

ISSAID Programme Day by Day | **Tuesday 8 April 2025**

08:30 - 10:00

Espace Adenauer

Opening ceremony

Moderated by Isabelle Koné-Paut (FR) & Erkan Demirkaya (CA)

08:30 President's address / [Erkan Demirkaya](#) (CA)

08:35 Welcome from local chair / [Isabelle Koné-Paut](#) (FR)

08:40 Surprise from CEREMAIA

09:00 Evolution, genetics of populations / [Lluís Quintana-Murci](#) (FR)

09:40 Announcement of award winners / [Isabelle Koné-Paut](#) (FR) & [Erkan Demirkaya](#) (CA)

10:00 - 10:30

Salon Honnorat

Coffee break & poster viewing

10:30 - 11:30

Espace Adenauer

Pathways of autoinflammation

10:30 JAK associated inflammatory disease / [Dusan Bogunovic](#) (US)

10:50 Regulations of NLRP3 inflammasomes / [Seth Masters](#) (AU)

11:10 ADA2 deficiency in 2025 / [Pui Lee](#) (US)

11:30 - 12:30

Espace Adenauer

Mechanisms of inflammasome activation

11:30 Modulization of missense variants / [Alessandra Carbone](#) (FR)

11:50 Functional tests of MEFV variants / [Thomas Henry](#) (FR)

12:10 The NLRP1 inflammasome / [Etienne Meunier](#) (FR)

12:30 - 13:30

Salon Gulbenkian

Outcome measures SAID meeting

12:30 - 13:30

Salon Honnorat

Break & Poster Walks 1 - 5

13:30 - 15:00

Espace Adenauer

Epidemiology/vasculitis of SAID

13:30 Extracellular vesicles, new protagonists in thrombo-inflammatory diseases / [Françoise Dignat-George](#) (FR)

13:50 SAID and vasculitis / [Paul Brogan](#) (UK)

14:10 Genetic mimickers / [Charalampia Papadopoulou](#) (UK)

14:30 SYSTEMIC INFLAMMATION, LYMPHOPROLIFERATION AND VASCULOPATHY IN A PATIENT WITH ARHGAP10 MUTATION / [Stefano Volpi](#) (IT)

14:40 BIOLOGICS IN THE TREATMENT OF PEDIATRIC BEHÇET'S DISEASE: RESULTS OF AN INTERNATIONAL COLLABORATIVE STUDY BY THE PRES VASCULITIS WORKING PARTY / [Fatma Gül Demirkan](#) (TR)

13:30 - 15:00

Salon Gulbenkian

Molecular determinants of innate immunity

13:30 The mechanism of cell autonomous inflammation in VEXAS syndrome is mediated by proteotoxic stress / [David Beck](#) (US)

13:50 Genetic Architecture of Incomplete Penetrance / [Dusan Bogunovic](#) (US)

14:10 Systemic autoinflammation due to gain of function mutations in STAT4 / [Lori Broderick](#) (US)

14:30 COFILIN-1 IS A REDOX SENSOR REGULATING THE NLRP3 INFLAMMASOME / [Jae Jin Chae](#) (US)

14:40 SGT1 CONTROLS NLRC4 INFLAMMASOME ACTIVATION IN AUTO-INFLAMMATORY DISEASES / [Cyrielle Hou](#) (CH)

14:50 GSDMD AND GSDME AMPLIFY NLRP3 ACTIVATION IN AUTOINFLAMMATION / [Janet Onyuru](#) (US)

15:00 - 16:00

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Year in review & award presentations

15:00 Basic science / [Ivona Aksentijevich](#) (US)

15:20 Clinical science / [Erkan Demirkaya](#) (CA)

15:40 Dan Kastner award presentations / [Alice Burleigh](#) (UK) & [Marcia Munoz](#) (AU)

16:00 - 16:30

Salon Honnorat

Coffee break & poster viewing

16:30 - 17:30

Espace Adenauer

SAIDs across the ages

16:30 Late complications of Familial Mediterranean Fever / [Sophie Georjin Lavalie](#) (FR)

