

- 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
Address	149 rue de Sèvres 75015 PARIS
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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
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