



EUROPEAN MEDICINES AGENCY
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Public summary of opinion on orphan designation

Ciclopirox for the treatment of congenital erythropoietic porphyria

On 17 January 2018, orphan designation (EU/3/17/1960) was granted by the European Commission to Atlas Molecular Pharma S.L., Spain, for ciclopirox for the treatment of congenital erythropoietic porphyria.

What is congenital erythropoietic porphyria?

Congenital erythropoietic porphyria is a genetic disease that affects the production of haem, one of the components of haemoglobin. Haemoglobin is the protein found in the red blood cells that carries oxygen around the body. As a result, toxic substances called porphyrins build up in the body, in particular in the red blood cells and the skin. This damages the red blood cells and causes anaemia (low red blood cell counts). When skin is exposed to sunlight, the porphyrins react with light and cause severe skin damage, including blisters, sores, thinning and disfigurement of the skin.

The disease is caused by mutations (changes) in the gene for an enzyme called uroporphyrinogen III synthase. This enzyme is involved in the production of haem. The mutations result in an altered enzyme that does not work properly, leading to the build-up of porphyrins.

Congenital erythropoietic porphyria is debilitating in the long term due to the severe skin damage and potentially life threatening due to anaemia and liver complications.

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

At the time of designation, congenital erythropoietic porphyria affected 0.1 in 10,000 people in the European Union (EU). This was equivalent to a total of around 5,000 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?

No satisfactory treatments were authorised in the EU for congenital erythropoietic porphyria at the time of designation.

^{*}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 515,700,000 (Eurostat 2017).



Strict avoidance of sunlight or other strong light is essential to manage the disease, as well as avoiding physical damage to the skin and meticulous skin care. Patients with severe disease may need blood transfusions.

How is this medicine expected to work?

The way ciclopirox works is not well understood; however, it appears that the medicine attaches to the altered uroporphyrinogen III synthase. This helps to stabilise this enzyme, improving its activity and so reducing the production of porphyrins in the body.

What is the stage of development of this medicine?

The effects of the medicine have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with ciclopirox in patients with congenital erythropoietic porphyria had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for congenital erythropoietic porphyria or designated as an orphan medicinal product elsewhere for this condition.

Ciclopirox is authorised in some European countries for the treatment of fungal infections on the skin and nails.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 7 December 2017 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 EMA website, on the medicine's [rare disease designations page](#).

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	Ciclopirox	Treatment of congenital erythropoietic porphyria
Bulgarian	Циклопирокс	Лечение на вродена еритропоетична порфирия
Croatian	Ciklopiroks	Liječenje kongenitalne eritropoetske porfirije
Czech	Cyklopirox	Léčba vrozené erythropoetické porfyrie
Danish	Ciclopirox	Behandling af kongenit erythropoietisk porfyri
Dutch	Ciclopirox	Behandeling van congenitale erythropoëtische porphyrie
Estonian	Tsiklopiroks	Kaasasündinud erütropoeetilise porfüüria ravi
Finnish	Siklopiroksi	Synnynnäisen erythropoieettisen porfyrian hoito
French	Ciclopirox	Traitement de la porphyrie érythropoïétique congénitale
German	Ciclopirox	Behandlung der kongenitalen erythropoetischen Porphyrie
Greek	Κυκλοπυρόξη	Θεραπεία της συγγενούς ερυθροποιητικής πορφυρίας
Hungarian	Ciklopirox	Kongenitális erythropoietikus porfíria kezelése
Italian	Ciclopirox	Terapia della porfiria eritropoietica congenita
Latvian	Ciklopirokss	Iedzimtas eritropoētiskas porfīrijas ārstēšana
Lithuanian	Ciklopiroksas	Įgimtos eritropoetinės porfirijos gydymas
Maltese	Ciclopirox	Kura tal-porfirja eritropojetika konġenitali
Polish	Cyklopiroks	Leczenie wrodzonej porfirii erythropoetycznej
Portuguese	Ciclopirox	Tratamento da porfíria eritropoiética congénita
Romanian	Ciclopirox	Tratamentul porfiriei eritropoietice congenitale
Slovak	Ciclopirox	Liečba vrodenej erythropoetickej porfýrie
Slovenian	Ciklopiroks	Zdravljenje kongenitalne eritropoetične porfirije
Spanish	Ciclopirox	Tratamiento de la profiria congénita eritropoyética
Swedish	Ciklopirox	Behandling av medfödd eythropoietisk porfyri
Norwegian	SykCiclopiroksx	Behandling av medfødt erythropoietisk porfyri
Icelandic	Cííklópríox	Til meðferðar við meðfæddri erytrópoietic porfýríu

¹ At the time of designation