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Public summary of opinion on orphan designation

Setmelanotide for the treatment of Bardet Biedl syndrome

On 21 August 2019, orphan designation EU/3/19/2192 was granted by the European Commission to TMC Pharma (EU) Limited, Ireland, for setmelanotide for the treatment of Bardet Biedl syndrome.

What is Bardet Biedl syndrome?

Bardet Biedl syndrome is a genetic disorder caused by mutations (changes) in genes responsible for cilia (hair-like structures on cells needed for them to function properly). In Bardet Biedl syndrome, cilia do not work properly leading to variable symptoms including blindness, obesity, reduced kidney function, hormone abnormalities, learning difficulties and polydactyly (extra fingers or toes). The condition is debilitating in the long-term because of the development of visual impairment, speech and learning difficulties, kidney failure and other serious symptoms.

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

At the time of designation, Bardet Biedl syndrome 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 designation, no satisfactory methods were authorised in the EU for the treatment of Bardet Biedl syndrome. Patients mainly received supportive treatment such as management of symptoms as they occur, physical therapy, speech therapy, diet and exercise.

How is this medicine expected to work?

Setmelanotide is a 'melanocortin-4 receptor (MC4R) agonist'. This means that it triggers receptors (targets) on nerve cells that are important for decreasing appetite and increasing the body's energy

*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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use. Patients with Bardet Biedl syndrome have problems regulating body weight, leading to obesity. By triggering these receptors, setmelanotide is expected to enable patients to better control their weight.

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 Bardet Biedl syndrome were ongoing.

At the time of submission, setmelanotide was not authorised anywhere in the EU for the treatment of Bardet Biedl syndrome 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	Setmelanotide	Treatment of Bardet-Biedl syndrome
Bulgarian	Сетмеланотид	Лечение на синдром на Барде-Бидъл
Croatian	Setmelanotid	Liječenje Bardet-Biedlovog sindroma
Czech	Setmelanotid	Léčba Bardetova-Biedlova syndromu
Danish	Setmelanotid	Behandling af Bardet-Biedls syndrom
Dutch	Setmelanotide	Behandeling van het syndroom van Bardet-Biedl
Estonian	Setmelanotiid	Bardet-Biedli sündroomi ravi
Finnish	Setmelanotidi	Bardet-Biedlin oireyhtymän hoito
French	Setmélanotide	Traitement du syndrome de Laurence-Moon-Bardet-Biedl
German	Setmelanotide	Behandlung des Bardet-Biedl-Syndroms
Greek	Σετμελανοτίδη	Θεραπεία του συνδρόμου Bardet-Biedl
Hungarian	Setmelanotide	A Bardet–Biedl-szindróma kezelése
Italian	Setmelanotide	Trattamento della sindrome di Bardet-Biedl
Latvian	Setmelanotīds	Bardē-Bīdla sindroma ārstēšana
Lithuanian	Setmelanotidas	*Bardet-Biedl* sindromo gydymas
Maltese	Setmelanotide	Kura għal Sindrome ta' Bardet-Biedl
Polish	Setmelanotyd	Leczenie zespołu Bardeta-Biedla
Portuguese	Setmelanotido	Tratamento da síndrome de Bardet-Biedl
Romanian	Setmelanotidă	Tratamentul sindromului Bardet-Biedl
Slovak	Setmelanotid	Liečba Bardetovho-Biedlovho syndrómu
Slovenian	Setmelanotid	Zdravljenje Bardet-Biedlovega sindroma
Spanish	Setmelanotida	Tratamiento del Síndrome de Bardet Biedl
Swedish	Setmelanotid	Behandling av Bardet-Biedls syndrom
Norwegian	Setmelanotid	Behandling av Bardet-Biedl syndrom
Icelandic	Setmelanótíð	Meðferð á Bardet-Biedl heilkenni

[1] At the time of designation