Incidence of Aicardi-Goutières syndrome (AGS) and KCNT1-related epilepsy in Denmark

First published: 28/10/2021

Last updated: 05/04/2024

Study Finalised

Administrative details

EU PAS number

EUPAS43860

Study ID

48852

DARWIN EU® study

No

Study countries

Denmark

Study description

Aicardi-Goutières syndrome (AGS) and Potassium channel subfamily T, member 1 (KCNT1)-related epilepsies are rare genetic encephalopathies with epileptic features. Due to the rarity of these diseases, the epidemiology of AGS and KCNT1-related epilepsy remains unclear. Accordingly, the prevalence and incidence for these diseases or the individuals with disease-related genetic mutations remain unknown. This is a retrospective, non-interventional, population-based study using aggregate data extracted from the Danish population register and hospital-based data sources in Denmark to estimate the incidence of AGS and KCNT1-related epilepsies. Cases of AGS and KCNT1-related epilepsy will be identified at three participating study sites: the Danish Epilepsy Centre, Filadelfia for KCNT1, and the Department of Clinical Genetics at the University Hospital Copenhagen and the Centre for Rare Diseases at the Aarhus University Hospital (AUH) for AGS.

Study status

Finalised

Research institutions and networks

Institutions

IQVIA

United Kingdom

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Institution **Non-Pharmaceutical company** **ENCePP partner**

Contact details

Study institution contact

Thorén Robyn PAS_registrations@iqvia.com

Study contact

PAS_registrations@iqvia.com

Primary lead investigator

Thorén Robyn

Primary lead investigator

Study timelines

Date when funding contract was signed

Planned: 26/06/2020

Actual: 06/07/2021

Study start date

Planned: 30/07/2021

Actual: 30/07/2021

Date of final study report

Planned: 23/03/2022

Actual: 22/03/2022

Sources of funding

- Pharmaceutical company and other private sector

More details on funding

Biogen

Study protocol

Biogen_ACS_KCNT-1_Protocol FINAL_June23_2021_CLEAN.pdf (893.53 KB)

Regulatory

Was the study required by a regulatory body?

No

Is the study required by a Risk Management Plan (RMP)?

Not applicable

Methodological aspects

Study type

Study type list

Study topic:

Disease /health condition

Study type:

Non-interventional study

Scope of the study:

Disease epidemiology

Data collection methods:

Secondary use of data

Main study objective:

The primary objective of the study is to estimate the incidence of KCNT1-related epilepsy and AGS in Denmark.

Study Design

Non-interventional study design

Cohort

Other

Non-interventional study design, other

Retrospective, population-based study

Study drug and medical condition

Medical condition to be studied

Aicardi-Goutieres syndrome

Additional medical condition(s)

Epilepsy – AGS & KCNT-1 related

Population studied

Short description of the study population

Patients with Aicardi Goutières syndrome (AGS) and KCNT1-related epilepsy identified from the Danish population register and hospital-based data sources in Denmark.

Inclusion criteria:

The following criteria must be met in order to be included in the AGS or KCNT1-related epilepsy patient population:

• Available information on year and month of birth.

• Confirmed diagnosis of AGS or KCNT1-related epilepsy during the study period

• Available information on year and month of diagnosis

• Patient is resident in Denmark at the time of diagnosis

Exclusion criteria:

• Patients not meeting all inclusion criteria are ineligible for study inclusion.

Age groups

- Infants and toddlers (28 days – 23 months)
- Children (2 to < 12 years)
- Adolescents (12 to < 18 years)
- Adults (18 to < 46 years)
- Adults (46 to < 65 years)
- Adults (65 to < 75 years)
- Adults (75 to < 85 years)
- Adults (85 years and over)

Estimated number of subjects

40

Study design details

Data analysis plan

Age-specific incidence will be analyzed separately for AGS and KCNT1-related epilepsy. Due to the rarity of the diseases, cases will be aggregated by age groups determined by the age distribution of the population at risk and the expected range of ages at diagnoses. Further, overall incidence (for all ages) will be estimated. The incidence rate will be calculated as the number of cases divided by the total size of the corresponding age-specific population (i.e. all residents registered as living in Denmark) in given calendar years or by period, as appropriate. The confidence interval for the incidence rate will be calculated using the Poisson assumption.

Documents

Study results

Biogen KCNT1 AGS report Final v1.0 22Mar2022 - ABSTRACT to ENCePP.pdf (81.8 KB)

Data management

ENCePP Seal

The use of the ENCePP Seal has been discontinued since February 2025. The ENCePP Seal fields are retained in the display mode for transparency but are no longer maintained.

Data sources

Data source(s), other

Danish Registries (access/analysis)

Data sources (types)

Administrative healthcare records (e.g., claims)

Other

Data sources (types), other

Hospital databases from participating study sites

Use of a Common Data Model (CDM)

CDM mapping

No

Data quality specifications

Check conformance

Unknown

Check completeness

Unknown

Check stability

Unknown

Check logical consistency

Unknown

Data characterisation

Data characterisation conducted

No