- COHORTE DE PATIENTS ATTEINTS DE SYNDROME D'ALPORT

Mise à jour : 04/03/2015

Responsable(s) :
Heidet Laurence

Organisme(s) responsable(s) :
Centre de référence Maladies Rénales Héréditaires de l'Enfant et de l'Adulte - Société française de néphrologie

Type de base de données

Bases de données issues d'enquêtes

Base de données issues d'enquêtes, précisions

Etudes de cohortes

Objectif principal

Connaître l'histoire naturelle de la maladie.
Comprendre les mécanismes de la progression de la maladie vers l'insuffisance rénale.

Critères d'inclusion

Patients atteints de syndrome d'Alport

RADICO-SEDVASC - COHORTE NATIONALE (FR) SUR LE SYNDROME D'EHLERS DANLOS VASCULAIRE

Mise à jour : 03/01/2017

Responsable(s) :
JEUNEMAITRE Xavier, Inserm UMR S 970

Organisme(s) responsable(s) :
Inserm

Type de base de données

Registres de morbidité

Objectif principal

- l'histoire naturelle de la maladie et en particulier l'ordre d'apparition et les liens entre les différents signes majeurs et mineurs du syndrome (artériels, digestifs, pulmonaires et utérins)

Objectifs Secondaires
- les relations génotype- phénotype du syndrome
- les relations phénotypiques intrafamiliales
- les effets à long terme du Celiprolol seul ou en association avec d'autres molécules
- le coût médico-économique de la maladie
- les répercussions de la maladie sur la qualité de vie des patients et leurs activités socio-professionnelles

(auto-questionnaire généraliste SF36 et échelle d'anxiété d'Hamilton)

Critères d'inclusion

Les patients concernés sont ceux (enfants et adultes) souffrant du syndrome d'Ehlers-Danlos vasculaire caractérisé sur le plan moléculaire (mutation du gène COL3A1), ayant signé un formulaire de consentement éclairé et suivis dans l'un des centres investigateurs participant à l'étude (toute la France étant couverte)

RADICO-ACOEIL - NATIONAL COHORT ON CONGENITAL DEFECTS OF THE EYE: NATURAL HISTORY, GENETIC DETERMINISMS AND IMPROVED OCULAR AND EXTRA-OCULAR OUTCOME PREDICTION FOR BETTER PATIENT MANAGEMENT

Mise à jour : 03/01/2017

Responsable(s) :
Chassaing Nicolas , Inserm U 1056
Calvas Patrick

Organisme(s) responsable(s) :
Institut National de la Santé et de la Recherche Médicale / French National Institute for Health and Medical Research (Inserm)

Type de base de données

Morbidity registers

Objectif principal

Main objective
The principal objective of this study is to delineate the long term outcomes of the patients with ocular developmental defects, focusing on visual and neuro-developmental issues.

Secondary objectives
I) Identification of prognostic factors (such as ocular defects, unilateral or bilateral involvement, extra-ocular malformations) that would be associated with unfavourable visual and/or neurologic outcome. These data will be essential for the formulation of recommendations to enhance diagnosis and patient management.
II) Repercussions of the ocular developmental defects on patients and families quality of life.

Exploratory objectives
Searching for potential genotype/phenotype correlations to unravel
- the frequency of implication of each gene in these ocular developmental defects;
- the phenotypic spectrum associated with mutations in these genes;
- the identification of novel genes involved in these ocular developmental defects.
Given genotyping will not be mandatory to participate to the cohort; this objective will involve only the patients who accepted it.

