



13th ISSAID Meeting

of The International Society of Systemic
Auto Inflammatory Diseases

Paris, France

8-10 April 2025

FINAL PROGRAMME

www.issaid.org/issaid2025



Réseau Rhumatismes Inflammatoires Pédiatriques en Île-de-France



Les pathologies prises en charge: Arthrite Juvénile Idiopathique, Connectivites (Lupus, DMJ, Sclérodermie...), Uvéite inflammatoire, Maladies Auto-Inflammatoires (PFAPA, FMF, MKD...), Sarcoïdose, Vascularite (Takayasu, PAN...), Kawasaki, OCMR, Behçet, etc

RESRIP

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Welcome & Committees

Welcome Message

Dear Colleagues,

We are delighted to invite you to the 13th Meeting of the International Society of Systemic Autoinflammatory Diseases (ISSAID), which will take place in Paris from April 8th to 10th, 2025, at the "Cité Internationale" - a magnificent haven of peace near many transport amenities where you will particularly enjoy the sumptuous Honorat lounge overlooking a lush green park.

April in Paris offers a delightful mix of mild weather, budding greenery, and a relaxed atmosphere, perfect for strolling along the Seine after a day of work.

The ISSAID 2025 meeting will provide an opportunity for pediatric and adult rheumatologists, immunologists, general pediatricians and internists, scientists, and others interested in systemic autoinflammatory diseases to come together to share new findings related to autoinflammation and to learn about recent advances in this field.

The ISSAID 2025 meeting will highlight the topic of neurologic inflammation, and we hope it will facilitate better assessment, care, and research on this particularly complex subject. We will present current topics such as artificial intelligence, mechanisms of inflammation, and new diseases.

The ISSAID 2025 meeting will promote interactions between old and new generations so that the science of SAIDs can be transmitted and best adapted to societal transformations and the challenges of tomorrow's medicine.

A young investigator meeting (YIM), and sessions dedicated to patients and families will be held, in parallel with the main sessions. Most importantly, the 13th ISSAID meeting will enable learning from colleagues, collaborators, and experts.

Please be sure to save the dates in your diary – April 08th to 10th, 2025!

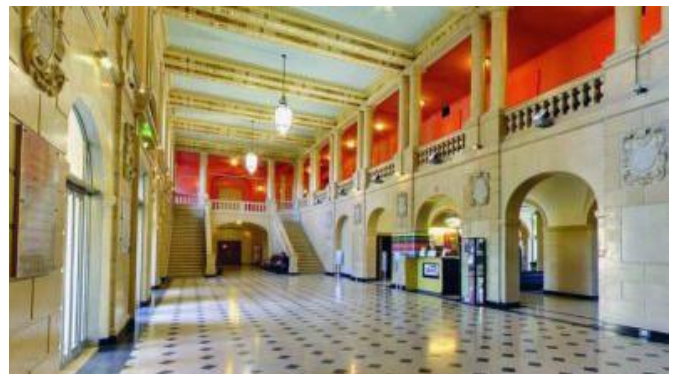
We look forward to welcoming you to Paris in 2025 for exciting science and discussions.

Kind regards,

Isabelle Koné Paut

Head of Pediatric Rheumatology Dept, Kremlin Bicêtre Hospital, FRANCE

Coordinator of the French Reference Center of Autoinflammatory Diseases & Inflammatory Amyloidosis (CEREMAIA)



Welcome & Committees

Executive Committee

Helen Lachmann (UK)
President

Erkan Demirkaya (CA)
President-Elect

Lori Broderick (US)
Secretary

Sinisa Savic (UK)
Treasurer

Karen Durrant (US)
Patient Advocate

Subcommittees

Fatma Dedeoglu (US)
Ovgu Kul Cinar (UK)
Roberta Caorsi (IT)
Education & Training Subcommittee

Sophie Georgin-Lavialle (FR)
Raju Khubchandani (IN)
Clinical Affairs Subcommittee

Dorota Rowczenio (UK)
Genetic Subcommittee

Seth Masters (AU)
Research Subcommittee

International Scientific Committee

Daniel Kastner (US)
Ivona Aksientjevich (US)
Seza Ozen (TR)
Helen Lachmann (UK)
Paul Brogan (UK)
Jordi Anton-Lopez (ES)
Fabrizio De Benedetti (IT)
Marco Gattorno (IT)
Rafaela Golbach-Mansky (US)
Jasmin Kuemmerle Deschner (DE)
Dirk Foell (DE)

