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Public summary of opinion on orphan designation

Soluble recombinant human fibroblast growth factor receptor 3 for the treatment of achondroplasia

On 27 February 2017, orphan designation (EU/3/17/1843) was granted by the European Commission to TherAchon SAS, France, for soluble recombinant human fibroblast growth factor receptor 3 (also known as rhFGFR3) 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 (change) in a gene responsible for making a protein 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.

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

At the time of designation, achondroplasia affected approximately 0.6 in 10,000 people in the European Union (EU). This was equivalent to a total of around 31,000 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 achondroplasia. Patients were given supportive care, as well as surgery to extend limb length, to correct spinal compression or to correct bowed legs.

[*]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 515,700,000 (Eurostat 2017).

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How is this medicine expected to work?

In the body, substances called fibroblast growth factors attach to FGFR3 receptors on cells to regulate cell growth and development. However, in people with achondroplasia, these receptors behave abnormally when fibroblast growth factors attach, and their abnormal activity results in abnormal bone growth.

This medicine is an inactive form of the receptor that mops up available fibroblast growth factors to stop the abnormal receptors from working. Through its action as a decoy, the medicine is expected to reduce the activity of these receptors, thereby helping to restore normal patterns of growth.

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, no clinical trials with the medicine in patients with achondroplasia had been started.

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 19 January 2017 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	Soluble recombinant human fibroblast growth factor receptor 3	Treatment of achondroplasia
Bulgarian	Разтворим рекомбинантен рецептор 3 за човешки фибробластен растежен фактор	Лечение на ахондроплазия
Croatian	Topivi rekombinantni humani receptor 3 fibroblastnog faktora rasta	Liječenje ahondroplazije
Czech	Solubilní rekombinantní lidský fibroblastový růstový faktor receptoru 3	Léčba achondroplazie
Danish	Opløselig rekombinant human fibroblastvækstfaktor receptor 3	Behandling af akondroplasi
Dutch	Vloeibare recombinante humane fibroblast-groeifactorreceptor 3	Behandeling van achondroplasie
Estonian	Lahustuv rekombinantse inimese fibroblasti kasvufaktori retseptor 3	Akondroplaasia ravi
Finnish	Ihmisen liukoinen, rekombinantti fibroblastikasvutekijäreseptori 3	Akondroplasian hoito
French	Récepteur 3 du facteur de croissance des fibroblastes humain recombinant soluble	Traitement de l'achondroplasie
German	Ungebundener rekombinanter humaner Fibroblasten-Wachstumsfaktor-Rezeptor 3	Behandlung der Achondroplasie
Greek	Διαλυτός ανασυνδυασμένος υποδοχέας 3 του ανθρώπινου ινοβλαστικού αυξητικού παράγοντα	Θεραπεία της αχονδροπλασίας
Hungarian	Oldható rekombináns human fibroblaszt növekedési faktorának 3-as receptora	Achondroplasia kezelése
Italian	Recettore 3 ricombinante solubile del fattore di crescita fibroblastico umano	Trattamento dell'acondroplasia
Latvian	Šķīstošs, rekombinants cilvēka fibroblastu augšanas faktora 3. receptors	Ahondroplāzijas ārstēšana
Lithuanian	Tirpaus rekombinantinio žmogaus fibroblastų augimo faktoriaus 3 receptorius	Achondroplazijos gydymas
Maltese	Riċettur 3 tal-fattur tat-tkabbir tal-fibrolasti tal-bniedem rikombinanti	Kura tal-akondroplasija
Polish	Rozpuszczalny rekombinowany ludzki receptor czynnika wzrostu fibroblastów 3	Leczenie achondroplazji
Portuguese	Receptor 3 do fator de crescimento de fibroblastos humanos recombinante, solúvel	Tratamento da acondroplasia
Romanian	Receptor 3 solubil al factorului uman recombinant de creștere fibroblastică	Tratamentul acondroplaziei
Slovak	Solubilný rekombinantný receptor 3 ľudského fibroblastového rastového faktora	Liečba achondroplázie
Slovenian	Topni rekombinantni humani receptor 3	Zdravljenje ahondroplazije

[1] At the time of designation

Language	Active ingredient	Indication
	fibroblastnega rastnega faktorja	
Spanish	Receptor 3 del factor de crecimiento de fibroblasto humano recombinante soluble	Tratamiento de la acondroplasia
Swedish	Löslig rekombinant human fibroblasttillväxtfaktor-receptor 3	Behandling av akondroplasi
Norwegian	Løselig, rekombinant reseptor 3 for human fibroblastvekstfaktor	Behandling av akondroplasi
Icelandic	Raðbrigða viðtaki 3 fyrir manna trefjakímfrumnavaxtarþátt	Meðferð við brjóskkröm (e. achondroplasia)