Critères d'inclusion

Patients from 0 to 7 years old
- Newborns and/or children from birth to 7 years old, affected with the following ocular defects:

? anophthalmia,
? microphthalmia
? aniridia
? anterior segment dysgnesis
whose parents will have properly evaluated risks (those related to the actual standard of care for these pathologies) and benefits (improvement of knowledge and standard of care) of the study, and will be given an informed consent to participate the protocol.
- Patients affiliated to the "Régime National d'Assurance Maladie"
- Inclusion of foreign patients will be possible through the French inclusion centers when they agreed to be charged for all medical fees.
Patients over 8 years old
- Children from 8 years old, affected with the following ocular defects :
? anophthalmia,
? microphthalmia
? aniridia
? anterior segment dysgenesis
whose parents will have properly evaluated risks and benefits of the study, and will be given an informed consent form to participate to the protocol.
- Patients affiliated to the "Régime National d'Assurance Maladie"
- Inclusion of foreign patients will be possible through the French inclusion centres when they agreed to be charged for all medical fees.
Adult Patients
- Adults affected with the following ocular defects :
? anophthalmia,
? microphthalmia
? aniridia
? anterior segment dysgenesis
- Adult patients under guardianship whose guardians will have properly evaluated risks (those related to the actual standard of care for these pathologies) and benefits (improvement of knowledge and standard of care) of the study, and will be given an informed consent to participate the protocol. Indeed, intellectual disability may be associated with the ocular defects and we will need to include these patients in order to evaluate incidence of this event.
- Adult patients able to properly evaluate risks (those related to the actual standard of care for these pathologies) and benefits (improvement of knowledge and standard of care) of the study and to give their informed consent to participate to the protocol.
- Adult parents of an affected child participating to the study and willing to participate to the inheritance study (results of DNA analysis).
- Patients affiliated to the "Régime National d'Assurance Maladie".
- Inclusion of foreign patients will be possible through the French inclusion centres when they agreed to be charged for all medical fees.
- Pregnant women can be included in the study (as examination proposed have no interference or adverse effect during pregnancies).

Non-inclusion Criteria
- Patients with ocular developmental defects other than the ones listed above.
- Patient or patients' parents/tutor not able to approve or declining participation to the protocol.
- French patients not affiliated to the "Régime National d'Assurance Maladie" or foreign patients not willing to pay charges of medical services.

RADICO-ECYSCO - EUROPEAN CYSTINOSIS COHORT

Mise à jour : 03/01/2017

Responsable(s) :
SERVAIS Aude, Inserm U983
Niaudet Patrick

Organisme(s) responsable(s) :
Institut National de la Santé et de la Recherche Médicale / French National Institute for Health and Medical Research (Inserm)

Type de base de données

Morbidity registers

Objectif principal

The primary objective of the RaDiCo-ECYSCO cohort is to understand the natural history and major long-term manifestations and outcomes of cystinosis in paediatric and adult cases.

Secondary Objectives are to:
? Evaluate the impact of disease and treatments on patients' quality of life
? Evaluate the effect of treatment on the complications
? appraise the long-term safety of treatment and compliance

Information Technology Objectives are to:
? Develop and diffuse an electronic tool of data collection from various sources linked to a database integrating a system of management and follow-up of data-management allowing collection of data for cystinosis paediatric and adult patients.
? Include data generated by patients and, where relevant, their parents and or carers.
? Expand the cohort to cover a broader European population.
? Promote the use of the RaDiCo-ECYSCO eCRF for mutualisation and harmonisation of data for cystinosis paediatric and adult patients within the expert sites.

Improvement of standard care objectives are to:
? Develop comprehensive evidence based guidelines for treatments as well as for follow-up of patients who will switch from paediatric to adult status,
? Propose a system of audit against the guidelines ensuring overall care is of the highest standard as well as identifying areas of concern for actions.

