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EMA/COMP/247581/2011
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Viral vector containing DNA encoding the human SMN protein for the treatment of 5q spinal muscular atrophy

On 21 June 2011, orphan designation (EU/3/11/876) was granted by the European Commission to the University of Sheffield, United Kingdom, for viral vector containing DNA encoding the human SMN protein for the treatment of 5q spinal muscular atrophy.

What is 5q spinal muscular atrophy?

5q spinal muscular atrophy is an inherited disease that affects the motor neurons (nerves from the brain and spinal cord that control muscle movements). Patients with the disease lack a protein called 'spinal motor neuron' (SMN) protein, 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. Muscle weakness is usually more severe in the proximal musculature (the muscles closest to the trunk). The disease is linked to a defect on chromosome 5q and is usually diagnosed in the first year of life.

5q spinal muscular atrophy disease 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, 5q spinal muscular atrophy affected approximately 0.3 in 10,000 people in the European Union (EU)[*]. This is equivalent to a total of around 15,000 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 5q spinal muscular atrophy. Patients received supportive treatment to help them and their family to cope

[*]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 27), Norway, Iceland and Liechtenstein. This represents a population of 506,300,000 (Eurostat 2011).

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with the symptoms of the disease. This included chest physiotherapy and physical aids to support muscular function, and ventilators to help with breathing.

How is this medicine expected to work?

This medicine is made of a virus that contains normal copies of the gene responsible for the production of the SMN protein. When injected into the patient, the virus is expected to carry the gene into the brain cells, where the gene is expected to enable the cells to start producing the SMN protein. This is expected to improve muscle function.

The type of virus used in this medicine ('adeno-associated virus'; AAV9) does not cause disease in humans.

What is the stage of development of this medicine?

At the time of submission of the application for orphan designation, the evaluation of the effects of 'viral vector containing DNA encoding the human SMN protein' in experimental models was ongoing.

At the time of submission of the application for orphan designation, no clinical trials with the medicine in patients with 5q spinal muscular atrophy had been started.

At the time of submission, this medicine was not authorised anywhere in the EU for 5q 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 9 March 2011 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

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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	Viral vector containing DNA encoding the human SMN protein	Treatment of 5q spinal muscular atrophy
Bulgarian	Вирусен вектор, съдържащ ДНК кодиране за човешкия SMN протеин	Лечение на 5q спинална мускулна атрофия
Czech	Virový vektor obsahující kȯd DNA pro lidský protein SMN	Léčba 5q spinální muskulární atrofie
Danish	Viral vektor der indeholder DNA kodende for humant SMN-protein	Behandling af 5q spinal muskelatrofi
Dutch	Virale vector welke de DNA-codering voor het humane SMN-proteïne bevat	Behandeling van 5q spinale spieratrofie
Estonian	Viirusvektorit sisaldav DNA, mis kodeerib inimese SMN valku	5q spinaalse lihasatroofia ravi
Finnish	Ihmisen SMN-proteiinia koodaavaa DNA:ta sisältävä virusvektori	Selkärangan 5q lihassurkastuman hoito
French	Vecteur viral porteur d'ADN codant pour la protéine SMN humaine	Traitement de l’amyotrophie spinale 5q
German	Viraler Vektor mit für humanes SMN-Protein kodierender DNA	Behandlung der 5q spinalen Muskelatrophie
Greek	Ιικός φορέας που φέρει DNA που κωδικοποιεί για την ανθρώπινη πρωτεΐνη SMN	Θεραπεία της νωτιαίας μυϊκής ατροφίας (5q)
Hungarian	Humán SMN fehérje DNS kódját tartalmazó vírusvektor	5q spinális izomatrophia kezelése
Italian	Vettore virale contenente il DNA codificante per la proteina umana SMN	Trattamento dell’atrofia muscolare spinale 5q
Latvian	Vīrālie vektori, kas satur cilvēka kustību neironu olbaltumvielu (SMN) DNS kodu	5q spinālas muskuļu atrofijas ārstēšana
Lithuanian	Viruso vektorius su žmogaus SMN baltymą koduojančia DNR	Nugaros raumenų atrofijos gydymas, esant 5q srities delecijoms
Maltese	Vettur virali li fih DNA li jikkodifika l-proteina SMN umana	Kura tal-atrofija muskolari spinali 5q
Polish	Wektor wirusowy zawierający DNA kodujące ludzkie białko SMN	Leczenie rdzeniowego zaniku mięśni 5q
Portuguese	Vector viral contendo código ADN para a proteína SMN humana	Tratamento da atrofia muscular espinal 5q
Romanian	Vector viral conţinând ADN ce codifică proteina SMN umană	Tratamentul amiotrofiei spinale 5q
Slovak	Vírusový vektor obsahujúci DNA kódujúcu ľudský proteín SMN	Liečba 5q spinálnej svalovej atrofie
Slovenian	Virusni vektor z DNK zapisom za humani protein SMN	Zdravljenje 5q spinalne mišične atrofije

[1] At the time of designation

Language	Active ingredient	Indication
Spanish	Vector vírico que contiene ADN que codifica la proteína humana SMN	Tratamiento de la atrofia muscular espinal 5q
Swedish	Virusvektor som innehåller DNA kodande för det humana SMN-proteinet	Behandling av 5q spinal muskelatrofi
Norwegian	Virusvektor som inneholder DNAkodende for humant SMN-protein	Behandling av 5q spinal muskelatrofi
Icelandic	Veiruferja sem inniheldur DNA-kóða fyrir manna SMN-prótein	Meðferð við 5q mænuvöðvarýrnunar