

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

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| Général                                                      |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
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| Identification                                               |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
| Nom détaillé                                                 | Study of the natural history of Alport Syndrome by establishment of an International database                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |
| Sigle ou acronyme                                            | RaDiCo-EURBIO-Alport                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              |
| Numéro d'enregistrement (ID-RCB ou EUDRACT, CNIL, CPP, etc.) | N° CCTIRS 16-087 / N° CPP 14130 ND / N° CNIL 916204 / N° MESR DC-2015-2564                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
| Thématiques générales                                        |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
| Domaine médical                                              | Cardiology<br>Ophthalmology<br>Otolaryngology or ENT<br>Pediatrics<br>Rare diseases<br>Urology, andrology and nephrology                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          |
| Etude en lien avec la Covid-19                               | No                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |
| Pathologie, précisions                                       | 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. |

## Responsable(s) scientifique(s)

|                    |                                                                          |
|--------------------|--------------------------------------------------------------------------|
| Nom du responsable | Heidet                                                                   |
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| Nom du responsable | Knebelmann                                                                                      |
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| Laboratoire        | U1151                                                                                           |
| Organisme          | French National Institute for Health and Medical<br>Research (Inserm)                           |

## Collaborations

|                                                           |     |
|-----------------------------------------------------------|-----|
| Participation à des projets, des réseaux, des consortiums | Yes |
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| Précisions | Rare Disease Healthcare Pathway (Orkid) / French national reference center for hereditary kidney diseases in children and adults (MARHEA)/ European Reference Network ERK-NET |
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## Financements

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| Financements | Public |
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| Précisions | 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. |
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## Gouvernance de la base de données

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|--------------------------------|--------------------------------------------------|
| Organisation(s) responsable(s) | French National Institute for Health and Medical |
|--------------------------------|--------------------------------------------------|

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| ou promoteur                                                                                  | Research (Inserm)                                                                                                                                                                                                                                                                                                                                                                                                                          |
| Statut de l'organisation                                                                      | Secteur Public                                                                                                                                                                                                                                                                                                                                                                                                                             |
| Existence de comités scientifique ou de pilotage                                              | Yes                                                                                                                                                                                                                                                                                                                                                                                                                                        |
| Labellisations et évaluations de la base de données                                           | Security audit certification of the database. Data management and continuous quality control of data.                                                                                                                                                                                                                                                                                                                                      |
| Contact(s) supplémentaire(s)                                                                  |                                                                                                                                                                                                                                                                                                                                                                                                                                            |
| Caractéristiques                                                                              |                                                                                                                                                                                                                                                                                                                                                                                                                                            |
| Type de base de données                                                                       |                                                                                                                                                                                                                                                                                                                                                                                                                                            |
| Type de base de données                                                                       | Morbidity registers                                                                                                                                                                                                                                                                                                                                                                                                                        |
| Base de données issues d'enquêtes, précisions                                                 | Cohort study                                                                                                                                                                                                                                                                                                                                                                                                                               |
| Origine du recrutement des participants                                                       | A selection of health institutions and services                                                                                                                                                                                                                                                                                                                                                                                            |
| Le recrutement dans la base de données s'effectue dans le cadre d'une étude interventionnelle | No                                                                                                                                                                                                                                                                                                                                                                                                                                         |
| Informations complémentaires concernant la constitution de l'échantillon                      | 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. |
| Objectif de la base de données                                                                |                                                                                                                                                                                                                                                                                                                                                                                                                                            |
| Objectif principal                                                                            | The main objective is to study the natural history of the Alport Syndrome.                                                                                                                                                                                                                                                                                                                                                                 |
| Critères d'inclusion                                                                          | <p>The inclusion criteria are :</p> <ul style="list-style-type: none"> <li>- 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.</li> <li>- Signed informed consent</li> </ul>                                                                                       |

There are no exclusion criteria.

