



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

2 August 2019
EMADOC-628903358-936

Public summary of opinion on orphan designation

Emixustat hydrochloride for the treatment of Stargardt's disease

On 29 May 2019, orphan designation (EU/3/19/2162) was granted by the European Commission to Pharma Gateway AB, Sweden, for emixustat hydrochloride for the treatment of Stargardt's disease.

What is Stargardt's disease?

Stargardt's disease is a hereditary disorder of the eye that leads to progressive loss of sight. In most cases Stargardt's disease is caused by abnormalities in a gene called *ABCA4*. The *ABCA4* gene is responsible for the production of a protein called ABCR that regulates the transport of vitamin A byproducts in and out of some cells in the retina (the light-sensitive surface at the back of the eye). In patients with Stargardt's disease, ABCR does not work properly. As a result, deposits known as lipofuscins, made of fats, proteins and harmful vitamin A byproducts, build up inside the retina cells, which become damaged and eventually die.

Stargardt's disease is a long-term debilitating disease, because the patient's sight becomes progressively worse and eventually leads to blindness.

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

At the time of designation, Stargardt's disease affected approximately 1 in 10,000 people in the European Union (EU). This was equivalent to a total of 52,000 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 for the treatment of Stargardt's disease.

*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).



Supportive care such as vision aids and visual rehabilitation were available. Patients were recommended to wear UV-blocking sunglasses and to avoid vitamin A supplements with the aim of slowing down progression of the disease.

How is this medicine expected to work?

The medicine reduces the action of an enzyme known as RPE65 that helps provide vitamin A to cells in the retina; vitamin A is necessary for vision. In patients with Stargardt's disease, vitamin A byproducts can form harmful substances that are not cleared away. These substances build up in the eyes as part of lipofuscins. By reducing RPE65's activity, the medicine is expected to reduce the build-up of these harmful substances, thus slowing down the loss of vision.

What is the stage of development of this medicine?

The effects of emixustat hydrochloride have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with emixustat hydrochloride in patients with Stargardt's disease were ongoing.

At the time of submission, emixustat hydrochloride was not authorised anywhere in the EU for the treatment of Stargardt's disease. Orphan designation 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 17 April 2019, 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:

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 languages¹, Norwegian and Icelandic

Language	Active ingredient	Indication
English	Emixustat hydrochloride	Treatment of Stargardt's disease
Bulgarian	Емиксустат хидрохлорид	Лечение на болест на Stargardt
Croatian	Emiksustat-hidroklorid	Liječenje Stargardtove bolesti
Czech	Emixustat hydrochlorid	Léčba Stargardtovy choroby
Danish	Emixustathydrochlorid	Behandling af Stargardt sygdom
Dutch	Emixustathydrochloride	Behandeling van de ziekte van Stargardt
Estonian	Emiksustaatesinikloriid	Stargardt'tõve ravi
Finnish	Emiksustaattihydrokloridi	Stargardtin taudin hoito
French	Chlorhydrate d'émixustat	Traitement de la maladie de Stargardt
German	Emixustat-hydrochlorid	Behandlung der Stargardt-Krankheit
Greek	Υδροχλωρική εμιξουστάτη	Θεραπεία της νόσου Stargardt
Hungarian	Emixusztát-hidroklorid	Stargardt-kór kezelése
Italian	Emixustat cloridrato	Trattamento della malattia di Stargardt
Latvian	Emiksustāta hidrohlorīds	Stargardta slimības ārstēšana
Lithuanian	Emiksustato hidrochloridas	Stargardt ligos gydymas
Maltese	Emissustat idroklorur	Kura tal-marda ta' Stargardt
Polish	Chlorowodorek emiksustatu	Leczenie choroby Stargadta
Portuguese	Cloridrato de emixustate	Tratamento da doença de Stargardt
Romanian	Clorhidrat de emixustat	Tratamentul bolii Stargardt
Slovak	Hydrochlorid emixustátu	Liečba Stargardtovej choroby
Slovenian	Emiksustat hidroklorid	Zdravljenje Stargardtove bolezni
Spanish	Emixustato hidrocloruro	Tratamiento de la enfermedad de Stargardt
Swedish	Emixustathydroklorid	Behandling av Stargadts sjukdom
Norwegian	Emiksustathydroklorid	Behandling av Stargadts sykdom
Icelandic	Emixustat hýdróklóríð	Meðferð við Stargadts sjúkdómi

¹ At the time of designation