16:50 SAIDs and pregnancy / [Huri Ozdogan](#) (TR)

17:10 Does everybody need life-long treatment? / [Helen Lachmann](#) (UK) & [Seza Ozen](#) (TR)

16:30 - 17:30

Salon Gulbenkian

ISSAID general assembly

17:30 - 19:00

Salon Honnorat

Welcome reception

08:30 - 10:00

Espace Adenauer

Contribution of algorithms and artificial intelligence in SAIDs

- 08:30 Contribution of AI to the diagnosis of SAIDs / [Kevin Yauy](#) (FR)
08:50 AI approaches to decipher the pathogenesis of SAIDs / [Vassili Soumelis](#) (FR)
09:10 PROTEIN ARRAY PROFILING IDENTIFIES DISTINCT AUTO ANTIBODY SIGNATURES IN SYSTEMIC AUTO INFLAMMATORY DISEASES / [Gabriel Alexander Vignolle](#) (AT)
09:20 VALIDATION OF HUMAN PHENOTYPE ONTOLOGY (HPO) TERMS AND DEVELOPMENT OF AN AI-BASED DIAGNOSTIC TOOL FOR SAIDS USING THE EUROFEVER REGISTRY: THE ODINO PROJECT / [Caterina Matucci Cerinic](#) (IT)
09:30 ASSESSING THE IMPACT OF FAMILIAL MEDITERRANEAN FEVER (FMF) ON PHYSICAL ACTIVITY IN CHILDREN USING THE PHYSICAL ACTIVITY QUESTIONNAIRE FOR CHILDREN (PAQ-C): A COMPARATIVE PRELIMINARY STUDY WITH HEALTHY CONTROLS / [Zelal Aydin](#) (TR)
09:40 INCORPORATION OF RECENT SELECTION SIGNALS IMPROVES VARIANT IMPACT PREDICTION IN IMMUNE-MEDIATED GENES / [Brynja Matthiasardottir](#) (US)
09:50 IMPROVED MACHINE LEARNING MODELS FOR PREDICTING COLCHICINE RESISTANCE IN FAMILIAL MEDITERRANEAN FEVER / [Admir Ozturk](#) (TR)

10:00 - 10:30

Salon Honnorat

Coffee break & poster viewing

10:30 - 11:30

Espace Adenauer

Neurologic inflammation and SAIDs

- 10:30 Neuroinflammation and SAIDs / [Pierre Ellul](#) (FR)
10:50 Functional MRI studies in neuroinflammation / [Neil Basu](#) (UK)
11:10 DIAGNOSIS OF CRYOPYRIN-ASSOCIATED PERIODIC SYNDROME (CAPS) IN ADULTHOOD: LESSONS FROM A FRENCH COHORT / [Yixiang Yves-Jean Zhu](#) (FR)
11:20 NEUTROPHIL EXTRAVASATION AND BBB DISRUPTION IN MURINE NOMID: INSIGHTS INTO NEUROINFLAMMATION / [Hal Hoffman](#) (US)

11:30 - 12:30

Espace Adenauer

Genetics

- 11:30 Whole genome sequencing and neonatal screening: The GUARDIAN experience / [Alban Ziegler](#) (FR)
11:50 Which techniques for the diagnosis of SAIDs in 2025 / [Alexander Hoishen](#) (NL)
12:10 IRAK2 DEFICIENCY CAUSES A NEW IMMUNE DYSREGULATION DISORDER / [Yudie Fei](#) (CN)
12:20 CHANGING THE LANDSCAPE OF ACQUIRED SAIDs - REPORT FROM THE UK REFERENCE GENETIC LABORATORY / [Dorota Rowczenio](#) (UK)

11:30 - 12:30

Salon Gulbenkian

Amyloidosis and renal complications in SAIDs

Moderated by Gilles Grateau (FR) & Ilan Ben-Zvi (IL)

- 11:30 Overview on AA amyloidosis worldwide in 2025 / [Helen Lachmann](#) (UK)
11:50 State of the art of AA amyloidosis complicating familial Mediterranean fever in the world in the last 50 years / [Ahmet Gul](#) (TR)
12:10 AA amyloidosis complicating monogenic autoinflammatory diseases / [Sophie Georgin Lavialle](#) (FR)

