Platform-Residras and Residras

First published: 01/02/2024

Last updated: 17/10/2024

Data source | Human | Disease registry | Primary care medical records | Pharmacy dispensing records | Other

Administrative details

Administrative details

Data source ID

49905

Data source acronym

Dravet-SCN1A-PCHD19 Registry

Data holder

Dravet Italia Onlus

Data source type

Disease registry

Primary care medical records

Pharmacy dispensing records

Other

Data source type, other

Prescription event monitoring,prospective studies database,case-control surveillance database

Main financial support

Funds from patients organisations, charity and foundations

Funding from industry or contract research

Care setting

Primary care – specialist level (e.g. paediatricians)

Secondary care – specialist level (ambulatory)

Hospital inpatient care

Hospital outpatient care

Data source qualification

If the data source has successfully undergone a formal qualification process (e.g., from the EMA, ISO or other certifications), this should be described.

Yes

Description of the qualification

Medical Validation

Data source website

https://www.dravet-registry.com

Contact details

Isabella Brambilla info@dravet-registry.com

Main

info@dravet-registry.com

Renzo Guerrini renzo.guerrini@meyer.it

Alternate

renzo.guerrini@meyer.it

Data source regions and languages

Data source countries

Italy

Data source languages

English

Data source establishment

Data source established

15/06/2013

Data source time span

First collection: 15/06/2013

The date when data started to be collected or extracted.

Publications

Data source publications

A registry for Dravet syndrome: the Italian experience. Epilepsia Open 2023 . doi: 10.1002/epi4.12730. Online ahead of print.

Multicenter prospective longitudinal study in 34 patients with Dravet syndrome: Neuropsychological development in the first six years of life - Brain & Development 43 (2021) 419–430 - https://doi.org/10.1016/j.braindev.2020.10.004

Dravet syndrome: Early electroclinical findings and long-term outcome in adolescents and adults - Epilepsia. 2019,60(S3):S49–S58. - DOI: 10.1111/epi.16297

Efficacy and safety of Fenfluramine hydrochloride for the treatment of seizures in Dravet syndrome: A real-world study - Epilepsia. 2020,00:1–10. - DOI: 10.1111/epi.16690

Clinical and genetic factors predicting Dravet syndrome in infants with SCN1A mutations - Neurology® 2017,88:1–8. doi: 10.1212/WNL.0000000000003716.&#xd,

Data elements collected

The data source contains the following information

Disease information

Does the data source collect information with a focus on a specific disease? This might be a patient registry or other similar initiatives.

Yes

Disease details (other)

Rare diseases involved: Dravet Syndorme with/without SCN1A mutation/deletion, Other Syndrome related SCN1A Mutation/Deletion and

Syndrome related PCDH19 Mutation/Deletion.
Disease information collected: Personal and Familiar history, genetic investigation, seizure Onset, Follow up seizures, neurological follow up, treatment follow up, EEG and other exams follow up, grow and cardio parameters, Hospitalisation, exceptional events, gait analysis follow up.

Rare diseases

Are rare diseases captured? In the European Union a rare disease is one that affects no more than 5 people in 10,000.

Yes

Pregnancy and/or neonates

Does the data source collect information on pregnant women and/or neonatal subpopulation (under 28 days of age)?

Yes

Hospital admission and/or discharge

Yes

ICU admission

Is information on intensive care unit admission available?

Yes

Cause of death

Captured

Cause of death vocabulary

Not coded (Free text)

Prescriptions of medicines

Not Captured

Dispensing of medicines

Not Captured

Advanced therapy medicinal products (ATMP)

Is information on advanced therapy medicinal products included? A medicinal product for human use that is either a gene therapy medicinal product, a somatic cell therapy product or a tissue engineered products as defined in Regulation (EC) No 1394/2007 [Reg (EC) No 1394/2007 Art 1(1)].

No

Contraception

Is information on the use of any type of contraception (oral, injectable, devices etc.) available?

No

Indication for use

Does the data source capture information on the therapeutic indication for the use of medicinal products?

Not Captured

Medical devices

Is information on medicinal devices (e.g., pens, syringes, inhalers) available?

No

Administration of vaccines

Yes

Procedures

Does the data source capture information on procedures (e.g., diagnostic tests, therapeutic, surgical interventions)?

Not Captured

Healthcare provider

Is information on the person providing healthcare (e.g., physician, pharmacist, specialist) available? The healthcare provider refers to individual health professionals or a health facility organisation licensed to provide health care diagnosis and treatment services including medication, surgery and medical devices.

Yes

Clinical measurements

Is information on clinical measurements (e.g., BMI, blood pressure, height) available?

Yes

Genetic data

Are data related to genotyping, genome sequencing available?

