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Public summary of opinion on orphan designation

Setmelanotide for the treatment of Alström syndrome

On 9 January 2020, orphan designation EU/3/19/2245 was granted by the European Commission to TMC Pharma (EU) Limited, Ireland, for setmelanotide for the treatment of Alström syndrome.

What is Alström syndrome?

Alström syndrome is a genetic disease that causes a variety of problems in several organs across the body. Signs and symptoms first occur in infancy and include vision impairment, hearing loss, obesity, diabetes, and problems with the heart, liver, kidneys and lungs.

The condition is caused by a defect in a gene called *ALMS1*, which produces a protein thought to play a role in the function and growth of many types of cells. Alström syndrome is inherited recessively (meaning that both parents must have the defective gene for a child to have the condition).

The condition is debilitating due to its wide-ranging effects and life threatening because it can lead to multiple organ failure. Life expectancy is lower than normal, with patients rarely living beyond 50 years.

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

At the time of designation, Alström syndrome affected approximately 0.01 in 10,000 people in the European Union (EU). This was equivalent to a total of around 500 people. 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 orphan designation, there were no satisfactory treatments for Alström syndrome authorised in the EU. There were clinical guidelines for managing all the complications of this disease.

How is this medicine expected to work?

In patients with Alström syndrome, one of the problems is that the signals that control appetite and how the body produces energy are disrupted. Setmelanotide is a small molecule that is expected to work by stimulating certain nerves in the brain involved in food intake and weight gain. This is

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expected to restore appetite control in patients with Alström syndrome and so reduce their food intake and weight gain.

What is the stage of development of this medicine?

The effects of setmelanotide have been evaluated in experimental models.

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

At the time of submission, setmelanotide was not authorised anywhere in the EU for the treatment of Alström syndrome. Orphan designation of the medicine had been granted in the United States for leptin receptor (LEPR) deficiency obesity.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 5 December 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	Setmelanotide	Treatment of Alström syndrome
Bulgarian	Сетмеланотид	Лечение на синдрома на Алстрьом
Croatian	Setmelanotid	Liječenje Alströmovog sindroma
Czech	Setmelanotid	Léčba Alströmova syndromu
Danish	Setmelanotid	Behandling af Alström syndrom
Dutch	Setmelanotide	Behandeling van het syndroom van Alström
Estonian	Setmelanotiid	Alströmi sündroomi ravi
Finnish	Setmelanotidi	Alströmin oireyhtymän hoito
French	Setmélanotide	Traitement du syndrome d'Alström
German	Setmelanotide	Behandlung des Alström-Syndroms
Greek	Σετμελανοτίδη	Θεραπεία του συνδρόμου Alström
Hungarian	Setmelanotide	Alström szindróma kezelésére
Italian	Setmelanotide	Trattamento della sindrome di Alström
Latvian	Setmelanotīds	Alstrēma (*Alström*) sindroma ārstēšana
Lithuanian	Setmelanotidas	*Alström* sindromo gydymas
Maltese	Setmelanotide	Kura tas-sindromu ta' Alström
Polish	Setmelanotyd	Leczenie zespołu Alströma
Portuguese	Setmelanotido	Tratamento da síndrome de Alström
Romanian	Setmelanotidă	Tratamentul sindromului Alström
Slovak	Setmelanotid	Liečbu Alströmovho syndrómu
Slovenian	Setmelanotid	Zdravljenje Alströmovega sindroma
Spanish	Setmelanotida	Tratamiento del síndrome de Alström
Swedish	Setmelanotid	Behandling av Alströms syndrom
Norwegian	Setmelanotid	Behandling av Alstrøm syndrom
Icelandic	Setmelanótíð	Til meðferðar á Alström-heilkenni

[1] At the time of designation