12:30 - 13:30

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Lunch sponsored symposium – supported by Sobi

13:30 - 15:00

Espace Adenauer

New clinical lessons in autoinflammation

- 13:30 Still's Disease (Results from the Immunaid) / [Bruno Fautrel](#) (FR)
- 13:50 When, why and how to monitor IL-18? / [Scott Canna](#) (US)
- 14:10 Inflammation impacts the HSC compartment: Consequences for gene therapy approaches / [Marina Cavazzana](#) (FR)
- 14:30 RARE TNFAIP3 HYPOMORPHIC VARIANTS ARE A MASSIVELY UNDERESTIMATED DRIVER OF HUMAN AUTOINFLAMMATORY DISEASE GLOBALLY / [Daniella Schwartz](#) (US)
- 14:40 JAK INHIBITORS ARE EFFECTIVE IN PEDIATRIC REFRACTORY NLRC4 GAIN OF FUNCTION MUTATION / [Jean Jacques De Bruycker](#) (CA)
- 14:50 DOMINANT NEGATIVE ADA2 MUTATIONS CAUSE ADA2 DEFICIENCY IN HETEROZYGOUS CARRIERS / [Marjon Wouters](#) (BE)

13:30 - 15:00

Salon Gulbenkian

Oral communications 1

- 13:30 PULMONARY ARTERIAL HYPERTENSION WITH STILL'S DISEASE: A NEW PULMONARY MANIFESTATION ASSOCIATED WITH HLA-DRB1*15 / [Stéphane Mitrovic](#) (FR)
- 13:40 FUNCTIONAL ANALYSIS OF C- AND N-TERMINAL CDC42 VARIANTS REVEALS DISTINCT PATHWAYS OF AUTOINFLAMMATION RESPONSIBLE FOR DIFFERENT CDC42-ASSOCIATED AUTOINFLAMMATORY DISEASES? / [Philippe Mertz](#) (FR)
- 13:50 CURRENT TREATMENT OF MACROPHAGE ACTIVATION SYNDROME WORLDWIDE: THE METAPHOR PROJECT, A PRES/PRINTO REAL-LIFE INTERNATIONAL SURVEY / [Francesca Minoia](#) (IT)
- 14:00 MANAGING PATIENTS WITH CRYOPYRIN ASSOCIATED PERIODIC SYNDROME (CAPS). HOW DOES INITIATING TREATMENT WITH IL-1 MEDICATIONS AFFECT PATIENT'S SYSTEMIC INFLAMMATION, SYMPTOM REPORTING AND QUALITY OF LIFE. EXPERIENCE OF A SPECIALISED UK CENTRE / [Michael Wood](#) (UK)
- 14:10 PROGRESS REPORT ON IL6 INHIBITION IN ROSAH AUTOINFLAMMATORY DISEASE / [Christina Kozycki](#) (US)
- 14:20 PROSPECTIVE FOLLOW UP OF 37 PREGNANCIES IN WOMEN RECEIVING IL1 INHIBITORS FOR SYSTEMIC AUTOINFLAMMATORY DISEASES: AN MULTICENTRIC FRENCH STUDY FROM THE GR2 COHORT / [Marion Delplanque](#) (FR)
- 14:30 Q&A

15:00 - 16:00

Espace Adenauer

Thieves market

- 15:00 Case 1 / [Adrien Schvartz](#) (FR)
- 15:10 Case 2 / [Toni Hospach](#) (DE)
- 15:20 Case 3 / [Archana Khan](#) (IN)
- 15:30 Case 4 / [Anna Agrusti](#) (IT)
- 15:40 Case 5 / [Federica Anselmi](#) (FR)
- 15:50 Q&A

15:00 - 16:00

Salon Gulbenkian

Workshop: Autoinflammation and osteitis - Part I

Moderated by Polly Ferguson (US) & Perrine Dusser (FR)

