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EMA/COMP/123657/2015
Committee for Orphan Medicinal Products

# Public summary of opinion on orphan designation

## Tideglusib for the treatment of fragile X syndrome



On 19 March 2015, orphan designation (EU/3/15/1452) was granted by the European Commission to QRC Consultants Ltd., United Kingdom, for tideglusib for the treatment of fragile X syndrome.

### What is fragile X syndrome?

Fragile X syndrome is a genetic disease characterised by moderate to severe mental retardation. Other symptoms include difficulty communicating and socialising, anxiety, hyperactivity, and repetitive and stereotyped behaviours.

The disease is caused by a defect in a gene on the X chromosome. The gene is responsible for the production of a protein called fragile X mental retardation protein (FMRP), which is necessary for the development of the brain. In patients with fragile X syndrome, the defective gene cannot produce normal levels of the FMRP protein and this leads to the mental retardation and other neurological symptoms. Women are normally less severely affected than men, because they have a second X chromosome that usually has a normal copy of the gene.

Fragile X syndrome is a long-term debilitating disease because of the behavioural and mental health problems it causes.

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

At the time of designation, fragile X syndrome affected approximately 2 in 10,000 people in the European Union (EU). This was equivalent to a total of around 103,000 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 fragile X syndrome. Patients were given general support, such as behavioural therapy and special

*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 512,900,000 (Eurostat 2015).

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education, and in some cases, antidepressants, stimulants and antipsychotics were used to treat the symptoms of the disease. Genetic counselling (discussion of the risks of passing the condition on to children) was recommended for families with a history of fragile X syndrome.

## How is this medicine expected to work?

Tideglusib is expected to work by blocking the activity of an enzyme in the brain called glycogen synthase kinase-3β (GSK-3β). In fragile X syndrome, the lack of the FMRP protein leads to excess activity of GSK-3β, which is thought to contribute to the disease. By blocking the activity of GSK-3β, tideglusib is expected to help reduce the severity of the disease symptoms.

## What is the stage of development of this medicine?

The effects of tideglusib have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with tideglusib in patients with fragile X syndrome had been started.

At the time of submission, tideglusib was not authorised anywhere in the EU for fragile X syndrome 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 12 February 2015 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 | Tideglusib | Treatment of fragile X syndrome |
| Bulgarian | Тидеглузиб | Лечение на синдрома на чупливата Х хромозома |
| Croatian | Tideglusib | Liječenje sindroma fragilnog X kromosoma |
| Czech | Tideglusib | Léčba syndromu fragilního X |
| Danish | Tideglusib | Behandling af fragilt X-syndrom |
| Dutch | Tideglusib | Behandeling van het fragiele-X-syndroom |
| Estonian | Tideglusib | Fragiilse X sündroomi ravi |
| Finnish | Tideglusibi | Särö-X-oireyhtymän hoito |
| French | Tidéglusib | Traitement du syndrome de l'X fragile |
| German | Tideglusib | Zur Behandlung des Fragilen-X-Syndroms |
| Greek | Τιδεγκλουσίμπη | Θεραπεία του συνδρόμου εύθραυστου Χ |
| Hungarian | Tideglusib | A fragilis X-szindróma kezelésére |
| Italian | Tideglusib | Trattamento della sindrome dell'X fragile |
| Latvian | Tideglusibs | Trauslā X sindroma ārstēšanai |
| Lithuanian | Tideglusibas | Lūžiosios X chromosomos sindromo gydymas |
| Maltese | Tideglusib | Kura tas-sindrome ta' X fraġli |
| Polish | Tideglusib | Leczenie zespołu łamliwego chromosomu X |
| Portuguese | Tideglusib | Tratamento da síndrome do X frágil |
| Romanian | Tideglusib | Tratamentul sindromului cromozomului X fragil |
| Slovak | Tideglusib | Liečba syndrómu fragilného chromozómu X |
| Slovenian | Tideglusib | Zdravljenje sindroma fragilnega kromosoma X |
| Spanish | Tideglusib | Tratamiento del síndrome de X frágil |
| Swedish | Tideglusib | Behandling av Fragil X-syndrom |
| Norwegian | Tideglusib | Behandling av Fragilt X-syndrom |
| Icelandic | Tídeglúsíb | Meðferð við heilkenni brotgjarns X (fragile X syndrome) |

[1] At the time of designation