European Medicines Agency
Pre-authorisation Evaluation of Medicines for Human Use

Document Date: London, 14 May 2009
Doc.Ref.: EMEA/COMP/257219/2008

Committee for Orphan Medicinal Products

Public summary of positive opinion for orphan designation of retroviral gamma c cDNA containing vector for the treatment of severe combined immunodeficiency (SCID)-Xl disease

On 30 May 2001, orphan designation (EU/3/01/038) was granted by the European Commission to Génopoïétic SAS, France, for retroviral gamma c cDNA containing vector for the treatment of severe combined immunodeficiency (SCID)-Xl disease.

What is severe combined immunodeficiency (SCID)-Xl disease?
The immune system is the body's natural defence system against infections. It is a complex system with various cell types and molecules involved in its activity and regulation. Interleukins are a type of proteins that have many functions in the immune process, including signalling between cells and activation of cells. The majority of patients affected by severe combined immunodeficiency disease (SCID) have an inherited genetic abnormality that affects the production of interleukins. As a result, patients' immune system does not operate normally and patients suffering from SCID are susceptible to serious infections almost as soon as they are born. The defective gene is called gamma c (γc) and it is located on the X chromosome which is why the disease is said to be X-linked (Xl). Women have two X chromosomes and men have one X chromosome along with one Y chromosome. This means that women who carry one defective gene will not the have the disease, unless both genes on both chromosomes are defective. This means that this is a recessive disease. Men however, only have one copy of the gene and will therefore have the disease if that copy is defective. Severe combined immunodeficiency (SCID)-Xl disease is chronically debilitating and life threatening.

What is the estimated number of patients affected by the condition[*]?
At the time of designation severe combined immunodeficiency (SCID)-Xl disease affected approximately 0.003 in 10,000 people in the European Union (EU)*. This is based on the information provided by the sponsor and knowledge of the Committee for Orphan Medicinal Products (COMP). This is below the threshold for orphan designation which is 5 in 10,000. This is equivalent to a total of around 100 people.

What treatments are available?
There are no medicinal products available for the treatment of the condition at the time of submission of application for orphan drug designation. Some patients received bone marrow transplantation, although this is not available to all. Patients were also treated with passive immunisation (antibodies against various pathogens such as bacteria, virus and fungus) and antibiotics (medicines that kill microorganisms).

[*]Disclaimer: For the purpose of the designation, the number of patients affected by the condition is estimated and assessed based on data from the European Union. This represents a population of 377,000,000 (Eurostat 2001).

7 Westferry Circus, Canary Wharf, London, E14 4HB, UK
Tel. (44-20) 74 18 84 00 Fax (44-20) 75 23 70 40
E-mail: orphandrugs@emea.europa.eu http://www.emea.europa.eu

How is this medicine expected to work?
Retroviral gamma c cDNA containing vector is a synthetic virus particle (the vector), modified to contain a normal copy (cDNA – copy DNA) of the defective gamma c gene. Retroviruses are specialised in inserting virus genes into human cells and this function is utilised by this medicinal product. In this way the medicinal product can modify cells from the patients to incorporate a normal gamma c gene and thus make them produce the normal protein.

What is the stage of development of this medicine?
The effects of retroviral gamma c cDNA containing vector were evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials in patients with severe combined immunodeficiency (SCID)-Xl disease were ongoing.

Retroviral gamma c cDNA containing vector was not authorised anywhere worldwide for severe combined immunodeficiency (SCID)-Xl disease, nor designated as orphan medicinal product elsewhere for this condition, at the time of submission.

According to Regulation (EC) No 141/2000 of 16 December 1999, the Committee for Orphan Medicinal Products (COMP) adopted on 10 April 2001 a positive opinion recommending the grant of the above-mentioned 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;
- and either the rarity of the condition (affecting not more than five in 10,000 people in the Community) or the 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 the quality, safety and efficacy is necessary before a product can be granted a marketing authorisation.

For more information:
Sponsor's contact details:
Génopoïétic SAS
1390 rue Centrale-Beynost
01708 Miribel Cedex
France
Telephone: +33 4 72 71 25 36
Telefax: +33 4 78 55 49 50
E-mail: genopoietic@genopoietic.fr

Patients' associations contact points:

International Patient Organisation for Primary Immunodeficiencies
Firside
Main Road, Downderry
Cornwall PL11 3LE
United Kingdom
Telephone: +44 1503 250 668
Telefax: +44 1503 250 668

Immuno-déficience héréditaire : Recherche, Information, Soutien
Association IRIS
BP 72
55102 Verdun Cedex
France
Telephone: +33 3 29 83 48 34
Telefax: +33 3 29 83 48 35

Deutsche Selbsthilfe angeborene Immundefekte e. V.
Hochschatzen 5
83530 Schnaitsee
Germany
Telephone: +49 8074 81 64
Telefax: +49 8074 97 34

Translations of the active ingredient and indication in all EU languages and Norwegian and Icelandic

LANGUAGE	Active Ingredient	Indication
English	Retroviral γc cDNA containing vector	Treatment of Severe Combined Immunodeficiency (SCID)-X1 Disease
Danish	Retroviral vektor indeholdende γc cDNA	Behandling af sygdommen SCID-X1 (Severe Combined Immunodeficiency)
Dutch	Retrovirale vector met γc-keten cDNA tussenvoeging	Behandeling van SCID-X1 (Human Severe Combined Immunodeficiency)
Finnish	Retroviraalisen cDNA:n sisältävä vektorivä	Vakava kombinoitu immuunipuutos (SCID)-X1 -taudin hoito
French	Vecteur rétroviral véhiculant l'ADNc de la chaîne γc	Traitement du Déficit Immunitaire Combiné Sévère (DICS-X1) lié à l'X
German	Retroviraler Vektor mit dem cDNA-Insert der γc-Kette	Behandlung der schweren kombinierten Immunschwäche (SCID)-X1 (Severe Combined Immunodeficiency Disease)
Greek	Φορέας ρετροϊού που εγκλύει το συμπληρωματικό DNA της αλυσίδας γc	Αγωγή για σύνδρομο συνδυασμένης ανοσολογικής ανεπάρκειας
Italian	Vettore retrovirale contenete il cDNA della catena γc	Trattamento della sindrome da immunodeficienza grave combinata (SCID)-X1
Potuguese	Vetor retroviral contendo a inserção do cDNA de cadeia γc	Tratamento da Doença da Imunodeficiência Severa Combinada (SCID)-X1
Spanish	Vector retroviral conteniendo un inserto cDNA de cadena γc	Tratamiento de la inmunodeficiencia severa combinada (SCID)-X1
Swedish	Vektor som innehåller ett Retroviralt γc cDNA-segment	Behandling av svår kombinerad könsbunden immunbrist (SCID)-X1