- 15:00 CRMO pathophysiology, diagnosis / [Christian Hedrich](#) (UK)
- 15:20 CRMO- associated diseases / [Herman Girschick](#) (DE)
- 15:40 OGFRL1 GENE MUTATIONS MAY LINK CHERUBISM TO CHRONIC RECURRENT MULTIFOCAL OSTEOMYELITIS (CRMO) / [Linda Rossi-Semerano](#) (FR)
- 15:50 EFFICACY AND TOLERABILITY OF BISPHOSPHONATES IN THE MANAGEMENT OF CHRONIC RECURRENT MULTIFOCAL OSTEOMYELITIS (CRMO) IN CHILDREN: A 30-PATIENT RETROSPECTIVE COHORT STUDY / [Perrine Dusser](#) (FR)

16:00 - 16:30

Salon Honnorat

Coffee break & poster viewing

16:30 - 18:00

Espace Adenauer

Navigating autoinflammation through generations and resources

- 16:30 Summary of Early Career Group meeting / [Fatma Dedeoglu](#) (CA)
- 16:45 Autoinflammatory Diseases in Emerging Countries: Challenges & Perspectives / [Rim Bourguiba](#) (TN)
- 17:00 2nd talk / [Marie-Louise Freymond](#) (FR)
- 17:15 3rd talk / [Raphaela Goldbach-Mansky](#) (US)
- 17:30 Panel discussion and Q&A

16:30 - 17:30

Salon Gulbenkian

Workshop: Autoinflammation and osteitis - Part II

Moderated by Polly Ferguson (US) & Perrine Dusser (FR)

- 16:30 Monogenic auto-inflammatory disease with bone osteitis / [Polly Ferguson](#) (US)
- 16:50 Radiologic findings of CRMO / [Sandrine Chilloh](#) (FR)
- 17:10 Debate: pro anti-TNF in CRMO VS pro Bisphosphonates in CRMO? / [Anja Schnabel](#) (DE) & [Perrine Dusser](#) (FR)

08:30 - 10:00

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Beyond genetics of SAIDs

- 08:30 Conceptualization of immune memory and trained immunity / Speaker TBC
- 08:50 Epigenetic determinants in inflammasomopathies / Florence Apparilly (FR)
- 09:10 Cholesterol metabolism and inflammation / Sam Lockhart (UK)
- 09:30 TRANSCRIPTOMIC INSIGHTS INTO PFAPA SYNDROME: RNA-SEQUENCING ANALYSIS OF FLARE RELATIVE TO NON-FLARE STATES / Sivia Kerry Lapidus (US)
- 09:40 STRUCTURE AND FUNCTION OF PYRIN INFLAMMASOME: MECHANISTIC LINK BETWEEN FMF AND NOCARH SYNDROME / Wonyong Lee (KR)
- 09:50 MEVALONATE KINASE DEFICIENCY – AN AUTOINFLAMMATORY DISEASE OF DYSREGULATED NK CELLS / Michael Rogers (AU)

10:00 - 10:30

Salon Honorat

Coffee break & poster viewing

10:30 - 11:30

Espace Adenauer

Clonal haematopoiesis and autoinflammation I

- 10:30 Clonal haematopoiesis and SAIDs / Jérôme Hadjadj (FR)
- 10:50 VEXAS syndrome in 2025 / David Beck (US)
- 11:10 INFLAMMATORY AND DYSIMMUNE MANIFESTATIONS IN T/NK-CELL TYPE CHRONIC ACTIVE EBV INFECTION: A DESCRIPTION OF 14 CASES OF A RARE AND HETEROGENEOUS CLONAL LYMPHOID HEMATOLOGICAL DISORDER / Benjamin Fournier (FR)
- 11:20 EMERGING TREATMENT STRATEGIES FOR VEXAS SYNDROME: A SYSTEMATIC REVIEW AND META-ANALYSIS / Berkay Kilic (TR)