Captured

Genetic data vocabulary

Other

Genetic data vocabulary, other

HPO

Biomarker data

Does the data source capture biomarker information? The term “biomarker” refers to a broad subcategory of medical signs (objective indications of medical state observed from outside the patient), which can be measured accurately and reproducibly. For example, haematological assays, infectious disease markers or metabolomic biomarkers.

Captured

Biomarker data vocabulary

HPO

Patient-reported outcomes

Is information on patient-reported outcomes (e.g., quality of life) available?

Yes

Patient-generated data

Is patient-generated information (e.g., from wearable devices) available?

No

Units of healthcare utilisation

Are units of healthcare utilisation (e.g., number of visits to GP per year, number of hospital days) available or can they be derived? Units of healthcare utilisation refer to the quantification of the use of services for the purpose of preventing or curing health problems.

Yes

Unique identifier for persons

Are patients uniquely identified in the data source?

Yes

Diagnostic codes

Captured

Diagnosis / medical event vocabulary

Orphanet Rare Disease Ontology (ORDO)

Human Phenotype Ontology (HPO)

Other

Diagnosis / medical event vocabulary, other

Unifield Medical Language

Medicinal product information

Captured

Medicinal product information collected

Formulation

Dose

Medicinal product vocabulary

Not coded (Free text)

Quality of life measurements

Captured

Quality of life measurements vocabulary

Not coded (Free text)

Lifestyle factors

Captured

Lifestyle factors

Diet

Other

Sociodemographic information

Captured

Sociodemographic information collected

Age

Gender

Country of origin

Other

Quantitative descriptors

Population Qualitative Data

Population age groups

Paediatric Population (< 18 years)

Preterm newborn infants (0 – 27 days)

Term newborn infants (0 – 27 days)

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)

Elderly (≥ 65 years)

Adults (65 to < 75 years)

Adults (75 to < 85 years)

Adults (85 years and over)

Estimated percentage of the population covered by the data source in the catchment area

Since it is a rare disease, it is hoped that as many patients as possible will be included.

Description of the population covered by the data source in the catchment area whose data are not collected (e.g., people who are registered only for private care)

Internation-wide

Family linkage

Family linkage available in the data source permanently or can be created on an ad hoc basis

Ad hoc

Population

Population size

631

Active population size

629

Population by age group

Age group	Population size	Active population size
Paediatric Population (< 18 years)	422	420
Preterm newborn infants (0 – 27 days)	0	0
Term newborn infants (0 – 27 days)	0	0
Infants and toddlers (28 days – 23 months)	6	6
Children (2 to < 12 years)	280	278
Adolescents (12 to < 18 years)	136	136
Adults (18 to < 46 years)	171	171
Adults (46 to < 65 years)	15	15
Elderly (≥ 65 years)	3	3
Adults (65 to < 75 years)	3	3
Adults (75 to < 85 years)	0	0
Adults (85 years and over)	0	0

Median observation time

Median time (years) between first and last available records for unique individuals captured in the data source

12.00

Median time (years) between first and last available records for unique active individuals (alive and currently registered) capt

12.00

Data flows and management

Access and validation

Governance details

Documents or webpages that describe the overall governance of the data source and processes and procedures for data capture and management, data quality check and validation results (governing data access or utilisation for research purposes).

https://www.dravet-registry.com/residras-request-data

Biospecimen access

Are biospecimens available in the data source (e.g., tissue samples)?

No

Access to subject details

Can individual patients/practitioners/practices included in the data source be contacted?

Yes

Description of data collection

The referring doctor of the accredited centre (with the approval of their Ethical Committee), following the patient's examination, enters the required data in the registry at least once a year

Event triggering registration

Event triggering registration of a person in the data source

Disease diagnosis

Event triggering de-registration of a person in the data source

Other

Event triggering de-registration of a person in the data source, other

Patient's withdrawal of consent as required by GDPR

Event triggering creation of a record in the data source

Onset and Visit follow up

Data source linkage

Linkage

Is the data source described created by the linkage of other data sources (prelinked data source) and/or can the data source be linked to other data source on an ad-hoc basis?

No

Data management specifications that apply for the data source

Data source refresh

Yearly

Informed consent for use of data for research

Other

Possibility of data validation

Can validity of the data in the data source be verified (e.g., access to original medical charts)?

Yes

Data source preservation

Are records preserved in the data source indefinitely?

Yes

Approval for publication

Is an approval needed for publishing the results of a study using the data source?

Yes

Informed consent, other

There is a committee to evaluate requests for data access

Data source last refresh

15/06/2023

Common Data Model (CDM) mapping

CDM mapping

Has the data source been converted (ETL-ed) to a common data model?

No