23 February 2015
EMA/COMP/735793/2014
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Plerixafor for the treatment of WHIM syndrome

On 16 December 2014, orphan designation (EU/3/14/1403) was granted by the European Commission to Groupe d'étude des neutropénies, France, for plerixafor for the treatment of WHIM syndrome.

What is WHIM syndrome?

WHIM syndrome is a hereditary condition in which the immune system (the body's natural defences) does not work properly, making patients more susceptible to viral and bacterial infections.

WHIM stands for warts (skin growths), hypogammaglobulinemia (low level of antibodies), infections and myelokathexis (a disorder causing low levels of white blood cells).

Patients with the condition have warts in the hands and feet caused by viral infections, and are at risk of recurrent bacterial infections due to low levels of neutrophils and lymphocytes (types of white blood cells) and of antibodies produced by the white bloods cells to fight infections.

WHIM syndrome is a long-term debilitating and life-threatening condition because of the recurrent infections which increase the risk of developing cancer.

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

At the time of designation, WHIM syndrome affected approximately 0.002 in 10,000 people in the European Union (EU). This was equivalent to a total of around 100 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 WHIM syndrome. Patients were given treatment to relieve the symptoms of the condition, including granulocyte-colony stimulating factor which stimulates the bone marrow to produce neutrophils.

[*]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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How is this medicine expected to work?

Patients with WHIM syndrome have mutations (defects) in the gene for the CXCR4 receptor, which plays a role in the movement of blood cells into and from the bone marrow (where blood cells are produced). Because of these mutations, the CXCR4 receptor is hyperactive. As a result, blood cells, particularly neutrophils, are retained in the bone marrow, leading to low levels of neutrophils in the blood.

Plerixafor is expected to work by attaching to the CXCR4 receptor and thereby reducing its activity. By reducing the activity of the CXCR4 receptor, plerixafor allows neutrophils to be released from the bone marrow into the blood stream, thereby helping the body to fight infections.

What is the stage of development of this medicine?

The effects of plerixafor have been evaluated in experimental models.

At the time of submission of the application for orphan designation, a clinical trial with plerixafor in patients with WHIM syndrome was planned.

At the time of submission, plerixafor was authorised in the EU as Mozobil as a treatment to mobilise haematopoietic stem cells from the bone marrow into the blood stream, in order to collect them prior to stem cell transplantation.

At the time of submission, plerixafor was not authorised anywhere in the EU for WHIM syndrome 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 13 November 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	Plerixafor	Treatment of WHIM syndrome
Bulgarian	Плериксафор	Лечение на синдрома WHIM
Croatian	Pleriksafor	Liječenje WHIM sindroma
Czech	Plerixafor	Léčba syndromu WHIM
Danish	Plerixafor	Behandling af WHIM syndrom
Dutch	Plerixafor	Behandeling van de WHIM syndroom
Estonian	Pleriksafoor	WHIM sündroomi ravi
Finnish	Pleriksafori	WHIM-oireyhtymän hoito
French	Plérixafor	Traitement du syndrome WHIM
German	Plerixafor	Die Behandlung des WHIM Syndroms
Greek	Πλεριξαφόρη	Θεραπεία του συνδρόμου WHIM
Hungarian	Plerixafor	WHIM szindróma kezelése
Italian	Plerixafor	Trattamento della sindrome WHIM
Latvian	Pleriksafors	WHIM sindroma ārstēšana
Lithuanian	Pleriksaforas	WHIM sindromo gydymas
Maltese	Plerixafor	Kura tas-sindrome WHIM
Polish	Pleryksafor	Leczenie zespołu WHIM
Portuguese	Plerixafor	Tratamento do síndrome de WHIM
Romanian	Plerixafor	Tratamentul sindromului WHIM
Slovak	Plerixafor	Liečba syndrómu WHIM
Slovenian	Pleriksafor	Zdravljenje sindroma WHIM
Spanish	Plerixafor	Tratamiento del síndrome de WHIM
Swedish	Plerixafor	Behandling av WHIM syndromet
Norwegian	Pleriksafor	Behandling av WHIM syndrom
Icelandic	Plerixafór	Meðferð á WHIM heilkenni

[1] At the time of designation