



EUROPEAN MEDICINES AGENCY  
SCIENCE MEDICINES HEALTH

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## Public summary of opinion on orphan designation

### Mavorixafor for the treatment of WHIM syndrome

On 25 July 2019, orphan designation EU/3/19/2183 was granted by the European Commission to Voisin Consulting S.A.R.L., France, for mavorixafor 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 bacterial and viral infections.

WHIM stands for warts (skin growths), hypogammaglobulinemia (low level of antibodies), infections and myelokathexis (a condition where immune cells are trapped in the bone marrow preventing them from fighting infections).

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

WHIM syndrome is a long-term debilitating and life-threatening condition because of the recurrent infections and the increased risk of developing viral-associated cancer over time.

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

At the time of designation, WHIM syndrome affected less than 0.01 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 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 WHIM syndrome. Patients were given treatment to relieve the symptoms of the condition, including

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\*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 518,400,000 (Eurostat 2019).



granulocyte-colony stimulating factor which stimulates the bone marrow to produce neutrophils and immunoglobulins (proteins naturally found in the blood that help the body to fight infections and other diseases).

### **How is this medicine expected to work?**

Patients with WHIM syndrome have mutations (changes) 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 and, as a consequence, blood cells (particularly neutrophils) are retained in the bone marrow, leading to low levels of neutrophils in the blood.

Mavoxifafor is expected to reduce the activity of the CXCR4 receptor and allow neutrophils to be released from the bone marrow into the blood stream. This increases the levels of neutrophils in the blood thereby helping the body to fight infections.

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

The effects of mavoxifafor have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with mavoxifafor in patients with WHIM syndrome were ongoing.

At the time of submission, mavoxifafor was not authorised anywhere in the EU for the treatment of WHIM syndrome. Orphan designation of mavoxifafor had been granted in the United States for this condition.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 20 June 2019, 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:

Contact details of the current sponsor for this orphan designation can be found on [EMA website](#).

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<sup>1</sup>, Norwegian and Icelandic

Language	Active ingredient	Indication
English	Mavorixafor	Treatment of WHIM syndrome
Bulgarian	Мавориксафор	Лечение на синдрома WHIM
Croatian	Mavoriksafor	Liječenje WHIM sindroma
Czech	Mavorixafor	Léčba syndromu WHIM
Danish	Mavorixafor	Behandling af WHIM syndrom
Dutch	Mavorixafor	Behandeling van de WHIM syndroom
Estonian	Mavoriksafoor	WHIM sündroomi ravi
Finnish	Mavoriksafori	WHIM-oireyhtymän hoito
French	Mavorixafor	Traitement du syndrome WHIM
German	Mavorixafor	Die Behandlung des WHIM Syndroms
Greek	Μαβοριξαφόρη	Θεραπεία του συνδρόμου WHIM
Hungarian	Mavorixafor	WHIM szindróma kezelése
Italian	Mavorixafor	Trattamento della sindrome WHIM
Latvian	Mavoriksafors	WHIM sindroma ārstēšana
Lithuanian	Mavoriksaforas	WHIM sindromo gydymas
Maltese	Mavorixafor	Kura tas-sindrome WHIM
Polish	Maworiksafor	Leczenie zespołu WHIM
Portuguese	Mavorixafor	Tratamento do síndrome de WHIM
Romanian	Mavorixafor	Tratamentul sindromului WHIM
Slovak	Mavorixafor	Liečba syndrómu WHIM
Slovenian	Mavoriksafor	Zdravljenje sindroma WHIM
Spanish	Mavorixafor	Tratamiento del síndrome de WHIM
Swedish	Mavorixafor	Behandling av WHIM syndromet
Norwegian	Mavoriksafor	Behandling av WHIM syndrom
Icelandic	Mavorixafor	Meðferð á WHIM heilkenni

<sup>1</sup> At the time of designation