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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Skin equivalent graft genetically corrected with a COL7A1-encoding SIN retroviral vector for the treatment of dystrophic epidermolysis bullosa

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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 30 April 2009, orphan designation (EU/3/09/630) was granted by the European Commission to Prof.Alain Hovnanian, France, for skin equivalent graft genetically corrected with a COL7A1-encoding SIN retroviral vector for the treatment of dystrophic epidermolysis bullosa.

What is epidermolysis bullosa?

Dystrophic epidermolysis bullosa is a genetic disease of the skin, in which the outer layer of the skin, the epidermis, separates from the inner layer, the dermis. This makes the skin very fragile and causes severe blistering and scarring. The disease is caused by abnormalities in the COL7A1 gene. This gene is responsible for the production of a protein called 'collagen VII', which normally links the epidermis and the dermis together.

Dystrophic epidermolysis bullosa is a long-term debilitating disease and may be life threatening mainly because of the severe blistering, which results in poor quality of life and shortened life expectancy.

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

At the time of designation, epidermolysis bullosa affected less than 0.5 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 25,000 people*, and is below the

*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 504,800,000 (Eurostat 2009).

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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 for the treatment of dystrophic epidermolysis bullosa. Patients were advised to maintain a high standard of personal hygiene and skincare to help blisters heal, avoid infection and protect the skin from damage. Painkillers were also used, and surgery in case of complications.

How is this medicine expected to work?

Skin equivalent graft genetically corrected with a COL7A1-encoding SIN retroviral vector is made up of skin cells taken from the patient, which have been grown in the laboratory to produce a layer of skin for grafting. These cells are modified by a virus so that they have normal copies of the COL7A1 gene inserted in their genes. When the modified skin cells are grafted onto the patient's skin, they are expected to start producing collagen VII, helping the dermis and epidermis to stick together and preventing blister formation. The type of virus used in this medicine ('SIN retrovirus') has been modified so that it does not cause disease in humans.

What is the stage of development of this medicine?

The evaluation of the effects of this medicine in experimental models is ongoing.

At the time of submission of the application for orphan designation, no clinical trials in patients with dystrophic epidermolysis bullosa had been started.

At the time of submission, this medicine was not authorised anywhere in the EU for dystrophic epidermolysis bullosa or designated as 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 4 March 2009 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

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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	Skin equivalent graft genetically corrected with a COL7A1-encoding SIN retroviral vector	Treatment of dystrophic epidermolysis bullosa
Bulgarian	Еквивалентна на кожа присадка, генетично коригирана със COL7A-кодиращ SIN ретровирусен вектор,	Лечение на дистрофична булозна епидермиолиза
Czech	Štěp ekvivalentní kůži geneticky modifikovaný pomocí COL7A1-kódovaného SIN retrovirovým vektorem	Léčba dystrofické bulózní epidermolýzy
Danish	Hudækvivalent graft genetisk korrigeret med en COL7A1-kodende SIN retroviral vektor	Behandling af dystrofisk epidermolysis bullosa
Dutch	Huidequivalent transplantaat, genetisch gecorrigeerd met een COL7A1-coderend SIN retrovirale vector	Behandeling van dystrophisch epidermolysis bullosa
Estonian	Nahaga ekvivalentset siirik, mida on geneetiliselt korrigeeritud COL7A1-kodeeriva SIN-retroviirusvektoriga	Bulloosse düstroofilise epidermolüüsi ravi
Finnish	COL7A1-geeniä koodaavalla SIN-retroviruksella geneettisesti korjattu ihon siirrännäisvastine	Dystrofisen epidermolysis bullosan hoito
French	Equivalent de peau génétiquement corrigée par un vecteur rétroviral SIN exprimant COL7A1	Traitement de l'épidermolyse bulleuse dystrophique
German	Mit einem COL7A1-kodierten SIN Retrovirus-Vektor genetisch korrigiertes Hautersatztransplantat	Behandlung der Dystrophischen Epidermolysis bullosa
Greek	Μόσχευμα ισοδύναμο με δέρμα γενετικά διορθωμένο με ρετροϊκό φορέα SIN κωδικοποίησης COL7A1	Θεραπεία της δυστροφικής πομφολυγώδους επιδερμόλυσης
Hungarian	Bőrrel azonos, genetikailag COL7A1-t kódoló SIN retrovírusos vektorral korrigált graft	Dystrophiás epidermolysis bullosa kezelése
Italian	Innesto cutaneo equivalente geneticamente modificato con un vettore retrovirale SIN codificante per COL7A1	Trattamento della epidermolisi bollosa distrofica
Latvian	Ādai līdzvērtīgs transplantāts, kas ģenētiski koriģēts ar COL7A1-kodējošu SIN retrovīrusu vektoru	Distrofiskās bulozās epidermolīzes ārstēšanai

[1] At the time of designation

Language	Active ingredient	Indication
Lithuanian	Odai ekvivalentiškas transplantatas, genetiškai koreguotas naudojant COL7A1- koduotą SIN retroviruso vektorių	Distrofinės pūslinės epidermolizės gydymas
Maltese	Trapjant ekwivalenti għall-ġilda kkoreġut ġenetikament b'vettur retrovirali SIN ikkodifikat minn COL7A1	Kura ta' l-epidermolisi bullosa distrofika
Polish	Opowiednik skóry genetycznie skorygowanej z COL7A1 kodującym SIN wektorem retrowirusowym używanym do przeszczepów	Leczenie postaci dystroficznej pęcherzowego oddzielania się naskórka
Portuguese	Enxerto de equivalente cutâneo, geneticamente corrigido com um vector retroviral SIN portador do gene COL7A1	Tratamento da epidermólise bulhosa distrófica
Romanian	Grefă echivalentă cu grefa de piele corectată genetic cu vectorul retroviral de tip SIN, care exprimă gena COL7A1	Tratament pentru epidermoliza buloasă distrofică
Slovak	Štep ekvivalentný koži, geneticky upravený COL7A1 kódujúcim SIN retrovírusový vektor	Liečba dystrofickej bulóznej epidermolýzy
Slovenian	Presadek ekvivalenta kože, genetsko popravljen s SIN retrovirusnim vektorjem, ki kodira COLA7A1	Zdravljenje distrofične bulozne epidrmolize
Spanish	Injerto de equivalente cutáneo genéticamente corregido con un vector retrovírico autoinactivable portador del gen COL7A1	Tratamiento de la epidermolisis bullosa distrófica
Swedish	Hudekvivalent transplantat genetiskt korrigerat med COL7A1-kodande retroviral vektor	Behandling av dystrofisk epidermolysis bullosa
Norwegian	Hud-ekvivalent graft genetisk korrigert med en COL7A1-kodet SIN retroviral vektor	Behandling av dystrofisk epidermolysis bullosa
Icelandic	Ígræðsla húðarjafngildi erfðafræðilega lagfært með COL7A1-kóðuðu SIN retróveiru ferju	Meðferð á vanþroska epidermolysis bullosa