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Public summary of opinion on orphan designation

Acetazolamide for the treatment of periodic paralysis

On 21 August 2019, orphan designation EU/3/19/2193 was granted by the European Commission to Laboratorios Tillomed Spain, S.L.U, Spain, for acetazolamide for the treatment of periodic paralysis.

What is periodic paralysis?

Periodic paralysis is a group of inherited muscle disorders that causes periodic attacks of weakness or paralysis (inability to move), which resolve spontaneously after some time. The attacks can be triggered by stress, excitement, physical activity, heat or cold. Muscle weakness can affect a small group of muscles or can be more generalised and affect the whole body.

Periodic paralysis is caused by abnormalities in the ion channels, tiny pores in the muscle cells that control the passage of charged particles (ions) such as sodium or chloride and play a key role in the contraction and relaxation of muscles.

Periodic paralysis is a long-term debilitating disorder due to permanent muscle weakness, muscle pain and the need for mobility aids. It can be life threatening because of the risk of heart arrhythmias (unstable heartbeat) which could lead to a heart attack.

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

At the time of designation, periodic paralysis affected approximately 0.2 in 10,000 people in the European Union (EU). This was equivalent to a total of around 10,000 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 submission, no satisfactory methods were authorised in the EU for the treatment of periodic paralysis. Patients received advice on diet and lifestyle to reduce the risk of triggering attacks.

*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).

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How is this medicine expected to work?

Acetazolamide has been available in the EU for many years for various uses including the treatment of certain types of epilepsy, fluid build-up in the tissues (oedema) and glaucoma (high pressure inside the eye). How it works in periodic paralysis is not fully understood, but it is thought to increase the removal of potassium through the urine and also to increase the acidity of the body, which can change the way the ion channels work in muscle cells.

What is the stage of development of this medicine?

The effects of acetazolamide have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with acetazolamide in patients with periodic paralysis had been started.

At the time of submission, acetazolamide was authorised in EU countries for glaucoma, epilepsy and as a diuretic (a medicine to increase urination) in patients with abnormal retention of fluids. It was not authorised anywhere in the EU for the treatment of periodic paralysis or designated as an orphan medicinal product elsewhere for this condition.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 18 July 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 languages[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Acetazolamide	Treatment of periodic paralysis
Bulgarian	Ацетазоламид	Лечение на периодична парализа
Croatian	Acetazolamid	Liječenje periodične paralize
Czech	Acetazolamid	Léčba periodické paralýzy
Danish	Acetazolamid	Behandling af periodisk paralyse
Dutch	Acetazolamide	Behandeling van periodieke verlamming
Estonian	Atsetazoolamiid	Perioodilise paralüüsi ravi
Finnish	Asetatsolamidi	Jaksoittaisen halvauksen hoito
French	Acétazolamide	Traitement de la paralysie périodique
German	Acetazolamid	Behandlung von periodischer Paralyse
Greek	Ακεταζολαμίδη	Θεραπεία της περιοδικής παράλυσης
Hungarian	Acetazolamid	Periodikus paralízis kezelése
Italian	Acetazolamide	Trattamento della paralisi periodica
Latvian	Acetazolamīds	Periodiskas paralīzes ārstēšana
Lithuanian	Acetazolamidas	Šeiminio periodinio paralyžiaus gydymas
Maltese	Aċetazolamide	Kura tal-paraliżi perjodika
Polish	Acetazolamid	Leczenie porażenia okresowego
Portuguese	Acetazolamida	Tratamento da paralisia periódica
Romanian	Acetazolamidă	Tratamentul paraliziei periodice
Slovak	Acetazolamid	Liečba periodického ochrnutia
Slovenian	Acetazolamid	Zdravljenje periodične paralize
Spanish	Acetazolamida	Tratamiento de la parálisis periódica
Swedish	Acetazolamid	Behandling av periodisk paralys
Norwegian	Acetazolamid	Behandling av periodisk paralyse
Icelandic	Asetasólamíð	Meðferð við reglubundnu lömun

[1] At the time of designation