ImmunAID
Immunome Consortium for AutoInflammatory Disorders

ImmunAID project:

for a better diagnosis and management of autoinflammatory diseases

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Project governance

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WP7 leader
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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?

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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

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Scientific strategy

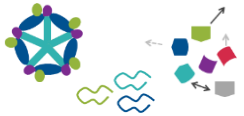
European-wide cohort

Samples from a cohort of 1616 adults and paediatrics patients

Exhaustive biological analyses

Unbiased strategy

Multi-omics analyses



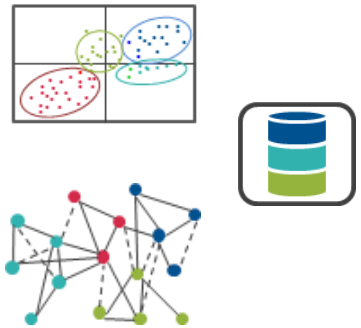
Inflammasome

Lipid mediators

Immune networks

Targeted strategy

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

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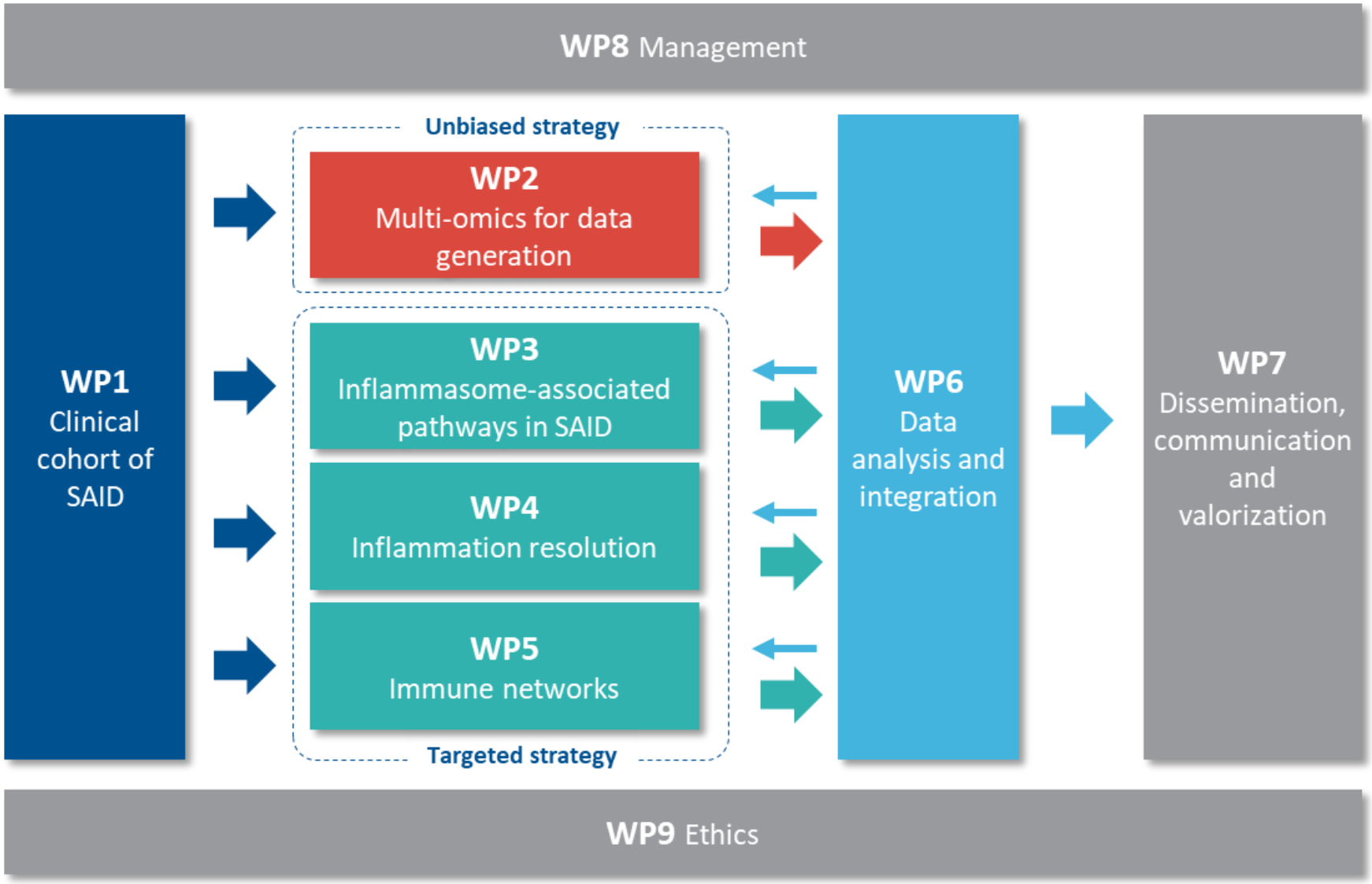
5

Workplan overview



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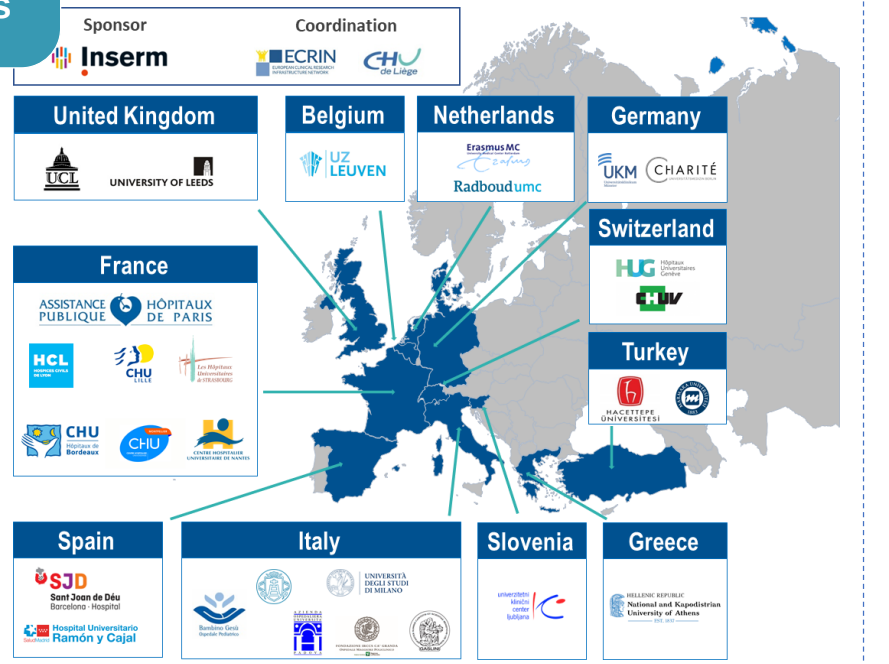
The ImmunAID consortium

24
Institutions from 12 European countries

37
clinical centers in 11 countries



€15.8 million
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