ImmunAID project: for a better diagnosis and management of autoinflammatory diseases
Project governance

Project Management Team

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Steering Committee

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- Bruno Fautrel
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- Hélène Esperou
  WP9 leader (Inserm)
General context and current challenges in Systemic AutoInflammatory Diseases (SAID)

- A complex and largely unknown set of rare diseases with unspecific symptoms (fever, rash, etc.)
- Very heterogeneous disease presentation within 2 main categories:
  - Monogenic familial disorders (mSAID)
  - Phenotype-based SAIDs without known mutation (guSAID):
    - Recurrent Pericarditis
    - Adult-onset Still’s disease or systemic juvenile idiopathic arthritis
    - Neutrophilic dermatosis
    - Schnitzler
    - Vasculitis (Kawasaki, Behçet, Takayasu arteritis)
    - Inflammation of unknown origin
    - Chronic/recurrent osteitis

- Unmet medical needs
  - Diagnosis is difficult: based on clinical signs and exclusion of other diseases
  - Diagnosis is long: up to 5 inappropriate treatments can be prescribed before the right diagnosis is made

- Open biological questions
  - Autoinflammation signatures related to SAID?
  - What is the dysregulation of inflammasome functions?
  - What is the cytokine network disequilibrium?
Project objectives

- **Clinical objectives:**
  - To deliver a new, comprehensive and pathogenesis-driven classification of SAID
  - To open the way to a rapid and accurate diagnosis across all the spectrum of SAID
  - To improve patient management through adapted guidelines with more relevant treatment options

- **Biological objectives:**
  - To identify and validate novel Omics- and pathway-based diagnostic biomarkers: genetic mutations (or combinations of genetic mutations), translational dysregulation, proteomics disequilibrium or microbiome alteration associated with SAID
  - To describe the potential dysregulation of inflammasome functions by studying known or suspected regulators and monitoring their functions in response to external stimuli
  - To assess the role of cytokine network disequilibrium in the expression of SAIDs
  - To explore inflammation resolution processes by measuring the activity of inflammation resolution effectors/regulators
Scientific strategy

1) New diagnostic biomarkers
2) Disentangle the spectrum of SAID
3) New omics- and pathogenesis-based SAID classifications
4) Robust clinical decision algorithm

European-wide cohort

Exhaustive biological analyses

Data integration and high-throughput analyses

Outputs

Unbiased strategy

Multi-omics analyses

Inflammasome
Lipid mediators
Immune networks

Targeted strategy

Samples from a cohort of 1616 adults and paediatrics patients

Data integration and high-throughput analyses

Outputs

1) New diagnostic biomarkers
2) Disentangle the spectrum of SAID
3) New omics- and pathogenesis-based SAID classifications
4) Robust clinical decision algorithm
Workplan overview

WP1 Clinical cohort of SAID

WP2 Multi-omics for data generation

WP3 Inflammasome-associated pathways in SAID

WP4 Inflammation resolution

WP5 Immune networks

WP6 Data analysis and integration

WP7 Dissemination, communication and valorization

WP9 Ethics
The ImmunAID consortium

24 Institutions from 12 European countries

€15.8 million funded by the Horizon 2020 program

37 clinical centers in 11 countries

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End date: April 30th, 2023