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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Carbamazepine for the treatment of metaphyseal chondrodysplasia, Schmid type

On 14 October 2016, orphan designation (EU/3/16/1746) was granted by the European Commission to University of Newcastle upon Tyne, United Kingdom, for carbamazepine for the treatment of metaphyseal chondrodysplasia, Schmid type.

What is metaphyseal chondrodysplasia, Schmid type?

Metaphyseal chondrodysplasia, Schmid type is an inherited disease of the bone. It is due to mutations (changes) in the genes responsible for making collagen X, a protein important in the development of cartilage and bone. As a result, abnormal forms of collagen X are produced that build up and interfere with bone development. Abnormal development of the bones causes patients to have short arms and legs, bowing of the legs and abnormal gait. Other bones may also be affected and the patient may have hip deformities.

Metaphyseal chondrodysplasia, Schmid type is debilitating in the long term because it causes pain, and hip and knee deformity.

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

At the time of designation, metaphyseal chondrodysplasia, Schmid type affected less than 0.01 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 500 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).

[*]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).

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What treatments are available?

At the time of designation, no satisfactory methods were authorised to stop limb deformity or joint pain in patients with metaphyseal chondrodysplasia, Schmid type. Painkillers and physiotherapy were used to relieve symptoms, and bone surgery was used to reduce deformities as they developed.

How is this medicine expected to work?

The way carbamazepine works in metaphyseal chondrodysplasia, Schmid type is not fully clear but it is thought to help the break-down of the abnormal collagen X associated with the condition. By reducing the build-up of abnormal collagen X, carbamazepine is expected to allow some normal bone development and reduce symptoms of the condition.

What is the stage of development of this medicine?

The effects of carbamazepine have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with the medicine in patients with metaphyseal chondrodysplasia, Schmid type had been started.

At the time of submission, carbamazepine was authorised in the EU for epilepsy, pain of trigeminal neuralgia and for preventing manic-depressive psychosis in patients with bipolar disorder. At the time of submission, the medicine was not authorised anywhere in the EU for metaphyseal chondrodysplasia, Schmid type 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 13 September 2016 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	Carbamazepine	Treatment of metaphyseal chondrodysplasia, Schmid type
Bulgarian	Карбамазепин	Лечение на метафизарна хондродисплазия, тип Schmid
Croatian	Karbamazepin	Liječenje metafizne hondrodisplazije, tipa Schmid
Czech	Karbamazepin	Léčba metafyzární chondrodysplazie Schmidova typu
Danish	Carbamazepin	Behandling af metafyseal kondrodysplasi, Schmid-type
Dutch	Carbamazepine	Behandeling van metafysaire chondrodysplasie, Schmid-type
Estonian	Karbamasepiin	Schmid-tüüpi metafüsaarse kondrodüsplaasia ravi
Finnish	Karbamatsepiini	Metafyseaalisen kondrodysplasian (tyyppi Schmid) hoito
French	Carbamazépine	Traitement de la chondrodysplasie métaphysaire de type Schmid
German	Carbamazepin	Behandlung von metaphysärer Chondrodysplasie, Typ Schmid
Greek	Καρβαμαζεπίνη	Θεραπεία της μεταφυσιακής χονδροδυσπλασίας τύπου Schmid
Hungarian	Karbamazepin	Schmid-típusú metaphysealis chondrodysplasia kezelése
Italian	Carbamazepina	Trattamento della condrodisplasia metafisaria, tipo Schmid
Latvian	Karbamazepīns	Šmida (*Schmid*) tipa metafizārās hondrodisplāzijas ārstēšana
Lithuanian	Karbamazepinas	Metafizinės chondrodisplazijos gydymas, *Schmid* tipas
Maltese	Carbamazepine	Kura tal-kondrodisplasija metafisarja, tat-tip Schmid
Polish	karbamazepina	Leczenie chondrodysplazji przynasadowej typu Schmida
Portuguese	Carbamazepina	Tratamento de condrodisplasia metafisária de tipo Schmid
Romanian	Carbamazepină	Tratamentul condrodisplaziei metafizare tip Schmid
Slovak	karbamazepín	Liečba metafýzovej chondrodysplázie, Schmidov typ
Slovenian	Karbamazepin	Zdravljenje metafizne hondrodisplazije Schmidovega tipa
Spanish	Carbamazepina	Tratamiento de la condrodisplasia metafisaria tipo Schmid
Swedish	Karbamazepin	Behandling av metafysär kondrodysplasi, Schmid typ
Norwegian	Karbamazepin	Behandling av metafysær chkondrodysplasia, Schmid -typen
Icelandic	Karbamazepín	Meðferð við brjóskrangvexti í beinfal, af Schmid gerð (MCDS-sjúkdómi)

[1] At the time of designation