18 February 2013
EMA/COMP/795251/2012
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Modified recombinant human C-type natriuretic peptide for the treatment of achondroplasia

On 24 January 2013, orphan designation (EU/3/12/1094) was granted by the European Commission to BioMarin Europe Ltd, United Kingdom, for modified recombinant human C-type natriuretic peptide for the treatment of achondroplasia.

What is achondroplasia?

Achondroplasia represents the most common form of short-limb dwarfism, a condition where the bones in the arms and legs do not form properly and are shorter than normal. Patients with achondroplasia have a short stature, an enlarged head with a prominent forehead, bowed legs, ear problems, compression of the spinal cord, as well as short fingers, toes, lower legs and upper arms.

Achondroplasia is an inherited disease caused by a mutation (defect) in a gene called fibroblast growth factor receptor 3 (FGFR3). Patients who have inherited the defective gene from both parents are the most severely affected and normally die around birth or a few months afterwards. In patients with only one defective FGFR3 gene, achondroplasia causes long-term disability and may result in a shorter life span because of its effects on the heart.

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

At the time of designation, achondroplasia affected approximately 0.42 in 10,000 people in the European Union (EU). This was equivalent to a total of around 21,000 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).

*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 509,000,000 (Eurostat 2013).

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What treatments are available?

At the time of designation, no satisfactory methods were authorised in the EU for the treatment of achondroplasia. Patients were given supportive care, as well as surgery to extend limb length, to correct spinal compression or to correct bowed legs.

How is this medicine expected to work?

In patients with achondroplasia, the gene FGFR3 is permanently 'switched on', and this prevents the normal growth of bones, ultimately leading to bones that are shorter than normal. This medicine is expected to work by attaching to a receptor called natriuretic peptide receptor type B (NPR-B) on the surface of cells, which is thought to 'switch off' the activity of FGFR3. This is expected to stimulate the normal growth of bones, thereby improving the symptoms of the disease.

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 the medicine in experimental models was ongoing.

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

At the time of submission, the medicine was not authorised anywhere in the EU for achondroplasia 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 December 2012 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:

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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	Modified recombinant human C-type natriuretic peptide	Treatment of achondroplasia
Bulgarian	Модифициран рекомбинантен човешки С-натриуретичен пептид	Лечение на ахондроплазия
Czech	Modifikovaný rekombinantní lidský natriuretický peptid typu C	Léčba achondroplazie
Danish	Modificeret rekombinant C-type natriuretisk peptid	Behandling af akondroplasi
Dutch	Gemodificeerd recombinant humaan C-type natriuretisch peptide	Behandeling van achondroplasie
Estonian	Modifitseeritud rekombinantne inimese C-tüüpi natriureetiline peptiid	Akondroplaasia ravi
Finnish	Modifioitu, yhdistelmä-DNA-tekniikalla tuotettu C-tyypin natriureettinen peptidi	Akondroplasian hoito
French	Peptide natriurétique de type C recombinant de séquence modifiée	Traitement de l'achondroplasie
German	Modifiziertes rekombinantes C-Typ natriuretisches Peptid	Behandlung der Achondroplasie
Greek	Τροποποιημένο ανασυνδυασμένο νατριουρητικό πεπτίδιο τύπου C	Θεραπεία της Αχονδροπλασίας
Hungarian	Módosított, rekombináns, C-típusú natriuretikus peptid	Achondroplasia (chondrodystrophia) kezelése
Italian	Peptide natriuretico di tipo C modificato tramite tecnica del DNA ricombinante	Trattamento dell'acondroplasia
Latvian	Modificēts rekombinantais C tipa nātrijurētiskais peptīds	Ahondroplāzijas ārstēšana
Lithuanian	Modifikuotas rekombinantinis žmogaus natriuretinis C-tipo peptidas	Achondroplazijos gydymas
Maltese	Peptide natriuretiku uman tat-tip Ċ rikombinanti mmodifikat	Kura tal-akondroplasija
Polish	Rekombinowany zmodyfikowany peptyd natriuretyczny typu C	Leczenie achondroplazji

[1] At the time of designation

Language	Active ingredient	Indication
Portuguese	Péptido natriurético do tipo C recombinante modificado	Tratamento da acondroplasia
Romanian	Peptid natriuretic de tip C uman recombinant modificat	Tratamentul acondroplaziei
Slovak	Modifikovaný rekombinantný ľudský natriuretický peptid typu C	Liečba achondroplázie
Slovenian	Modificirani rekombinantni natriuretični peptid tipa C	Zdravljenje ahondroplazije
Spanish	Péptido natriurético recombinante de tipo C modificado	Tratamiento de la acondroplasia
Swedish	Modifierad rekombinant human natriuretisk peptid av C-typ	Behandling av akondroplasi
Norwegian	Modifisert rekombinant humant C-type natriuretisk peptid	Behandling av akondroplasi
Icelandic	Umbreytt, raðbrigða manna natríumræsihormón af C-gerð	Meðferð við brjóskkröm (e. achondroplasia)