Critères d'inclusion

The RaDiCo-ECYSCO Cohort inclusion criteria are the following:
? Confirmed diagnosis of cystinosis (based on cystine dosage, presence of crystals at eye examination or molecular diagnosis)
? Signed informed consent

Non-inclusion Criteria
? Patients not able to give their informed consent.
No other non-inclusion criteria (patients with associated disease should be enrolled)

RADICO-DCP - DYSKINÉSIES CILIAIRES PRIMITIVES : IDENTIFICATION DE CRITÈRES DE SÉVÉRITÉ SPÉCIFIQUES ET RECHERCHE DE CORRÉLATION GÉNOTYPE-PHÉNOTYPE

Mise à jour : 03/01/2017

Responsable(s) :
ESCUDIER Estelle, Inserm UMR S 933

Organisme(s) responsable(s) :
Institut National de la Santé et de la Recherche Médicale / French National Institute for Health and Medical Research (Inserm)

Type de base de données

Registres de morbidité

Objectif principal

Objectif principal
L'objectif principal est d'identifier dans une grande cohorte de patients DCP, des facteurs prédictifs précoces de sévérité afin d'améliorer la prise en charge personnalisée des patients.

Objectifs secondaires
Les objectifs secondaires visent à :
? Valider l'implication de nouveaux gènes de DCP
? Evaluer l'impact sur la qualité de vie de deux atteintes importantes de la DCP :
o L'atteinte ORL selon sa sévérité (en particulier chez l'enfant, surdité avec retard de langage, et chez l'adulte, intensité des rhinosinusites chroniques avec recours à la chirurgie),
o L'infertilité de l'adulte qui nécessite souvent le recours à l'assistance médicale à la procréation, tout particulièrement chez les patients masculins.

Objectifs exploratoires
? Evaluer l'impact sur la qualité de vie des autres atteintes de la DCP
? Rechercher des corrélations entre les différents aspects phénotypiques cliniques (en particulier les facteurs prédictifs de sévérité identifiés, les manifestations ORL perturbant la qualité de vie, l'existence d'une infertilité), le phénotype ciliaire, et le génotype.

Objectifs informatiques
? Développer et diffuser un outil électronique de recueil de données de diverses sources. Cet outil sera adossé à une base de données et intégrera un système de gestion de suivi du data-management. Il permettra la saisie des données pédiatriques et adultes chez les patients atteints de DCP.
? Promouvoir l'utilisation de cet outil auprès des Centres de Compétence et de Référence Maladies Rares dédiés à la DCP et harmoniser le recueil des données, pour la première fois en France, des patients adultes et pédiatriques.

Critères d'inclusion

Tous les patients prévalents et incidents inclus dans la cohorte RaDiCo-DCP doivent :
? Avoir un diagnostic confirmé de DCP basé sur au moins un des critères de diagnostic objectif suivant : syndrome de Kartagener (association de sinusites chroniques, de bronchiectasie et d'un situs inversus), et/ou mise en évidence d'anomalies spécifiques de l'ultrastructure ciliaire, et/ou identification de mutations non ambiguës dans un gène de DCP.
? Avoir au minimum une visite annuelle de suivi conformément à la pratique courante.

Critères de non inclusion
Les patients correspondants aux critères suivants, ne pourront être inclus :

? Patient avec un diagnostic de DCP non confirmé (ne répondant pas aux critères du paragraphe 3.2).
? Patient avec une pathologie concomitante évolutive pouvant interférer avec l'évaluation des manifestations liées à la DCP.

RADICO-PID - IDIOPATHIC INTERSTITIAL PNEUMONIA: FROM INFANCY TO ELDERLY

Mise à jour : 03/01/2017

Responsable(s) :
CLEMENT Annick, Inserm UMR S 933

Organisme(s) responsable(s) :
Institut National de la Santé et de la Recherche Médicale / French National Institute for Health and Medical Research (Inserm)

Type de base de données

Morbidity registers

Objectif principal

Primary Objective
The main objective is to describe the phenotypic features of the paediatric and adult patients with IIP/PID, at diagnosis and during the follow-up. These data will be critical for the description of the natural history of the various forms of IIP/PID.