| Type de population                                   |                                                                                                                                                                                                                                                                                                   |
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| Age                                                  | Early childhood (2 to 5 years)<br>Childhood (6 to 13 years)<br>Adolescence (13 to 18 years)<br>Adulthood (19 to 24 years)<br>Adulthood (25 to 44 years)<br>Adulthood (45 to 64 years)<br>Elderly (65 to 79 years)<br>Great age (80 years and more)                                                |
| Population concernée                                 | Sick population                                                                                                                                                                                                                                                                                   |
| Pathologie                                           | Q64 - Other congenital malformations of urinary system                                                                                                                                                                                                                                            |
| Sexe                                                 | Male<br>Woman                                                                                                                                                                                                                                                                                     |
| Champ géographique                                   | International                                                                                                                                                                                                                                                                                     |
| Collecte                                             |                                                                                                                                                                                                                                                                                                   |
| Dates                                                |                                                                                                                                                                                                                                                                                                   |
| Année du premier recueil                             | 2017                                                                                                                                                                                                                                                                                              |
| Année du dernier recueil                             | 2025                                                                                                                                                                                                                                                                                              |
| Taille de la base de données                         |                                                                                                                                                                                                                                                                                                   |
| Taille de la base de données (en nombre d'individus) | [500-1000[ individuals                                                                                                                                                                                                                                                                            |
| Détail du nombre d'individus                         | 642                                                                                                                                                                                                                                                                                               |
| Données                                              |                                                                                                                                                                                                                                                                                                   |
| Activité de la base                                  | Current data collection                                                                                                                                                                                                                                                                           |
| Type de données recueillies                          | Clinical data<br>Declarative data<br>Paraclinical data<br>Biological data                                                                                                                                                                                                                         |
| Détail des données cliniques recueillies             | 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.

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| Données déclaratives, précisions                                                 | Paper self-questionnaire<br>Internet self-questionnaire<br>Face to face interview                                                               |
| Détail des données déclaratives recueillies                                      | SF-36 (adults) / SF-10 (children)                                                                                                               |
| Données biologiques, précisions                                                  | Biochemical, hematological, and renal parameters (ESRD, dialysis...)                                                                            |
| Existence d'une biothèque                                                        | Yes                                                                                                                                             |
| Contenu de la biothèque                                                          | Fluids (saliva, urine, amniotic fluid, ?)                                                                                                       |
| Détail des éléments conservés                                                    | urine                                                                                                                                           |
| Paramètres de santé étudiés                                                      | Health event/morbidity<br>Health event/mortality<br>Quality of life/health perception                                                           |
| Modalités                                                                        |                                                                                                                                                 |
| Mode de recueil des données                                                      | eCRF in secure web access, secure cloud and HADS hosting                                                                                        |
| Procédures qualité utilisées                                                     | Data Management Plan and Data Validation Plan. Continuous data management (automatic control rules and "queries" system).                       |
| Suivi des participants                                                           | Yes                                                                                                                                             |
| Modalités de suivi des participants                                              | Monitoring by convocation of the participant<br>Monitoring by contact with the referring doctor                                                 |
| Appariement avec des sources administratives                                     | No                                                                                                                                              |
| Valorisation et accès                                                            |                                                                                                                                                 |
| Valorisation et accès                                                            |                                                                                                                                                 |
| Accès                                                                            |                                                                                                                                                 |
| Existence d'un document qui répertorie les variables et les modalités de codage  | Yes                                                                                                                                             |
| Charte d'accès aux données (convention de mise à disposition, format de données) | Requests for access to RaDiCo-EURBIO data (aggregated or individual) will be reviewed by the scientific committee following the submission of a |

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| et délais de mise à disposition) | synopsis of a Specific Research Project (PRS), as defined in the Access to Resources Charter. Requests should be sent to <a href="mailto:eurbio@radico.fr">eurbio@radico.fr</a> . |
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| Accès aux données agrégées | Access on specific project only |
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| Accès aux données individuelles | Access on specific project only |
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