16 September 2029
EMADOC-628903358-1033

Public summary of opinion on orphan designation

Gaboxadol monohydrate for the treatment of Angelman syndrome

On 28 June 2019, orphan designation EU/3/19/2172 was granted by the European Commission to FGK Representative Service GmbH, Germany, for gaboxadol monohydrate for the treatment of Angelman syndrome.

What is Angelman syndrome?

Angelman syndrome is an inherited disorder that mainly affects the nervous system (nerve cells, brain and spinal cord). It is caused by a mutation (change) in the gene needed to make the enzyme E6-AP ubiquitin ligase, which is essential for normal development of the nervous system. Children with this condition have delayed development, intellectual disability, severe speech impairment, problems with movement and balance, recurrent seizures (fits) which are often difficult to treat and sociable behaviour with frequent smiling.

Angelman syndrome is a long-term debilitating condition due to the developmental delay, problems with movement and seizures.

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

At the time of designation, Angelman syndrome affected approximately 1 in 10,000 people in the European Union (EU). This was equivalent to a total of around 52,000 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 medicines were authorised in the EU for the treatment of Angelman syndrome. Available treatments were aimed at treating some of the symptoms of the disease and included medicines for epilepsy and behavioural therapy.

*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 518,400,000 (Eurostat 2019).

Official address Domenico Scarlattilaan 6 ● 1083 HS Amsterdam ● The Netherlands
Address for visits and deliveries Refer to www.ema.europa.eu/how-to-find-us
Send us a question Go to www.ema.europa.eu/contact **Telephone** +31 (0)88 781 6000 An agency of the European Union

How is this medicine expected to work?

It is thought that in patients with Angelman syndrome the lack of the enzyme E6-AP ubiquitin ligase leads to reduced levels of GABA, the main substance in nerve cells responsible for switching off electrical activity in the brain, leading to some of the symptoms of the disease. This medicine is expected to attach to $GABA_A$ receptors (targets) found on the surface of nerve cells as GABA would, thereby reducing some of the symptoms of the condition.

What is the stage of development of this medicine?

The effects of gaboxadol monohydrate have been evaluated in experimental models.

At the time of submission of the application for orphan designation, a clinical trial with gaboxadol monohydrate in patients with Angelman syndrome had been completed.

At the time of submission, gaboxadol monohydrate was not authorised anywhere in the EU for the treatment of Angelman syndrome. Orphan designation of this medicine had been granted in the United States for this condition.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 23 May 2019, 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.

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	Gaboxadol monohydrate	Treatment of Angelman syndrome
Bulgarian	Габоксадол монохидрат	Лечение на синдрома на Ангелман
Croatian	Monohidrat gaboksadol	Liječenje Angelmanovog sindroma
Czech	Gaboxadol monohydrát	Léčba Angelmanova syndromu
Danish	Gaboxadolmonohydrat	Behandling af Angelman syndrom
Dutch	Gaboxadolmonohydraat	Behandeling van het syndroom van Angelman
Estonian	Gaboksadoolmonohüdraat	Angelmani sündroomi ravi
Finnish	Gaboksadoli monohydraatti	Angelmanin oireyhtymän hoito
French	Gaboxadol monohydraté	Traitement du syndrome d'Angelman
German	Gaboxadol Monohydrat	Behandlung des Angelman Syndroms
Greek	Γαμποξαδόλη μονοϋδρική	Θεραπεία του συνδρόμου Angelman
Hungarian	Gaboxadol monohidrát	Angelman szindróma kezelése
Italian	Gaboxadol monoidrato	Trattamento della syndrome di Angelman
Latvian	Gaboksadola monohidrāts	Angelmana sindroma ārstēšana
Lithuanian	Gaboksadolio monohidratas	Angelman sindromo gydymas
Maltese	Gaboxadol monohydrate	Kura tas-sindrome ta' Angelman
Polish	Jednowodny gaboksadol	Leczenie zespołu Angelmana
Portuguese	Gaboxadol mono-hidratado	Tratamento da síndrome de Angelman
Romanian	Gaboxadol monohidrat	Tratamentul sindromului Angelman
Slovak	Monohydrát gaboxadol	Liečba Angelmanovho syndrómu
Slovenian	Gaboksadol monohidrat	Zdravljenje Angelmanovega sindroma
Spanish	Gaboxadol monohidrato	Tratamiento del sindrome de Angelman
Swedish	Gaboxadol monohydrat	Behandling av Angelmans syndrom
Norwegian	Gaboksadolmonohydrat	Behandling av Angelmans syndrom
Icelandic	Gaboxadól einhýdrat	Meðferð við Angelman heilkenni

[1] At the time of designation