- Cohort of Patients with Alport Syndrome

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General

Identification

Detailed name	Cohort of Patients with Alport Syndrome

General Aspects

Medical area	Rare diseases
Health determinants	Genetic
Keywords	Alport syndrome, hereditary disease, rare disease

Scientific investigator(s) (Contact)

Name of the director	Heidet
Surname	Laurence
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Organization	Necker Hospital for Children

Collaborations

Participation in projects, networks and consortia	Yes
Details	Under the RADICO (Rare Disease Cohorts) programme, the RaDiCo-EURBIO-Alport will be implemented to expand the Alport syndrome database across Europe.

Funding

Funding status	Private
Details	Association for Information and Research on Renal Genetic Diseases

Governance of the database

Sponsor(s) or organisation(s) responsible	Centre de référence Maladies Rénales Héréditaires de l'Enfant et de l'Adulte - Société française de néphrologie
Organisation status	Private

Additional contact

Main features

Type of database

Type of database	Study databases
Study databases (details)	Cohort study
Database recruitment is carried out by an intermediary	A selection of health institutions and services
Database recruitment is carried out as part of an interventional study	No
Additional information regarding sample selection.	From several adult and paediatric nephrology centres.

Database objective

Main objective	Knowledge of the natural history of the disease. Understanding the disease mechanisms for developing kidney failure.
Inclusion criteria	Patients with Alport syndrome.

Population type

Age	Newborns (birth to 28 days) Infant (28 days to 2 years) Early childhood (2 to 5 years) Childhood (6 to 13 years) Adolescence (13 to 18 years) Adulthood (19 to 24 years) Adulthood (25 to 44 years) Adulthood (45 to 64 years) Elderly (65 to 79 years) Great age (80 years and more)
Population covered	Sick population
Gender	Male Woman
Geography area	National

Detail of the geography area	France

Data collection

Dates

Size of the database

Size of the database (number of individuals)	< 500 individuals
Details of the number of individuals	440

Data

Database activity	Current data collection
Type of data collected	Clinical data Biological data
Clinical data (detail)	Direct physical measures
Details of collected clinical data	Family history (consanguinity, blood in urine, hearing loss, ocular abnormalities, anomalies in collagen expression); transmission method; molecular studies; extrarenal signs; kidney biopsy; immunofluorescence studies.
Biological data (detail)	Microalbuminuria and proteinuria; renal function; renin-angiotensin system inhibitors
Presence of a biobank	No
Health parameters studied	Health event/morbidity

Procedures

Participant monitoring	Yes
Links to administrative sources	No

Promotion and access

Promotion

Access

Terms of data access (charter for data provision, format of data, availability delay)	Contact the scientist in charge.

Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only