



EUROPEAN MEDICINES AGENCY
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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Ecopipam for the treatment of Lesch-Nyhan disease

On 3 February 2010, orphan designation (EU/3/09/717) was granted by the European Commission to Dr Alain Munoz, France, for ecopipam for the treatment of Lesch-Nyhan disease.

What is Lesch-Nyhan disease?

Lesch-Nyhan disease is a genetic disease affecting boys that is caused by the lack of an enzyme called hypoxanthine-guanine phosphoribosyl-transferase (HPRT). This enzyme is normally involved in recycling purines, substances used by the body to build cells. The lack of HPRT leads to the purines in the body being destroyed, which causes the waste product uric acid to build up in the blood and to form crystals in the joints and the kidneys. This causes a severe form of gout, with symptoms such as arthritis (pain and inflammation in the joints), tophi (larger deposits of urate crystals that can cause joint and bone damage) and kidney stones. Patients with Lesch-Nyhan disease also have poor muscle control and dystonia (involuntary muscle contractions), which are so severe that patients are unable to walk without assistance and they need help to perform daily activities such as eating and keeping clean. Patients also self-harm, with behaviours such as chewing off of the fingertips and lips, and hitting the head or face if they are not restrained.

Lesch-Nyhan disease is a severe and long-term debilitating disease because the symptoms of the disease appear in the first few years of life and persist throughout life even if they do not get worse with age.

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

At the time of designation, Lesch-Nyhan disease 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 threshold for orphan designation, which is 5 people in 10,000. This is based on the information provided by the sponsor and knowledge of the Committee for Orphan Medicinal Products (COMP).

*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 504,800,000 (Eurostat 2009).



What treatments are available?

At the time of designation, no satisfactory methods were authorised in the EU for the treatment of Lesch-Nyhan disease. Different treatments were used to relieve the symptoms of the disease: for the arthritis, tophi and kidney stones, allopurinol was used to control the overproduction of uric acid, while for the dystonia and self-harming behaviour, patients were offered specially designed wheelchairs with physical restraints. To control self-harming, antipsychotic medicines and sedatives were also used, and in some cases the teeth were removed.

How is this medicine expected to work?

The way that ecopipam is expected to work in Lesch-Nyhan disease is not fully understood, but it is thought to block a group of receptors in the brain that are usually stimulated by the neurotransmitter dopamine (a chemical messenger in the brain). There are different types of dopamine receptors. The ones blocked by ecopipam are of the subtypes D1 and D5. The lack of HPRT in Lesch-Nyhan disease seems to have an impact on the way that dopamine and its receptors work in the brain, which are thought to cause the self-harming behaviour. By blocking the D1/D5 receptors, ecopipam is expected to reduce the self-harming behaviour.

What is the stage of development of this medicine?

The effects of ecopipam have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with the designated product in patients with Lesch-Nyhan disease had been started.

At the time of submission, ecopipam was not authorised anywhere in the EU for Lesch-Nyhan disease. Orphan designation of ecopipam had been granted in the United States of America for the symptomatic treatment of self-injurious behaviours in patients with Lesch-Nyhan disease.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 3 December 2009 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 Community) 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

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

Language	Active ingredient	Indication
English	Escopipam	Treatment of Lesch-Nyhan disease
Bulgarian	Екопипам	Лечение синдрома на Леш-Нихан
Czech	Екопипам	Léčba Lesch-Nyhanovy nemoci
Danish	Escopipam	Behandling af Lesch-Nyhan syndrom
Dutch	Escopipam	Behandeling van Lesch-Nyhan syndroom
Estonian	Ekopipaam	Lesch-Nyhani sündroomi ravi
Finnish	Ekopipaami	Lesch-Nyhanin taudin hoito
French	Escopipam	Traitement du syndrome de Lesch-Nyhan
German	Escopipam	Behandlung des Lesch-Nyhan-Syndroms
Greek	Εκοπιπάμη	Θεραπεία του συνδρόμου Lesch-Nyhan
Hungarian	Ekopipam	A Lesch-Nyhan betegség kezelése
Italian	Escopipam	Trattamento della sindrome di Lesch-Nyhan
Latvian	Ekopipams	Leša-Nīhana (Lesch-Nyhan) sindroma ārstēšanai
Lithuanian	Ekopipamas	Lesch-Nyhan sindromo gydymas
Maltese	Escopipam	Kura tal-marda ta' Lesch-Nyhan
Polish	Ekopipam	Leczenie zespołu Lescha-Nyhana
Portuguese	Escopipam	Tratamento da doença de Lesch-Nyhan
Romanian	Escopipam	Tratamentul sindromului Lesch-Nyhan
Slovak	Ekopipam	Liečba Leschovej - Nyhanovej choroby
Slovenian	Ekopipam	Zdravljenje Lesch-Nyhanovega sindroma
Spanish	Escopipam	Tratamiento del síndrome de Lesch-Nyhan
Swedish	Ekopipam	Behandling av Lesch-Nyhans syndrom
Norwegian	Escopipam	Behandling av Lesch-Nyhan syndrom
Icelandic	Ecópipam	Meðferð á Lesch-Nyhan heilkenni