Local Organising Committee

Isabelle Koné-Paut
Sophie Georgin Lavialle
Véronique Hentgen
Co-chairs

Isabelle Touitou
Gilles Grateau
Honorary Presidents

Erkan Demirkaya
Fatma Dedeoglu
International ISSAID representatives

Isabelle Koné-Paut
Perrine Dusser
Sophie Georgin Lavialle
Lea Savey
Veronique Hentgen
Mathilde Labouret
David Saadoun
Bruno Fautrel
Arsène Mékinian
Jérôme Hadjadj
Yvan Jamilloux
Thomas Henry
Guislaine Boursier
Eric Jeziorski
CEREMAIA (French Reference Center of Autoinflammatory Diseases & Inflammatory Amyloidosis)

Fatma Dedeoglu (US)
Maryam Piram (CA)
Ronald Laxer (CA)
Erkan Demirkaya (CA)
Sulaiman Al Mayouf (SA)
Marielle van Gijn (NL)
Josef Uziel (IL)
Eldad Benchetrit (IL)
Yelda Biliginer (TR)
Carine Wouters (BE)
Luca Cantarini (IT)

General Information

Meeting Venue

The 13th ISSAID Meeting is located on the first floor of the Maison Internationale.



Maison Internationale
Cité Internationale Universitaire de Paris
17 Boulevard Jourdan
75014 Paris
France
www.ciup.fr

Located in the 14th arrondissement of Paris, the Cité Internationale has an exceptional architectural, artistic and landscape heritage. It is a major heritage site housing architecture, decor and furniture signed by world-renowned creators. Its 34-hectare park is a vast wooded area with more than 400 different plant species.

The Maison Internationale was entirely built thanks to a donation from the American philanthropist John D. Rockefeller Junior. It was designed by the American architect Jens Fredrick Larson in 1936.

How to Get There

Nearest public transport stations

	RER B : Cité Universitaire
	Métro ligne 4 : Porte d'Orléans
	Tramway T3 : Cité Universitaire
	Bus 21 : Cité Universitaire
	Bus 88 : Jourdan-Montsouris
	Bus 67 : Stade Charléty

Parisian transports

A map and some useful information about rates and schedule may be found on the RATP website

<https://www.ratp.fr/en>

Tourism office

All activities and information about the news of the moment in Paris are available on the Tourism office website:

www.parisjetaime.com/eng

Venue Accessibility

The Cité Internationale has PMR access (people with reduced mobility) as well as an elevator allowing access to the upper floor. The accessibility of the Maison Internationale has been improved with the construction of exterior staircases and the installation of lighting ramps. The PRM access map is accessible here:

<https://www.ciup.fr/wp-content/uploads/2022/01/Plan-des-acces-PMR-de-la-Maison-internationale.pdf>

Professional Congress Organiser

MCI Suisse SA

9 Rue du Pré-Bouvier, 1242 Satigny, Switzerland

Email: issaid@mci-group.com

Language

The official Meeting language is English. No simultaneous translation is provided.

Attendance

Session rooms vary in size. Should the Scientific Session of your choice be fully attended, we invite you to attend a different scientific session. Neither ISSAID nor MCI can be held responsible should a delegate be unable to attend a scientific session due to the capacity of the allocated meeting room.

General Information

Meeting Documents

Documents can be collected at the registration area. Badges must be worn at all times during the Meeting.

Phones

As a courtesy to all, phones and other electronic devices must be operated in silent mode during sessions.

Registration Opening Hours

Tuesday 8 April 2025 08:00 - 19:00

Wednesday 9 April 2025 08:00 - 18:00

Thursday 10 April 2025 08:00 - 17:30

Exhibition Opening Hours

The commercial exhibition is located in Salon Honnorat.

Tuesday 8 April 2025 09:30 - 19:00

Wednesday 9 April 2025 09:30 - 16:30

Thursday 10 April 2025 09:30 - 15:30

Poster Viewing

The poster exhibition is located in Salon Honnorat. Presenting authors will be available at their poster for informal discussions during the official breaks and the welcome reception.

Poster session 1 Tuesday 8 April 2025 during coffee breaks and welcome reception

Poster session 2 Wednesday 9 & Thursday 10 April 2025 during coffee breaks

Poster Walks

The moderated poster walks will be located in Galerie Wilson during lunch breaks as follows:

Poster walks 1-5 Tuesday 8 April 2025 12:30 - 13:30

Poster walks 6-10 Thursday 10 April 2025 12:30 - 13:30

Food & Beverage

Catering during official coffee and lunch breaks are included in the registration fee and will be served in designated areas. Additionally, there are two cafés inside the Maison Internationale:

Café du Théâtre: This café offers vegetarian cuisine and products from organic or sustainable farming. The restaurant is also labeled Ecotable, which attests to their commitment to an approach that respects the environment and health. Open Monday to Saturday (12 p.m. to 8 p.m.)

