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Committee for Orphan Medicinal Products

# Public summary of opinion on orphan designation

## Recombinant human alpha 1 chain homotrimer of type VII collagen for the treatment of epidermolysis bullosa

On 4 June 2014, orphan designation (EU/03/14/1272) was granted by the European Commission to Shire Pharmaceuticals (Ireland) Limited, Ireland, for recombinant human alpha 1 chain homotrimer of type VII collagen for the treatment of epidermolysis bullosa.

### What is epidermolysis bullosa?

Epidermolysis bullosa describes a group of diseases of the skin, in which the skin is very fragile and forms severe blisters upon minor mechanical friction or injury. The condition is usually present from birth, although some forms occur in adults. The diseases are caused by abnormalities 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 also reduce life expectancy.

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

At the time of designation, epidermolysis bullosa affected not more than 0.8 in 10,000 people in the European Union (EU). This was equivalent to a total of not more than 41,000 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. A high standard of personal hygiene and wound care were recommended to help blisters heal, to avoid infections and to protect the skin from damage. Painkillers and antibiotics were also used to

*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).

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manage pain and infections. Surgery was sometimes necessary if there were complications such as deformed hands or the development of skin cancer.

## How is this medicine expected to work?

In patients with a type of epidermolysis bullosa known as dystrophic epidermolysis bullosa, the blistering is caused by a faulty gene for a protein known as collagen VII that is needed to hold different skin layers together.

This medicine contains functional collagen VII that is lacking in patients with dystrophic epidermolysis bullosa. The medicine is to be given by injection and it is expected that the collagen will be incorporated into the skin, where they will help to hold the skin layers together and prevent the formation of blisters.

The collagen VII in the medicine is made by a method known as 'recombinant DNA technology': it is made by cells into which a gene (DNA) has been introduced that makes them able to produce the protein.

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

At the time of submission of the application for orphan designation, the evaluation of the effects of the medicine in experimental models was ongoing.

At the time of submission, no clinical trials with the medicine in patients with epidermolysis bullosa had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for epidermolysis bullosa. Orphan designation has been granted in the United States for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 9 April 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:

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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 languages[1], Norwegian and Icelandic

| Language | Active ingredient | Indication |
|---|---|---|
| English | Recombinant human alpha 1 chain homotrimer of type VII collagen | Treatment of epidermolysis bullosa |
| Bulgarian | рекомбинантен човешки алфа 1 верижен хомотример на колаген тип VII | Лечение на булозна епидермолиза |
| Czech | Rekombinantní lidský homotrimer řetězce alfa-1 kolagenu typu VII | Léčba bulózní epidermolýzy |
| Croatian | Rekombinantni ljudski homotrimer alfa-1 lanca kolagena tipa VII | Liječenje bulozne epidermolize |
| Danish | Rekombinant human alfa-1-kæde homotrimer af type VII-kollagen | Behandling af epidermolysis bullosa |
| Dutch | Recombinant humaan alfa-1-keten homotrimeer van collageen type VII | Behandeling van epidermolysis bullosa |
| Estonian | Rekombinantne inimese VII tüüpi kollageeni alfa-1 ahela homotrimeer | Bulloosse epidermolüüsi ravi |
| Finnish | Alfa 1-ketjun, tyypin VII kollageenia oleva, rekombinantti humaani homotrimeeri | Epidermolysis bullosan hoito |
| French | Homotrimère humain recombinant composé de chaînes alpha-1 de collagène de type VII | Traitement de l'épidermolyse bulleuse |
| German | Rekombinantes humanes Alpha-1-Ketten-Homotrimer des Typ-VII-Kollagens | Behandlung der Epidermolysis bullosa |
| Greek | ομοτριμερές αλυσίδας α-1 ανασυνδυασμένου ανθρώπινου κολλαγόνου τύπου VII | Θεραπεία της πομφολυγώδους επιδερμόλυσης |
| Hungarian | VII-es típusú kollagén rekombináns humán alfa-1 láncának homotrimerje | Epidermolysis bullosa kezelése |
| Italian | Omotrimero umano ricombinante di catene alfa 1 di collagene tipo VII | Trattamento della epidermolisi bollosa |
| Latvian | Rekombinants VII tipa kolagēna cilvēka alfa 1 ķēdes homotrimērs | Bulozās epidermolīzes ārstēšanai |
| Lithuanian | Rekombinantinis žmogaus alfa 1 grandinės VII tipo kolageno homotrimeras | Pūslinės epidermolizės gydymas |
| Maltese | Omotrimer uman rikombinanti b'katina alfa 1 ta' kollaġni tat-tip VII | Kura tal-epidermolisi bullosa |
| Polish | Rekombinowany ludzki homotrimer zbudowany z łańcucha alfa1 kolagenu typu VII | Pęcherzowe oddzielanie się naskórka |
| Portuguese | Colagénio humano recombinante de tipo VII de homotrímero de cadeia alfa-1 | Tratamento da epidermólise bulhosa |
| Romanian | Homotrimer din lanţuri alfa 1 de colagen de tip VII uman recombinant | Tratamentul epidermolizei buloase |

[1] At the time of designation

| Language | Active ingredient | Indication |
|---|---|---|
| Slovak | Rekombinantný ľudský homotrimér reťazca alfa-1 kolagénu typu VII | Liečba epidermolysis bullosa |
| Slovenian | Rekombinantni humani homotrimer verig alfa-1 kolagena tipa VII | Zdravljenje bulozne epidrmolize |
| Spanish | Homotrímero de cadenas alfa 1 del colágeno tipo VII humano recombinante | Tratamiento de la epidermolisis bullosa |
| Swedish | Rekombinant human alfa-1-kedjehomotrimer av typ VII-kollagen | Behandling av epidermolysis bullosa |
| Norwegian | Rekombinant human alfa 1 kjedehomotrimer av type VII kollagen | Behandling av epidermolysis bullosa |
| Icelandic | Raðbrigða, manna alfa-1-keðju þrennd (homotrimer) af tegund VII-kollageni | Meðferð á epidermolysis bullosa |