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Public summary of opinion on orphan designation

Sirolimus for the treatment of pachyonychia congenita

On 17 July 2017, orphan designation (EU/3/17/1896) was granted by the European Commission to Raremoon Consulting Ltd, United Kingdom, for sirolimus (also known as PTX-001) for the treatment of pachyonychia congenita.

What is pachyonychia congenita?

Pachyonychia congenita is an inherited skin condition in which the patient develops painful blisters and callouses on the palms and soles of the feet, thickened finger and toenails, and ulcers on the inside of the mouth. The disease is caused by mutations (defects) in the genes for keratin (the tough fibrous protein found in the skin and nails), which result in the production of abnormal keratin.

Pachyonychia congenita is a long-term debilitating disease because the painful blisters and callouses make walking difficult or impossible.

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

At the time of designation, pachyonychia congenita affected less than 0.01 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 500 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 the application for orphan designation, no satisfactory methods were authorised in the EU for treating pachyonychia congenita. Treatment focused on the relief of symptoms, personal hygiene to help blisters heal, to avoid infections and to protect the skin from damage, and the treatment of any infections that may occur in the affected skin.

*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).

30 Churchill Place ● Canary Wharf ● London E14 5EU ● United Kingdom
Telephone +44 (0)20 3660 6000 **Facsimile** +44 (0)20 3660 5555
Send a question via our website www.ema.europa.eu/contact

How is this medicine expected to work?

The medicine is a cream to apply to the skin which contains the active substance sirolimus. Sirolimus attaches to and blocks a protein called 'mammalian target of rapamycin' (mTOR). Since mTOR is involved in the control of keratin production, sirolimus is expected to reduce the amount of abnormal keratin produced thereby preventing the formation of blisters and callouses.

Sirolimus oral solution and tablets have been used to prevent kidney transplant rejection since 2001.

What is the stage of development of this medicine?

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

At the time of submission of the application for orphan designation, a clinical trial with the medicine in patients with pachyonychia congenita had been completed.

At the time of submission, sirolimus was not authorised anywhere in the EU for pachyonychia congenita. Orphan designation of sirolimus had been granted in the US for pachyonychia congenita.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 15 June 2017 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

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 languages[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Sirolimus	Treatment of pachyonychia congenita
Bulgarian	Сиролимус	Лечение на вродена пахионхия
Croatian	Sirolimus	Liječenje prirođene pahionihije
Czech	Sirolimus	Léčba kongenitální pachyonychie
Danish	Sirolimus	Behandling af pachyonychia congenita
Dutch	Sirolimus	Behandeling van pachyonychia congenita
Estonian	Siroliimus	Kaasasündinud küünepaksenemuse ravi
Finnish	Sirolimuusi	Synnynnäisen pakyonykian hoito
French	Sirolimus	Traitement de la pachyonychie congénitale
German	Sirolimus	Behandlung von Pachyonychia congenita
Greek	Σιρόλιμους	Θεραπεία της συγγενούς παχυονυχίας
Hungarian	Szirolimusz	Veleszületett pachyonychia kezelése
Italian	Sirolimus	Trattamento della Pachionichia congenital
Latvian	Sirolimus	Iedzimtas pahionīhijas ārstēšana
Lithuanian	Sirolimuzas	Įgimtos pachionichijos gydymas
Maltese	Sirolimus	Kura tal-pakionikja konġenitali
Polish	Syrolimus	Leczenie wrodzonego zgrubienia paznokci
Portuguese	Sirolimus	Tratamento de paquioníquia congénita
Romanian	Sirolimus	Tratamentul pahionichiei congenitale
Slovak	Sirolimus	Liečba pachyonychia congenital
Slovenian	Sirolimus	Zdravljenje kongenitalne pahionihije ionychia congenital
Spanish	Sirolimus	Tratamiento de la paquioniquia congénita
Swedish	Sirolimus	Behandling av pachyonychia congenita
Norwegian	Sirolimus	Behandling av pachyonychia congenita
Icelandic	Sírólímus	Meðferð pachyonychia congenita

[1] At the time of designation