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Public summary of opinion on orphan designation

Reldesemtiv for the treatment of spinal muscular atrophy

On 28 June 2019, orphan designation EU/3/19/2169 was granted by the European Commission to Pharma Gateway AB, Sweden, for reldesemtiv 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 muscle wasting 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 0.3 in 10,000 people in the European Union (EU). This was equivalent to a total of around 16,000 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, the medicine Spinraza (nusinersen) was authorised for the treatment of spinal muscular atrophy. Spinraza is given by injection into the spine. 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.

The sponsor has provided sufficient information to show that the medicine might be of significant benefit for patients with spinal muscular atrophy. Data from laboratory studies indicate that when used

*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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together with nusinersen this medicine could improve patients' muscle function compared with results seen with Spinraza alone.

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?

This medicine is expected to activate voluntary muscles by attaching to a protein known as troponin, which is involved in muscle contractions. By doing so, the medicine is expected to strengthen the muscles' response to nerve stimulation, thereby improving muscle function in patients with spinal muscular atrophy.

What is the stage of development of this medicine?

The effects of reldesemtiv have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with reldesemtiv in patients with spinal muscular atrophy had been started.

At the time of submission, reldesemtiv was not authorised anywhere in the EU for the treatment of spinal muscular atrophy. Orphan designation of reldesemtiv had been granted in the United States for spinal muscular atrophy.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a Positive opinion on 23 May 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	Reldesemtiv	Treatment of spinal muscular atrophy
Bulgarian	Релдесемтив	Лечение на спинална мускулна атрофия
Croatian	Reldesemtiv	Liječenje spinalne mišićne atrofije
Czech	Reldesemtiv	Léčba spinální muskulární atrofie
Danish	Reldesemtiv	Behandling af spinal muskelatrofi
Dutch	Reldesemtiv	Behandeling van spinale spieratrofie
Estonian	Reldesemtiiv	Spinaalse lihasatroofia ravi
Finnish	Reldesemtiv	Spinaalisen lihasatrofian hoito
French	Reldesemtiv	Traitement de l'amyotrophie spinale
German	Reldesemtiv	Behandlung der spinalen Muskelatrophie
Greek	Ρελντεσεμτίβη	Θεραπεία της νωτιαίας μυϊκής ατροφίας
Hungarian	Reldesemtiv	Spinális izomatrophia kezelése
Italian	Reldesemtiv	Trattamento dell'atrofia muscolare spinale
Latvian	Reldesemtivs	Spinālās muskuļu atrofijas ārstēšana
Lithuanian	Reldesemtivas	Spinalinės raumenų atrofijos gydymas
Maltese	Reldesemtiv	Kura tal-atrofija muskolari tas-sinsla
Polish	Reldesemtiw	Leczenie rdzeniowego zaniku mięśni
Portuguese	Reldesemtiv	Tratamento da atrofia muscular espinal
Romanian	Reldesemtiv	Tratamentul amiotrofiei spinale
Slovak	Reldesemtiv	Liečba spinálnej svalovej atrofie
Slovenian	Reldesemtiv	Zdravljenje spinalne mišične atrofije
Spanish	Reldesemtiv	Tratamiento de la atrofia muscular espinal
Swedish	Reldesemtiv	Behandling av spinal muskelatrofi
Norwegian	Reldesemtiv	Behandling av spinal muskelatrofi
Icelandic	Reldesemtiv	Meðferð við mænuvöðvarýrnunar

[1] At the time of designation