



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

13 December 2016
EMA/685908/2016
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Ascorbic acid for the treatment of facioscapulohumeral muscular dystrophy

On 18 November 2016, orphan designation (EU/3/16/1776) was granted by the European Commission to Université de Montpellier, France, for ascorbic acid for the treatment of facioscapulohumeral muscular dystrophy.

What is facioscapulohumeral muscular dystrophy?

Facioscapulohumeral muscular dystrophy is an inherited condition that causes weakness and wasting of the muscles, usually starting with the muscles of the face, shoulders and arms and gradually extending to the muscles of the torso and lower limbs. Symptoms usually start in adulthood but there is also a more severe form of the disease that starts in childhood (infantile onset).

The condition is long-term debilitating due to progressive muscle weakness and resulting difficulty moving around. There may also be problems with vision and hearing, and the infantile onset form is considered life threatening.

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

At the time of designation, facioscapulohumeral muscular dystrophy affected less than 1 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 51,000 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 to treat facioscapulohumeral muscular dystrophy. Patients mainly received supportive treatment including physical therapies such as physiotherapy and medicines to manage muscle pain and inflammation.

^{*}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 513,700,000 (Eurostat 2016).



How is this medicine expected to work?

Muscle weakness in patients with facioscapulohumeral muscular dystrophy is believed to be linked to a chemical reaction known as oxidation. Ascorbic acid (also known as vitamin C) is an antioxidant that is expected to work by stopping or neutralising these harmful chemical reactions, and thereby improve or delay patients' symptoms.

Ascorbic acid is being developed for use in combination with 3 other medicines with orphan designation: alpha-tocopherol (a form of vitamin E), L-selenomethionine and zinc gluconate.

What is the stage of development of this medicine?

The effects of the medicine 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 facioscapulohumeral muscular dystrophy were ongoing.

At the time of submission, the medicine was not authorised anywhere in the EU for facioscapulohumeral muscular dystrophy 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 6 October 2016 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, 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 languages¹, Norwegian and Icelandic

Language	Active ingredient	Indication
English	Ascorbic acid	Treatment of facioscapulohumeral muscular dystrophy
Bulgarian	Аскорбинова киселина	Лечение на фацио-скапуло-хумерална мускулна дистрофия
Croatian	Askorbinska kiselina	Liječenje fascioskapulohumeralne mišićne distrofije
Czech	Kyselina askorbova	Léčba facioskapulohumerální svalové dystrofie
Danish	Ascorbinsyre	Behandling af facioscapulohumeral muskeldystrofi
Dutch	Ascorbinezuur	Behandeling van facioscapulohumerale spierdystrofie
Estonian	Askorbiinhape	Facioskapulohumeraalse lihasküstroofia ravi
Finnish	Askorbiinihappo	Facioskapulohumeraalisen lihasküstroofian hoito
French	Acide ascorbique	Traitement de la dystrophie musculaire facio-scapulo-humérale
German	Ascorbinsäure	Behandlung der fazioskapulohumeralen Muskeldystrophie
Greek	Ασκορβικό οξύ	Θεραπεία της προσωπομοβραχιόνιας μυϊκής δυστροφίας
Hungarian	Aszkorbinsav	Facioscapulohumeralis izomdisztrófia kezelése
Italian	Acido ascorbico	Trattamento della distrofia muscolare facio-scapolo-omeraale
Latvian	Askorbīnskābe	Facioskapulohumerālās muskuļu distrofijas ārstēšana
Lithuanian	Askorbo rūgštis	Veido-mentės-žasto raumenų distrofijos gydymas
Maltese	Aċidu askorbiku	Kura ta' distrofija muskolari faċjo-skapulo-umerali
Polish	Kwas askorbinowy	Leczenie dystrofii mięśniowej twarzowo-łopatkowo-ramieniowej
Portuguese	Ácido ascórbico	Tratamento da distrofia muscular facioescapuloumeral
Romanian	Acid ascorbic	Tratamentul distrofiei musculare facio-scapulo-humerale
Slovak	Kyselina askorbová	Liečba facioskapulohumerálnej svalovej dystrofie
Slovenian	Askorbinska kislina	Zdravljenje facioskapulohumeralne mišične distrofije
Spanish	Ácido ascórbico	Tratamiento de la distrofia muscular facioescapulohumeral
Swedish	Askorbinsyra	Behandling av facioskapulohumeral muskeldystrofi
Norwegian	Askorbinsyre	Behandling av facioscapulohumeral muskel dystrofi
Icelandic	Askorbínsýra	Meðferð á facioscapulohumeral-vöðvarýrnun

¹ At the time of designation