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Public summary of opinion on orphan designation

Branaplam for the treatment of spinal muscular atrophy

On 16 April 2018, orphan designation (EU/3/18/2010) was granted by the European Commission to Novartis Europharm Limited, United Kingdom, for branaplam for the treatment of spinal muscular atrophy.

What is spinal muscular atrophy?

Spinal muscular atrophy is an inherited disease usually diagnosed in the first year of life that affects the motor neurons (nerves from the brain and spinal cord that control muscle movements). Patients with the disease lack a protein called 'survival motor neuron' (SMN), which is essential for the normal functioning and survival of motor neurons. Without this protein, the motor neurons deteriorate and eventually die. This causes the muscles to fall into disuse, leading to muscle wasting (atrophy) and weakness.

Spinal muscular atrophy is a long-term debilitating and life-threatening disease because it causes breathing problems and paralysis that worsens over time.

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

At the time of designation, spinal muscular atrophy affected approximately 0.7 in 10,000 people in the European Union (EU). This was equivalent to a total of around 36,000 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, one medicine, Spinraza, was authorised for the treatment of spinal muscular atrophy. Patients also received supportive treatment to help them and their families cope with the symptoms of the disease. This included chest physiotherapy and physical aids to support muscle function, and ventilators to help with breathing.

*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 517,400,000 (Eurostat 2018).

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The sponsor has provided sufficient information to show that the medicine might be of significant benefit for patients with spinal muscular atrophy. Early data showed that branaplam improved muscle function and reduced the need for assisted feeding and ventilation. Also, branaplam is given by mouth while Spinraza is given by intrathecal injection (into the lower back, directly into the spine).

This assumption will need to be confirmed at the time of marketing authorisation, in order to maintain the orphan status.

How is this medicine expected to work?

The SMN protein is produced by two genes, the *SMN1* and *SMN2* genes. Most patients with spinal muscular atrophy lack the *SMN1* gene but have the *SMN2* gene, which mostly produces a short SMN protein that does not work as well as a full-length protein.

Branaplam is expected to enable the *SMN2* gene to produce a full-length protein that works normally. This is expected to increase survival of motor neurons and reduce symptoms of the disease.

What is the stage of development of this medicine?

The effects of branaplam have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with the medicine in patients with spinal muscular atrophy were ongoing.

At the time of submission, the medicine was not authorised anywhere in the EU for spinal muscular atrophy or designated as an orphan medicinal product elsewhere for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 15 March 2018 recommending the granting of this designation.

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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, on the medicine's rare disease designations page.

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	Branaplam	Treatment of spinal muscular atrophy
Bulgarian	Бранаплам	Лечение на спинална мускулна атрофия
Croatian	Branaplam	Liječenje spinalne mišićne atrofije
Czech	Branaplam	Léčba spinální muskulární atrofie
Danish	Branaplam	Behandling af spinal muskelatrofi
Dutch	Branaplam	Behandeling van spinale spieratrofie
Estonian	Branaplaam	Spinaalse lihasatroofia ravi
Finnish	Branaplaami	Spinaalisen lihasatrofian hoito
French	Branaplam	Traitement de l'amyotrophie spinale
German	Branaplam	Behandlung der spinalen Muskelatrophie
Greek	Μπρανάπλάμη	Θεραπεία της νωτιαίας μυϊκής ατροφίας
Hungarian	Branaplám	Spinális izomatrophia kezelése
Italian	Branaplam	Trattamento dell'atrofia muscolare spinale
Latvian	Branaplams	Spinālās muskuļu atrofijas ārstēšana
Lithuanian	Branaplamas	Spinalinės raumenų atrofijos gydymas
Maltese	Branaplam	Kura tal-atrofija muskolari tas-sinsla
Polish	Branaplam	Leczenie rdzeniowego zaniku mięśni
Portuguese	Branaplam	Tratamento da atrofia muscular espinal
Romanian	Branaplamum	Tratamentul amiotrofiei spinale
Slovak	Branaplam	Liečba spinálnej svalovej atrofie
Slovenian	Branaplam	Zdravljenje spinalne mišične atrofije
Spanish	Branaplam	Tratamiento de la atrofia muscular espinal
Swedish	Branaplam	Behandling av spinal muskelatrofi
Norwegian	Branaplam	Behandling av spinal muskelatrofi
Icelandic	Branaplam	Meðferð við mænuvöðvarýrnun

[1] At the time of designation