9 March 2015
EMA/COMP/45098/2009 Rev.1
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

2,2-dimethylbutyric acid, sodium salt for the treatment of beta thalassaemia intermedia and major

First publication	22 June 2009
Rev.1: sponsor's change of address	9 March 2015

Disclaimer

Please note that revisions to the Public Summary of Opinion are purely administrative updates. Therefore, the scientific content of the document reflects the outcome of the Committee for Orphan Medicinal Products (COMP) at the time of designation and is not updated after first publication.

On 27 February 2009, orphan designation (EU/3/09/617) was granted by the European Commission to Isabelle Ramírez, Germany, for 2,2-dimethylbutyric acid, sodium salt for the treatment of beta thalassaemia intermedia and major.

What are beta thalassaemia intermedia and major?

Beta thalassaemia is an inherited disease in which patients are unable to make enough haemoglobin, the protein found in red blood cells that carries oxygen around the body. Beta thalassaemia major is a severe form of the disease in which patients need frequent blood transfusions. Beta thalassaemia intermedia is a less severe form, which may get worse with age.

Beta thalassaemia intermedia and major are debilitating diseases that are long lasting and may be life threatening because of anaemia (the lack of haemoglobin) and the risk of complications associated with blood transfusion (such as iron deposits and infections).

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

At the time of designation, beta thalassaemia intermedia and major affected less than 1 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 50,000 people*, and is

*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.
At the time of designation, this represented a population of 504,800,000 (Eurostat 2009).

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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, the main treatment for beta thalassaemia intermedia and major was blood transfusion. This is usually combined with iron chelators (medicines used to reduce iron levels in the body). High iron levels result from repeated blood transfusions. In some cases, bone marrow transplantation from a matched donor was used to allow the patient to produce red blood cells with normal levels of haemoglobin.

The sponsor has provided sufficient information to show that 2,2-dimethylbutyric acid, sodium salt, a medicine to be taken by mouth, might be of potential significant benefit for the patients because it is expected to reduce the need for blood transfusions. This assumption will need to be confirmed at the time of marketing authorisation, in order to maintain the orphan status.

How is this medicine expected to work?

2,2-Dimethylbutyric acid, sodium salt is expected to increase the amount of haemoglobin in the blood by promoting the production of foetal haemoglobin. This is the main type of haemoglobin found in unborn children. The levels of foetal haemoglobin usually decrease to around 1% of the total haemoglobin after two years of age. In beta thalassaemia, foetal haemoglobin is expected to replace the missing haemoglobin, reducing the need for blood transfusions.

What is the stage of development of this medicine?

The effects of 2,2-dimethylbutyric acid, sodium salt have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials in healthy volunteers were ongoing.

At the time of submission, 2,2-dimethylbutyric acid, sodium salt was not authorised anywhere in the world for beta thalassaemia intermedia and major.

Orphan designation of the medicine had been granted in the United States of America for beta thalassaemia.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 7 January 2009 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

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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	2,2-dimethylbutyric acid, sodium salt	Treatment of beta-thalassaemia intermedia and major
Bulgarian	2,2-диметил-бутирова(маслена)киселина, натриева сол	Лечение на бета таласемия интермедия и майор
Czech	2,2-dimetyl kyselina máslová, sodíková sůl	Léčení beta thalasémie intermedia a major
Danish	2,2-dimethyl-butansyre, natriumsalt	Behandling af beta-thalassæmia intermedia og major
Dutch	2,2-dimethylbutaanzuur, natriumzout	Behandeling van bètathalassemie intermedia en major
Estonian	2,2-dimetüülvõihappe naatriumsool	Keskmise ja raske beetatalasseemia ravi
Finnish	2,2-(dimetyyli)voihappo, natriumsuola	Beetatalassemia intermedia- ja major-tyypin hoito
French	Acide 2,2-diméthylbutyrique, sel de sodium	Traitement de la bêta-thalassémie intermédiaire et majeure
German	2,2-Dimethylbuttersäure, Natriumsalz	Behandlung der Beta-Thalassämie (intermediäre und Major-Form)
Greek	2,2-διμεθυλοβουτυρικό οξύ, άλας του νατρίου	Θεραπεία της β-μεσογειακής αναιμίας, ενδιάμεσης και μείζονος
Hungarian	2,2-dimetil-vajsav, nátriumsó	Béta-talasszémia intermedia és major kezelése
Italian	2,2-dimetilbutirrato sodico	Trattamento della beta-talassemia intermedia e major
Latvian	2,2-dimetilsviestskābes nātrija sāls	Vidēji izteiktas un izteiktas bēta talasēmijas ārstēšana
Lithuanian	2,2-dimetilsviesto rūgštis, natrio druska	Vidutinio sunkumo ir sunkios β-talasemijos gydymas
Maltese	2,2-dimethylbutyric acid, sodium salt	Kura tal-beta talassemija intermedja u maġġuri
Polish	Sól sodowa kwasu 2,2-dimetylomasłowego	Leczenie talasemii beta- intermedia i major
Portuguese	Ácido 2,2-dimetilbutírico, sal sódico	Tratamento da beta talassémia intermédia e major
Romanian	Acid 2,2-dimetilbutiric, sare de sodiu	Tratamentul beta talasemiei intermediare şi majore
Slovak	Kyselina 2,2-dimetylmaslová, sodná soľ	Liečba stredne závažnej a závažnej beta talasémie
Slovenian	2,2-dimetilbutanojska kislina, natrijeva sol	Zdravljenje srednje in velike talasemije beta

[1] At the time of designation

Language	Active Ingredient	Indication
Spanish	Ácido 2,2-dimetilbutírico, sal sódica	Tratamiento de la beta talasemia intermedia y mayor
Swedish	2,2-dimetylsmörsyra, natriumsalt	Behandling av beta-thalassaemia intermedia och major
Norwegian	2,2-dimetylbutansyre, natriumsalt	Behandling av beta-thalassemia intermedia og beta-thalassemia major
Icelandic	2,2-dímetýlbútansýra, natríumsalt	Meðferð á meðalbráðu Beta-Miðjarðarhafsblóðleysi og bráðu Beta-Miðjarðarhafsblóðleysi