Secondary Objectives
The secondary objectives are to:
? Identify gene factors involved in disease initiation and progression,
? Investigate the extent to which environmental and co-morbidity factors may influence disease severity and outcome
? Identify and validate biomarkers for disease diagnosis and progression

Exploratory objectives
? Production of improved strategies for patient recruitment and enrolment into clinical trials
? Development of novel strategy for patient follow-up
? Development of novel diagnostic approaches
? Evaluation of effect on natural history of disease, and tolerance of therapy, in a large population in real life
? Development of novel therapeutic approaches

Information Technology Objectives
? Develop and diffuse an electronic tool of data collection from various sources linked to a database integrating a system of management and follow-up of data-management allowing collection of data for IIP/PID paediatric and adult patients.
? Include data generated by patients and, where relevant, their parents and/or carers.

Critères d'inclusion

? Patient with a diagnosis of IIP/PID
IIP/PID diagnosis is established on presenting history, clinical, radiological and functional and if available pathological findings. Inclusion criteria include:

? Clinical criteria: chronic respiratory insufficiency manifestations including dyspnea/tachypnea, cough, and cyanosis during exercise or at rest
? Radiological criteria: characteristic chest High-Resolution Computed Tomography (HRCT) abnormalities including widespread ground glass or alveolar attenuation, reticulation often associated with traction bronchiectasis, and honeycombing
? Functional criteria: pulmonary function test abnormalities reflecting a restrictive pattern and including: loss of lung volume, vital capacity (VC), total lung capacity (TLC); reduction in the diffusion capacity of the lung for carbon monoxide (DLCO), gas exchange abnormalities, and altered ventilatory response to exercise
? Patients (parents/guardians for paediatric/patients) having given an informed consent to participate in the protocol
? Patients affiliated to the ?Regime National d'Assurance Maladie?

Non-inclusion Criteria
? Patients with diffuse parenchymal lung diseases caused by drug toxicity, immunodeficiency, proliferative disorders including histiocytosis, and metabolic disorders
? Patients (parents/guardians for paediatric patient) not able to approve/understand the protocol

RADICO-IDMET - NATIONAL COHORT ON IMPRINTING DISORDERS AND THEIR METABOLIC CONSEQUENCES

Mise à jour : 03/01/2017

Responsable(s) :
LINGLART Agnes, Inserm U 1169
NETCHINE Irène, INSERM U938, équipe 4

Organisme(s) responsable(s) :
Institut National de la Santé et de la Recherche Médicale / French National Institute for Health and Medical Research (Inserm)

Type de base de données

Morbidity registers

Objectif principal

Main objective
The main objective of this study is to describe the natural history of imprinting disorders (IDs) according to their metabolic profile.

Secondary objectives
Secondary objectives are:
? Evaluate the correlation between phenotypes and metabolic profiles at the time of diagnosis.
? Evaluate the risk factor of the various metabolic profiles
? Identify common therapeutic approaches for all IDs (this might lead to the identification of extended applications to all IDs or a larger group of IDs for drugs with so far restricted Marketing Authorization (MA).
? Assess the impact of IDs on quality of life
? Analyse inheritance data of the diseases (search for transmission of (epi)genetic mutations in parents of probands).

Exploratory objectives
? To evaluate the feasibility to use metabolic profiles for clinical classification of IDs
? To develop comprehensive, evidence based guidelines for diagnostic, treatments as well as for follow-up of patients
? To establish a homogenous group of French IDs patients in order to improve knowledge and medical management of IDs.
? To explore the correlation between microbiotia and metabolic profiles in IDs.
? To explore the possibility of using a therapeutic approach already in use for one ID also for other IDs

Information Technology Objectives
? Develop and diffuse an electronic tool of data collection from various sources linked to a database integrating a system of management and follow-up of data-management allowing collection of data for IDs patients.
? Include data generated by patients and, where relevant, their parents and/or carers.

Critères d'inclusion

Inclusion period will last 5 years.
Patients (adults and children) affected with an ID regardless of the severity of the disease,
- with a confirmed diagnosis of ID (based on molecular diagnosis)
- with a signed informed consent for adults or signed informed consent of parents/guardians of minors/ protected adult.

There are no non-inclusion criteria