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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

[Nle4, D-Phe7]-alpha-melanocyte stimulating hormone for the treatment of erythropoietic protoporphyria

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Rev.1: sponsor's change of address	20 August 2009
Rev.2: information about Marketing Authorisation	3 March 2015
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 8 May 2008, orphan designation (EU/3/08/541) was granted by the European Commission to Clinuvel UK Limited, United Kingdom, for [Nle4, D-Phe7]-alfa-melanocyte stimulating hormone for the treatment of erythropoietic protoporphyria.

What is erythropoietic protoporphyria?

Porphyrias are a group of disorders of certain enzymes (proteins that speed up the conversion of certain substances into other substances) responsible for the chemical steps of heme production. Heme is normally present in the body and it is the basic component of haemoglobin, the molecule that carries oxygen in the blood. It is mainly found in the blood, bone marrow, and liver. The signs and symptoms of the disorder vary depending on which chemical step of the heme production that is affected. The porphyrias can be classified according to different criteria, such as the main location of the abnormal enzyme, the leading symptom, or the characteristics of its clinical presentation.

Erythropoietic protoporphyria is characterised by the involvement of blood forming organs, skin and the liver. In erythropoietic protoporphyria there is a lack of one enzyme important for the heme production, and therefore the the red blood cells accumulate substances that they normally do not accumulate. These substances also accumulate in the liver and in the skin. When skin is exposed to sunlight the accumulated substances react with light and cause symptoms such as burning sensation, itching, red skin and ulceration.



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

At the time of designation, erythropoietic protoporphyria affected less than 0.2 in 10,000 people in the European Union (EU). This was equivalent to a total of 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).

What treatments are available?

No satisfactory methods exist that were authorised at the time of application.

Avoidance of sunlight or strong light altogether is essential to manage the disease.

How is this medicine expected to work?

The product, [Nle⁴, D-Phe⁷] alpha-melanocyte stimulating hormone is a synthetic product with similar properties to the naturally occurring alpha-melanocyte stimulating hormone or melanotropin. This hormone is a naturally occurring hormone that stimulates the production of eumelanin in skin cells. Eumelanin, the natural black-brown pigment in the skin, has the capacity to protect cells from irradiation (sun exposure) and its damaging effects on the cellular genetic material. It is thought that the product stimulates eumelanin production in the skin cells and stops cellular damage by inhibiting (blocking) sun light's interaction with the accumulated substances that are found in the skin of patients with erythropoietic protoporphyria.

What is the stage of development of this medicine?

At the time of submission of the application for orphan designation, clinical trials in patients with erythropoietic protoporphyria were ongoing.

[Nle⁴, D-Phe⁷] alpha-melanocyte stimulating hormone was not authorised anywhere worldwide for erythropoietic protoporphyria or designated as orphan medicinal product elsewhere for this condition, at the time of submission.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 4 March 2008 recommending the granting of this designation.

Update: [Nle⁴, D-Phe⁷]-alfa-melanocyte stimulating hormone, afamelanotide (Scenesse) has been authorised in the EU since 22 December 2014 for prevention of phototoxicity in adult patients with erythropoietic protoporphyria (EPP).

More information on Scenesse can be found in the European public assessment report (EPAR) on the Agency's website: ema.europa.eu/Find_medicine/Human_medicines/European_Public_Assessment_Reports

*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 502,800,000 (Eurostat 2008).

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

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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	[Nle ⁴ , D-Phe ⁷]-alpha-melanocyte stimulating hormone	Treatment of erythropoietic protoporphyria
Bulgarian	[Nle ⁴ , D-Phe ⁷]- алфа-меланоцитостимулиращ хормон	Лечение на еритропоетична протопорфирия
Czech	[Nle ⁴ , D-Phe ⁷]-alfa melanocyty stimuluující hormon	Léčba erythropoetické protoporfyrie
Danish	[Nle ⁴ , D-Phe ⁷]-alfa-melanocytstimulerende hormon	Behandling af erythropoietisk protoporfyri
Dutch	[Nle ⁴ , D-Phe ⁷]-alfa-melanocytstimulerend hormoon	Behandeling van erythropeëtische protoporphyrie
Estonian	[Nle ⁴ , D-Phe ⁷]-alfa melanotsüüte stimuleeriv hormoon	Erütropoetilise protoporfüüria ravi
Finnish	[Nle ⁴ , D-Phe ⁷]-alfamelanosyyttejä stimuloiva hormoni	Erythropeettisen protoporfyrin hoito
French	[Nle ⁴ , D-Phe ⁷]-Mélanostimuline alpha	Traitement de la protoporphyrie érythropeïétique
German	[Nle ⁴ , D-Phe ⁷]-alpha-melanozytenstimulierendes Hormon	Behandlung der erythropeitischen Protoporphyrie
Greek	[Nle ⁴ , D-Phe ⁷]άλφα-ορμόνη διέγερσης των μελανοκυττάρων	Θεραπεία της συγγενούς ερυθροποιητικής πορφυρίας
Hungarian	[Nle ⁴ , D-Phe ⁷]-alfa-melanocyt-stimuláló hormon	Erithropoietikus protoporfíria kezelésé
Italian	[Nle ⁴ , D-Phe ⁷]-alfa-melanotropina	Terapia della protoporfiria eritropoietica
Latvian	[Nle ⁴ , D-Phe ⁷]-alfa-melanocītus stimulējošais hormons	Eritropoētiskas protoporfirijas ārstēšana
Lithuanian	[Nle ⁴ , D-Phe ⁷]-alfa-melanocitus stimuliuojantis hormonas	Eritropoetinės protoporfirijos gydymas
Maltese	[Nle ⁴ , D-Phe ⁷]-Ormon li jistimula l-alfa-melanocite	Kura tal-protoporfirja eritropojetika
Polish	[Nle ⁴ , D-Phe ⁷] hormon stymulujący melanocyty alfa	Leczenie wrodzonej protoporfirii erythropoetycznej
Portuguese	[Nle ⁴ , D-Phe ⁷]-Hormona estimuladora dos melanocitos alfa-	Tratamento da protoporfiria eritropoietica
Romanian	[Nle ⁴ , D-Phe ⁷]-hormon melanocitostimulator alfa	Tratamentul protoporfiriei eritropoietice
Slovak	[Nle ⁴ , D-Phe ⁷]-alfa melanocyty stimulujući hormón	Liečba erythropeetickej protoporfýrie
Slovenian	[Nle ⁴ , D-Phe ⁷]-alfa-melanocite stimulirajoči hormon	Zdravljenje eritropoetične porfirije
Spanish	[Nle ⁴ , D-Phe ⁷]- hormona estimulante de los melanocitos alfa	Tratamiento de la protoporfiria eritropoyética

¹ At the time of designation

Language	Active Ingredient	Indication
Swedish	[Nle ⁴ , D-Phe ⁷]-alfa-melanocytstimulerande hormon	Behandling av erythropoietisk protoporfyri
Norwegian	[Nle ⁴ , D-Phe ⁷]-alfa-melanocytstimulerende hormon	Behandling av erythropoietisk protoporfyri
Icelandic	[Nle ⁴ , D-Phe ⁷]-alfa-sortufrumnahvatahormón	Meðferð á erythropoietic prótóporfýríu