International Registries for Rare Conditions Affecting Sex Development & Maturation (SDMregistries; formerly known as I-DSD/I-CAH/I-TS /I-HH)

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Data source Human Disease registry

Administrative details

Administrative details

Data source ID

1000000994

Data source acronym

SDMregistries

Data holder

The Office for Rare Conditions Registries (ORC Registries)

Data source type

Disease registry

Main financial support

Funding by own institution

Funding from industry or contract research

Care setting

Hospital outpatient care

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

Secondary care – specialist level (ambulatory)

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.

No

Data source website

sdmregistries.org

Contact details

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Main

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Data source regions and languages

Data source countries

Angola

Argentina

Armenia

Australia

Austria

Bangladesh

Belgium

Bosnia and Herzegovina

Brazil

Bulgaria

Cameroon

Canada

China

Croatia

Cyprus

Czechia

Denmark

Egypt

France

Georgia

Germany

Greece

Hong Kong

Hungary

India

Indonesia

Ireland

Israel

Italy

Japan

Jordan

Kenya

Kuwait

Lithuania

Malaysia

Mexico

Montenegro

Morocco

Netherlands

Nigeria

Norway

Oman

Pakistan

Philippines

Poland

Portugal

Romania

Russian Federation

Serbia

Singapore

Slovakia

South Africa

Spain

Sri Lanka

Sudan

Sweden

Switzerland

Taiwan

Tanzania, United Republic of

Thailand

Türkiye

Uganda

Ukraine

United Arab Emirates

United Kingdom

United States

Uzbekistan

Viet Nam

Data source languages

English

Data source establishment

Data source established

17/12/2008

Data source time span

First collection: 17/12/2008

The date when data started to be collected or extracted.

Publications

Data source publications

The European disorder of sex development registry: a virtual research environment

An assessment of the quality of the I-DSD and the I-CAH registries - international registries for rare conditions affecting sex development

The I-CAH Registry: A platform for international collaboration for improving knowledge and clinical care in congenital adrenal hyperplasia

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

Disorder of sex development

Hyperplasia adrenal

Turner's syndrome

Hypogonadism

Congenital hypogonadotropic hypogonadism

Klinefelter's syndrome

Disease details (other)

Disorder of gonadal development, Disorder of androgen synthesis, Disorder of androgen action, Leydig cell defects, Persistent mullerian duct syndrome, Defects of mullerian development, Non-specific XY DSD, Cloacal anomaly, Chromosomal DSD

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

No

ICU admission

Is information on intensive care unit admission available?

No

Cause of death

Captured

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

No

Procedures

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

Captured

Procedures vocabulary

Not coded (Free text)

Other

Procedures vocabulary, other

Defined list of procedures

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

Free text

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

Other

Biomarker vocabulary, other

Defined list of biomarkers

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.

No

Unique identifier for persons

Are patients uniquely identified in the data source?

Yes

Diagnostic codes

Not Captured

Medicinal product information

Captured

Medicinal product information collected

Brand name

Dosage regime

Dose

Formulation

Route of administration

Quality of life measurements

Not Captured

Lifestyle factors

Not Captured

Sociodemographic information

Captured

Sociodemographic information collected

Age

Country of origin

Gender

Sex

Quantitative descriptors

Population Qualitative Data

Population age groups

All

In utero

Paediatric Population (< 18 years)

Neonate

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)

Adult and elderly population (≥18 years)

Adults (18 to < 65 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

Information on patients who consented to the registry can only entered and used for research.

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)

Information on patients who have not consented to the registry can only entered and used for research.

Family linkage

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

Ad hoc

Population

Population size

11169

Active population size

11158

Population by age group

Age group	Population size	Active population size
Paediatric Population (< 18 years)	4978	4973
Adult and elderly population (≥18 years)	6191	6185

Median observation time

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

3.00

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

3.00

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).

sdmregistries.org/about/

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 Registry was conceived following the Chicago Consensus meeting in 2005 and the prototype ESPE DSD Register became the EuroDSD Registry in 2008 and the International DSD (I-DSD) Registry in 2011. The original platform subsequently developed dedicated modules for CAH (I-CAH, 2014), TS (I-TS, 2022), HH (I-HH, 2025) and this common platform was renamed the SDMregistries platform. Klinefelter's Syndrome module is currently being developed.

In 2026, SDMregistries has a network reaching 264 centres in 70 countries on all the continents. Of these, 180 active centres from 48 countries use the registries and had entered over 11,000 cases for supporting research.

The platform is managed by the Office for Rare Conditions Registries team at the University of Glasgow and it is supported through a wide range of sources that includes fees incurred by investigators for obtaining data for research, project grants, income from the biennial symposium. For further information visit the Studies and Learning & Training pages.

Event triggering registration

Event triggering registration of a person in the data source

Birth

Disease diagnosis

Start of treatment

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

Death

Emigration

Loss to follow up

Practice deregistration

Event triggering creation of a record in the data source

New patient in the specialist clinic/centre or existing patients in the specialist clinic/centre who started to contribute data to the registry.

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?

Yes

Linkage description, pre-linked

The data is pseudonymized but there are potential data linkages. These linkages however are only available to the Registry team and not provided to any study PI.

Linkage description, possible linkage

Date of birth, Sex at birth, current gender.

Data management specifications that apply for the data source

Data source refresh

Every 6 months

Informed consent for use of data for research

Required for all studies

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?

No

Approval for publication

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

No

Data source last refresh

15/03/2026

Common Data Model (CDM) mapping

CDM mapping

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

No