2 August 2019
EMADOC-628903358-934

Public summary of opinion on orphan designation

Diacerein for the treatment of epidermolysis bullosa

On 29 May 2019, orphan designation (EU/3/19/2161) was granted by the European Commission to Worphmed World Orphan Medicines Limited, United Kingdom, for diacerein for the treatment of epidermolysis bullosa.

What is epidermolysis bullosa?

Epidermolysis bullosa is a group of inherited diseases in which the skin is very fragile and forms severe blisters after even minor friction (rubbing) or injury. In most cases, symptoms of epidermolysis bullosa appear from birth, but for some forms, symptoms may not occur until adulthood. The diseases are caused by mutations (changes) in the genes responsible for the production of certain proteins that make the skin strong and elastic, such as collagen or keratins.

Epidermolysis bullosa is a long-term debilitating and life-threatening condition because the severe blistering and associated scarring and deformities result in poor quality of life and may reduce life expectancy.

What is the estimated number of patients affected by epidermolysis bullosa?

At the time of designation, epidermolysis bullosa affected 0.90 in 10,000 people in the European Union (EU). This was equivalent to a total of around 47,000 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, no satisfactory methods were authorised in the EU to treat epidermolysis bullosa. Good personal hygiene and skincare were recommended to help blisters heal, to avoid infections and to protect the skin from damage. Painkillers were also used. Surgery was sometimes necessary for complications such as deformed hands.

[*]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 518,400,000 (Eurostat 2019).

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How is this medicine expected to work?

Diacerein (taken by mouth) is authorised in some EU countries to treat symptoms of osteoarthritis (swelling and pain in the joints) and other degenerative joint diseases.

Diacerein belongs to a class of substances called anthraquinones. It works by blocking the action of interleukin-1 beta, a protein involved in the inflammation process. Interleukin-1 beta is thought to be over-activated in some patients with epidermolysis bullosa. By blocking its action, diacerein is expected to reduce the inflammation and the number of blisters in patients with the disease.

This medicine is expected to be available as a cream.

What is the stage of development of this medicine?

The effects of diacerein have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with this medicine in patients with epidermolysis bullosa were ongoing.

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

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 17 April 2019, 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 languages[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Diacerein	Treatment of epidermolysis bullosa
Bulgarian	Диацереин	Лечение на булозна епидермолиза
Croatian	Diacerein	Liječenje bulozne epidermolize
Czech	Diacerein	Léčba bulózní epidermolýzy
Danish	Diacerein	Behandling af epidermolysis bullosa
Dutch	Diacerein	Behandeling van epidermolysis bullosa
Estonian	Diatsereiin	Bulloosse epidermolüüsi ravi
Finnish	Diasereiini	Epidermolysis bullosan hoito
French	Diacerein	Traitement de l'épidermolyse bulleuse
German	Diacerein	Behandlung der Epidermolysis bullosa
Greek	Διασερεΐνη	Θεραπεία της πομφολυγώδους επιδερμόλυσης
Hungarian	Diacerein	Epidermolysis bullosa kezelése
Italian	Diacereina	Trattamento della epidermolisi bollosa
Latvian	Diacereīns	Bulozās epidermolīzes ārstēšanai
Lithuanian	Diacereinas	Pūslinės epidermolizės gydymas
Maltese	Diacerein	Kura tal-epidermolisi bullosa
Polish	Diacereina	Pęcherzowe oddzielanie się naskórka
Portuguese	Diacereina	Tratamento da epidermólise bulhosa
Romanian	Diacerein	Tratamentul epidermolizei buloase
Slovak	Diacereín	Liečba epidermolysis bullosa
Slovenian	Diacerein	Zdravljenje bulozne epidrmolize
Spanish	Diacereina	Tratamiento de la epidermolisis bullosa
Swedish	Diacerein	Behandling av epidermolysis bullosa
Norwegian	Diacerein	Behandling av epidermolysis bullosa
Icelandic	Díacerein	Meðferð á epidermolysis bullosa

[1] At the time of designation