

- 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 Héritaires 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
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Data collection

Dates

Size of the database

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

Database activity	Current data collection
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Type of data collected	Clinical data Biological data
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Clinical data (detail)	Direct physical measures
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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.
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Biological data (detail)	Microalbuminuria and proteinuria; renal function; renin-angiotensin system inhibitors
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Presence of a biobank	No
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Health parameters studied	Health event/morbidity
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Procedures

Participant monitoring	Yes
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Links to administrative sources	No
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Promotion and access

Promotion

Access

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

Access on specific project only

Access to individual data

Access on specific project only