

11 July 2014  
EMA/COMP/317253/2014  
Committee for Orphan Medicinal Products

## Public summary of opinion on orphan designation

### Afamelanotide for the treatment of familial benign chronic pemphigus (Hailey-Hailey disease)

On 4 July 2014, orphan designation (EU/3/14/1285) was granted by the European Commission to Clinuvel UK Limited, United Kingdom, for afamelanotide for the treatment of familial benign chronic pemphigus.

#### What is familial benign chronic pemphigus?

Familial benign chronic pemphigus, also known as Hailey-Hailey disease, is an inherited disease marked by painful sores and blisters in areas of the skin exposed to friction and also, rarely, in other surfaces such as the lining inside the mouth and throat.

The disease is inherited as a dominant trait (which means that it can be passed on to a child from only one parent) and is caused by a mutation (defect) in a gene responsible for a protein that helps glue together the cells of the epidermis (the outer skin). This causes skin cells to separate from each other, causing blisters that turn into sores. Symptoms of the disease normally first appear in the third and fourth decades of life.

Familial benign chronic pemphigus is a long-term debilitating disease because of the recurring painful blisters that can become infected and reduce the patient's quality of life.

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

At the time of designation, familial benign chronic pemphigus affected not more than 0.2 in 10,000 people in the European Union (EU). This was equivalent to a total of not more than 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).

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\*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 511,100,000 (Eurostat 2014).

## What treatments are available?

At the time of the application for orphan designation, no satisfactory methods of treatment were authorised in the EU for familial benign chronic pemphigus. Patients' symptoms were managed with medicines such as corticosteroids applied to the sores and antibiotics for infection.

## How is this medicine expected to work?

Afamelanotide is a copy of the natural hormone  $\alpha$ -melanocyte stimulating hormone ( $\alpha$ -MSH), which stimulates production of melanin, the pigment that gives the skin its colour. Afamelanotide has been designed to last longer in the body than normal  $\alpha$ -MSH. The way it acts on the disease is not clearly understood but it is known to have effects on inflammation and antioxidant properties which may assist the healthy development of the protective outer layers of skin, thus preventing the formation of skin lesions observed in Hailey-Hailey disease.

## What is the stage of development of this medicine?

The effects of afamelanotide have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with afamelanotide in patients with familial benign chronic pemphigus were ongoing.

At the time of submission, afamelanotide was not authorised anywhere in the EU for familial benign chronic pemphigus 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 14 May 2014 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:

Clinuvel UK Limited  
C/o- Reed Smith  
Broadgate Tower, Third Floor  
20 Primrose Street  
London EC2A 2RS  
United Kingdom  
Tel. +44 203 116 3000  
Fax +44 203 116 3999  
E-mail: [mail@clinuvel.com](mailto:mail@clinuvel.com)

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<sup>1</sup>, Norwegian and Icelandic

Language	Active ingredient	Indication
English	Afamelanotide	Treatment of familial benign chronic pemphigus (Hailey-Hailey disease)
Bulgarian	Афамеланотид	Лечение на фамилен доброкачествен хроничен пемфигус (болест на Хейли-Хейли)
Croatian	Afamelanotid	Liječenje familijarnog benignog kroničnog pemfigusa (bolest Hailey-Hailey)
Czech	Afamelanotid	Léčba benigního chronického familiárního pemfigu (Hailey-Hailey)
Danish	Afamelanotid	Behandling af godartet kroniske familiær pemphigus (Hailey-Hailey sygdom)
Dutch	Afamelanotide	Behandeling van goedaardige chronische familiale pemphigus (ziekte van Hailey-Hailey)
Estonian	Afamelanotiid	Perekondliku healoomulise kroonilise pemphiguse ravi (Hailey-Hailey haiguse)
Finnish	Afamelanotidi	Familiaalisen hyvänlaatuisen kroonisen pemfiguksen hoito (Hailey-Hailey tauti)
French	Afamélanotide	Traitement du Pemphigus bénin familial chronique (maladie de Hailey-Hailey)
German	Afamelanotid	Behandlung von familiärem benignem chronischen Pemphigus (Hailey-Hailey-Krankheit)
Greek	Αφαμελανοτιδη	Θεραπεία της οικογενούς καλοήθους χρόνιας πέμφιγας (νόσος Hailey-Hailey)
Hungarian	Afamelanotid	Krónikus familiáris benignus pemphigus kezelése (Hailey-Hailey betegség)
Italian	Afamelanotide	Terapia del pemfigo cronico familiare benigno (malattia di Hailey-Hailey)
Latvian	Afamelanotīds	Labdabīga hroniska ģimenes <i>pemphigus</i> (Hailey-Hailey slimības) ārstēšana
Lithuanian	Afamelanotidas	Šeiminės gerybinės lėtinės pūslinės gydymas (Hailey-Hailey ligos)
Maltese	Afamelanotide	Kura ta' pemfigus kroniku beninn li jintiret (marda ta' Hailey-Hailey)
Polish	Afamelanotyd	Leczenie rodzinnej łagodnej przewlekłej pęcherzycy (choroby Haileya-Haileya)
Portuguese	Afamelanotide	Tratamento do pênfigo crónico familiar benigno (doença de Hailey-Hailey)
Romanian	Afamelanotidă	Tratamentul pemfigusului cronic familial benign (boala Hailey-Hailey)
Slovak	Afamelanotid	Liečba familiárneho benígneho chronického pemfigu (Haileyova-Haileyova choroba)

<sup>1</sup> At the time of designation

Language	Active ingredient	Indication
Slovenian	Afamelanotid	Zdravljenje družinskega benignega kroničnega pemfigusa (Hailey-Haileyova bolezen)
Spanish	Afamelanotida	Tratamiento del pénfigo crónico benigno familiar (enfermedad de Hailey-Hailey)
Swedish	Afamelanotid	Behandling av familjär benign kronisk pemfigus (Hailey-Halleys sjukdom)
Norwegian	Afamelanotid	Behandling av familiær godartet kronisk pemfigus (Hailey-Hailey sykdom)
Icelandic	Afamelanótíð	Meðferð á ættgengum góðkynja langvinnum pemphigus (Hailey-Hailey sjúkdómi)