10:30 - 11:30

Salon Gulbenkian

Biomarkers & outcome measures in SAIDs

- 10:30 Neutrophil activation markers in autoinflammation / Dirk Foell (DE)
- 10:50 Quality of life in SAIDs / Maryam Piram (CA)
- 11:10 ROLE OF IL-18 AS A BIOMARKER IN MONITORING PEDIATRIC PATIENTS WITH STILL'S DISEASE / Matteo Trevisan (IT)
- 11:20 EVALUATING SIGLEC-1 EXPRESSION ON MONOCYTES AS A DIAGNOSTIC BIOMARKER FOR TYPE I IFN-RELATED PEDIATRIC AUTOINFLAMMATORY DISEASES / Valentina Matteo (IT)

11:30 - 12:30

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Clonal haematopoiesis and autoinflammation II

- 11:30 Tri 8 associated autoinflammatory disorders / Kalpana Manthiram (US)
- 11:50 Somatic mosaicism and TLR8 / Bertrand Boisson (FR)
- 12:10 Clonal haematopoiesis and IMID's / Sinisa Savic (UK)

11:30 - 12:30

Salon Gulbenkian

Oral communications 2

- 11:30 DEEP PHENOTYPING IDENTIFIES INFLAMMATORY PATHWAYS ASSOCIATED WITH DISEASE ACTIVITY OF VEXAS SYNDROME / [Kana Higashitani](#) (JP)
- 11:40 IMPROVEMENT OF REFRACTORY STILL'S/SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS LUNG DISEASE IN 6/7 CHILDREN TREATED WITH A NOVEL, BI-SPECIFIC IL-1BETA/IL-18 NEUTRALIZING ANTIBODY / [Scott Canna](#) (US)
- 11:50 PHOSPHOMEVALONATE KINASE DEFICIENCY: UNCOVERING NEW DIMENSIONS OF THE DISEASE PHENOTYPE / [Ezgi Deniz Batu](#) (US)
- 12:00 CHARACTERISTICS AND PROBLEMS OF JAPANESE PATIENTS WITH UBA1 VARIANT-NEGATIVE VEXAS SYNDROME-LIKE AUTOINFLAMMATORY DISEASE / [Ayaka Maeda](#) (JP)
- 12:10 TARGETING THE DYSREGULATED TYPE I IFN RESPONSE IN ADENOSINE DEAMINASE 2 DEFICIENCY EFFECTIVELY MITIGATES INFLAMMATION VIA PATHWAY INHIBITION AND GENE THERAPY/ [Dimitri Bulté](#) (IT)
- 12:20 FIRST GLOBAL SERIES OF VEXAS SYNDROME IN WOMEN: A COMPARATIVE ANALYSIS OF 14 FEMALE AND 274 MALE CASES / [Rim Bourguiba](#) (TN)

12:30 - 13:30

Salon Gulbenkian

Outcome measures SAID meeting (CARRA study group)

12:30 - 13:30

Salon Honnorat

Break & Poster Walks 6 - 10

13:30 - 15:00

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New targets for the treatment of SAID

- 13:30 Marrow transplant and stem cell transplant for SAID / [Isabelle Meyts](#) (BE)
- 13:50 Which treatment for NFkB related diseases? / [Carine Wouters](#) (BE)
- 14:10 DADA2 / [Marco Gattorno](#) (IT)
- 14:30 Debate: On demand or continuous treatment with IL-1 inhibition? / [Veronique Hentgen](#) (FR) & [Ilan Ben-Zvi](#) (IL)

15:00 - 15:30

Salon Honnorat

Coffee break & poster viewing

15:30 - 16:30

Espace Adenauer

Equity and access to innovative therapeutics in rare diseases

- 15:30 Patients' odyssey / [Jean-Philippe Plancon](#) (FR)
- 15:45 European public policies and rare diseases / [Johanna Benesty](#) (FR)
- 16:00 India: Overcoming the challenges of awareness availability and access / [Raju Khubchandani](#) (IN)
- 16:15 Kenya: Overcoming the challenges of awareness availability and access / [Angela Migowa](#) (KE)

16:30 - 17:00

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Closing remarks

- 16:30 Closing remarks
- 16:45 Announcement of ISSAID 2027