 HS Amsterdam ● The Netherlands
Address for visits and deliveries Refer to www.ema.europa.eu/how-to-find-us
Send us a question Go to www.ema.europa.eu/contact **Telephone** +31 (0)88 781 6000 An agency of the European Union

How is this medicine expected to work?

The way acetylleucine works in ataxia telangiectasia is not clear but it is thought to stabilise nerve cells responsible for balance and for coordinating movement. This is expected to improve movement control and coordination in patients with the disease.

What is the stage of development of this medicine?

The effects of acetylleucine have been evaluated in experimental models.

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

At the time of submission, acetylleucine was not authorised anywhere in the EU for ataxia telangiectasia 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 December 2018 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:

- Contact details of the current sponsor for this orphan designation can be found on the EMA website.

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 orphan condition in all official EU languages[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Acetylleucine	Treatment of ataxia telangiectasia
Bulgarian	Ацетиллевцин	Лечение на атаксия телангиектазия
Croatian	Acetilleucin	Léčba ataxie telangiectasie
Czech	Acetylleucin	Liječenje ataksije-teleangiektazije
Danish	Acetylleucine	Behandling af ataxia telangiectasia
Dutch	Acetylleucine	Behandeling van ataxia telangiëctasie
Estonian	Atsetüülleutsiin	Teleangiektaasataksia ravi
Finnish	Asetyylileusiini	Ataksia-teleangiektasian hoito
French	Acétylleucine	Traitement de l'ataxie télangiectasie
German	Acetylleucin	Behandlung der Ataxia teleangiectatica/Louis-Bar-Syndrom
Greek	Ακετυλευκίνη	Θεραπεία της αταξίας-τηλαγγειεκτασίας
Hungarian	Acetilleucin	Ataxia-telangiectasia kezelése
Italian	Acetilleucina	Trattamento della Ataxia telangiectasia
Latvian	Acetilleicīns	Teleangektāziskās ataksijas ārstēšana
Lithuanian	Acetilleucinas	Ataksijos telangiektazijos gydymas
Maltese	Acetylleucine	Kura tal-atassja telanġektasija
Polish	Acetyloleucyna	Leczenie zespołu ataksji-teleangiektazji
Portuguese	Acetilleucina	Tratamento da ataxia-telangiectasia
Romanian	Acetilleucină	Tratamentul ataxiei telangiectaziei
Slovak	Acetylleucín	Liečba ataxie teleangiektázie
Slovenian	Acetillevcin	Zdravljenje ataksija-teleangiopatije
Spanish	Acetileucina	Tratamiento de la ataxia telangiectasia
Swedish	Acetylleucin	Behandling av ataxia telangiectasia
Norwegian	Acetylleucin	Behandling av ataxia telangiectasia
Icelandic	Asetýlleucín	Meðferð á ataxia telangiectasia

[1] At the time of designation