EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant human dyskerin for the treatment of dyskeratosis congenita

On 8 November 2012, orphan designation (EU/3/12/1070) was granted by the European Commission to Advanced Medical Projects, Spain, for recombinant human dyskerin for the treatment of dyskeratosis congenita.

What is dyskeratosis congenita?

Dyskeratosis congenita is an inherited genetic disease that causes a reduction in the activity of an enzyme called telomerase, which is important for cell division. A lack of telomerase activity stops the cells from replicating, thereby promoting the ageing of tissues. The condition typically causes signs and symptoms in children that resemble premature aging, such as abnormal skin pigmentation on the face and chest, abnormal shapes to nails and white patches in the mouth.

Dyskeratosis congenita is a long-term debilitating and life-threatening disease because it is associated with serious health complications including bone marrow failure and the development of various types of cancer.

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

At the time of designation, dyskeratosis congenita affected approximately 0.01 in 10,000 people in the European Union (EU)[*]. This is equivalent to a total of around 500 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 dyskeratosis congenita.

[*]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. This represents a population of 506,300,000 (Eurostat 2011).

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How is this medicine expected to work?

In patients with dyskeratosis congenita there is a defect in one of the parts of telomerase, called dyskerin. Recombinant human dyskerin is expected to replace the defective dyskerin, thereby allowing the telomerase to function properly in cells.

The medicine is made by a method known as 'recombinant DNA technology': it is made by bacteria that have received a gene (DNA) that makes them able to produce dyskerin. It has also been modified by a process called 'pegylation'. This means that a chemical called 'polyethylene glycol' has been attached to dyskerin. This is expected to decrease the rate at which dyskerin is removed from the body, allowing the medicine to be given less often.

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 recombinant human dyskerin in experimental models was ongoing.

At the time of submission of the application for orphan designation, no clinical trials with recombinant human dyskerin in patients with dyskeratosis congenita had been started.

At the time of submission, recombinant human dyskerin was not authorised anywhere in the EU for dyskeratosis congenita 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 5 October 2012 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:

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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 dyskerin	Treatment of dyskeratosis congenita
Bulgarian	Рекомбинантен човешки дискерин	Лечение на вродена дискератоза
Czech	Rekombinantní humánní dyskerin	Léčba kongenitální dyskeratózy
Danish	Rekombinant humant dyskerin	Behandling af dyskeratosis congenita
Dutch	Recombinant humaan dyskerine	Behandeling van congenitale dyskeratosis
Estonian	Rekombinantne inimese düskeriin	Kaasasündinud düskeratoosi ravi
Finnish	Rekombinanttitekniikalla tehty ihmisen dyskeriini	Synnynnäisen dyskeratoosin hoito
French	Recombinante dyskérine humaine	Traitment de la dyskératosis congénitale
German	Rekombinantes humanes Dyskerin	Behandlung der angeborenen Dyskeratose
Greek	Ανασυνδυασμένη ανθρώπινη δυσκερίνη	Θεραπεία της συγγενούς δυσκεράτωσης.
Hungarian	Rekombináns humán dyskerin	Veleszületett dyskeratosis kezelése
Italian	Discherina umana ricombinante	Trattamento della discheratosi congenita
Latvian	Rekombinēts cilvēka diskerīns	Iedzimtas diskeratozes ārstēsana
Lithuanian	Rekombinantinis žmogaus diskerinas	Įgimtos diskeratozės gydymas
Maltese	Diskerin uman rikombinanti	Kura tad-diskeratosi konġenitali
Polish	Rekombinowana dyskeryna ludzka	Leczenie dyskeratozy wrodzonej
Portuguese	Disquerina humana recombinante	Tratamento da disqueratose congénita
Romanian	Diskerina umana recombinata	Tratamentul diskeratozei congenitale
Slovak	Rekombinantný ľudský dyskerín	Liečba vrodenej dyskeratózy
Slovenian	rekombinantni humani diskerin	Zdravljenje kongenitalne diskeratoze
Spanish	Disquerina humana recombinante	Tratamiento de la diskeratosis congenita
Swedish	Rekombinant humant dyskerin	Behandling av dyskeratosis congenita
Norwegian	Rekombinant humant dyskerin	Behandling av dyskeratosis congenita
Icelandic	Raðbrigða manna dýskerín	Meðferð meðfætts dyskeratósis

[1] At the time of designation