RaDiCo-EURBIO-Alport - Study of the natural history of Alport Syndrome by establishment of an International database

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General

Identification

Detailed name	Study of the natural history of Alport Syndrome by establishment of an International database
Sign or acronym	RaDiCo-EURBIO-Alport
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	N° CCTIRS 16-087 / N° CPP 14130 ND / N° CNIL 916204 / N° MESR DC-2015-2564

General Aspects

Medical area	Cardiology Ophthalmology Otolaryngology or ENT Pediatrics Rare diseases Urology, andrology and nephrology
Study in connection with Covid-19	No
Pathology (details)	Alport Syndrome (AS) is an inherited disease characterized by the association of a glomerular nephropathy, a sensorineural deafness, and retinal or corneal defects. Its frequency is about 1/5000. It is associated with mutations in one of the three genes encoding the alpha 3, 4, and 5 chains of type IV collagen, which form a distinct network in the glomerular basement membrane essential for the long-term stability of the glomerular filtration barrier. The disease can be inherited as a dominant X-linked, autosomal recessive, or autosomal dominant trait. Patients initially present with hematuria, followed by proteinuria and progressive renal failure. The median age at end-stage renal failure is about 20, but there is a large inter- and intra-familial variability. The progression of the disease can be divided into 4 stages: isolated hematuria, microalbuminuria, macroproteinuria, and

	progressive renal failure. Ear and ocular defects also exhibit progressive evolution.

Scientific investigator(s) (Contact)

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Unit	U1151
Organization	French National Institute for Health and Medical Research (Inserm)

Collaborations

Participation in projects, networks and consortia	Yes
Details	Rare Disease Healthcare Pathway (Orkid) / French national reference center for hereditary kidney diseases in children and adults (MARHEA)/ European Reference Network ERK-NET

Funding

Funding status	Public
Details	The RaDiCRaDiCo-EURBIO cohort initially received funding from the state managed by the National Research Agency (ANR) as part of the "Investissements d'Avenir" cohorts program.

Governance of the database

Sponsor(s) or organisation(s) responsible	French National Institute for Health and Medical Research (Inserm)
Organisation status	Public
Presence of scientific or steering committees	Yes
Labelling and database evaluation	Security audit certification of the database. Data management and continuous quality control of data.

Additional contact

Main features

Type of database

Type of database	Morbidity registers
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.	Paediatric and adult patients will be mainly recruited through the network of reference competence and recognised expert centres of rare kidney diseases. Investigators will inform patients meeting the inclusion criteria about the RaDiCo-EURBIO-Alport cohort and invite them to participate during regular care follow-up visit for prevalent patient and during their first regular care visit (postdiagnosis) for incident patient.

Database objective

Main objective	The main objective is to study the natural history of the Alport Syndrome.
Inclusion criteria	The inclusion criteria are : - Diagnosis of AS based on (i) electron microscopic examination of the renal biopsy and/or (ii) molecular studies and/or (iii) abnormal expression of type IV collagen chains on skin and/or glomerular basement membranes. - Signed informed consent

	There are no exclusion criteria.
Population type	
Age	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
Pathology	Q64 - Other congenital malformations of urinary system
Gender	Male Woman
Geography area	International
Data collection	
Dates	
Date of first collection (YYYY or MM/YYYY)	2017
Date of last collection (YYYY or MM/YYYY)	2025
Size of the database	
Size of the database (number of individuals)	[500-1000[individuals
Details of the number of individuals	642
Data	
Database activity	Current data collection
Type of data collected	Clinical data Declarative data Paraclinical data Biological data
Details of collected clinical data	The main variables collected, in addition to the CEMARA data already imported, are: demographics,

	family history, ocular symptoms, data on deafness and audiogram, treatments, molecular diagnosis, biochemical and renal parameters (ESRD, dialysis...), clinical examinations, as well as self-questionnaires on quality of life.
Declarative data (detail)	Paper self-questionnaire Internet self-questionnaire Face to face interview
Details of collected declarative data	SF-36 (adults) / SF-10 (children)
Biological data (detail)	Biochemical, hematological, and renal parameters (ESRD, dialysis...)
Presence of a biobank	Yes
Contents of biobank	Fluids (saliva, urine, amniotic fluid, ?)
Details of biobank content	urine
Health parameters studied	Health event/morbidity Health event/mortality Quality of life/health perception
Procedures	
Data collection method	eCRF in secure web access, secure cloud and HADS hosting
Quality procedure(s) used	Data Management Plan and Data Validation Plan. Continuous data management (automatic control rules and "queries" system).
Participant monitoring	Yes
Monitoring procedures	Monitoring by convocation of the participant Monitoring by contact with the referring doctor
Links to administrative sources	No
Promotion and access	
Promotion	
Access	
Presence of document that lists variables and coding procedures	Yes
Terms of data access (charter	Requests for access to RaDiCo-EURBIO data

for data provision, format of data, availability delay)	(aggregated or individual) will be reviewed by the scientific committee following the submission of a synopsis of a Specific Research Project (PRS), as defined in the Access to Resources Charter. Requests should be sent to eurbio@radico.fr.
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only