Le Café-Restaurant Universitaire: The café-restaurant offers balanced meals, prepared from fresh produce. Vegetarian menus are offered. The prices are attractive: a full meal at EUR 3.30 for non-scholarship holders. Open Monday to Friday, 11:30 a.m. to 2 p.m. and 6:30 p.m. to 9 p.m.

Wi-Fi Access

Username: **WIFI-COLLOQUES** // Password: **adenauer**

Certificate of Attendance

The certificate of attendance will be sent by email to all delegates after the Meeting.

General Information

Official Meeting Social Events

Welcome Reception: on Tuesday 8 April 2025 from 17:30 - 19:00

Networking Dinner*: on Wednesday 9 April 2025 from 19:30

Networking Dinner*

Time: Wednesday 9 April 2025 from 19:30 to 23:30

Place: Les Salons Hoches: 9 Avenue Hoche, 75008 Paris

Accessible by public transport

- RER B: Cité Universitaire to Châtelet-Les Halles (10 minutes – 5 stops)
- RER A: Châtelet-Les Halles to Charles de Gaulle – Etoile (6 minutes – 2 stops)

**Upon additional registration on a “first come, first served” basis.*

Programme Overview

📍 Espace Adenauer

📍 Salon Gulbenkian

Tuesday 8 April 2025

Opening Ceremony
08:30 - 10:00

Coffee Break & Poster Viewing - **10:00 - 10:30**

Pathways of Autoinflammation
10:30 - 11:30

Mechanisms of Inflammasome Activation
11:30 - 12:30

Lunch Break & Poster Walks 1-5
12:30 - 13:30

Working Parties Updates
12:30 - 13:30

Epidemiology/Vasculitis of SAID
13:30 - 15:00

Molecular Determinants of Innate Immunity
13:30 - 15:00

Year in Review & Award Presentations
15:00 - 16:00

Coffee Break & Poster Viewing - **16:00 - 16:30**

SAIDs Across the Ages
16:30 - 17:30

ISSAID General Assembly
16:30 - 17:30

Welcome Reception
17:30 - 19:00

Programme Overview

📍 Espace Adenauer

📍 Salon Gulbenkian

Wednesday 9 April 2025

Contribution of Algorithms and AI in SAIDs
08:30 - 10:00

Coffee Break & Poster Viewing - **10:00 - 10:30**

Neurologic Inflammation and SAIDs
10:30 - 11:30

Genetics
11:30 - 12:30

Amyloidosis and Renal Complications in SAIDs
11:30 - 12:30

Lunch symposium - supported by Sobi
12:30 - 13:30

New Clinical Lessons in Autoinflammation
13:30 - 15:00

Selected Oral Communications 1
13:30 - 15:00

Thieves Market
15:00 - 16:00

Workshop Autoinflammation and Osteitis 1
15:00 - 16:00

Coffee Break & Poster Viewing - **16:00 - 16:30**

Navigating Autoinflammation through
Generations and Ressources **16:30 - 18:00**

Workshop Autoinflammation and Osteitis 2
16:30 - 17:30

Thursday 10 April 2025

Beyond genetics of SAIDs
08:30 - 10:00

Coffee Break & Poster Viewing - **10:00 - 10:30**

Clonal Haematopoiesis and Autoinflammation 1
10:30 - 11:30

Biomarkers & Outcome Measures in SAIDs
10:30 - 11:30

Clonal Haematopoiesis and Autoinflammation 2
11:30 - 12:30

Selected Oral Communications 2
11:30 - 12:30

Lunch Break & Poster Walks 6-10
12:30 - 13:30

Working Parties Updates
12:30 - 13:30

New Targets for the Treatment of SAID
13:30 - 15:00

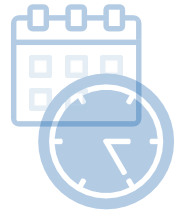
Coffee Break & Poster Viewing - **15:00 - 15:30**

Equity and Access to Innovative Therapeutics
15:30 - 16:30

Closing Ceremony
16:30 - 17:00

Programme Day 1

Tuesday 8 April 2025



08:30 - 10:00

📍 Espace Adenauer

Opening ceremony

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

- 08:30 President's address
Erkan Demirkaya (CA)
- 08:35 Welcome from local chair
Isabelle Koné-Paut (FR)
- 08:40 Announcement of award winners
Erkan Demirkaya (CA) & Isabelle Koné-Paut (FR)
- 09:00 Evolution, genetics of populations
Luis Quintana Murcie (FR)
- 09:20 Surprise from CEREMAIA

10:00 - 10:30

📍 Salon Honnorat

Coffee break, visit of the exhibition & poster viewing - *detailed posters on pages 28 - 33*

10:30 - 11:30

📍 Espace Adenauer

Pathways of autoinflammation

Moderated by Fabio Martinon (CH) & Sinisa Savic (UK)

- 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

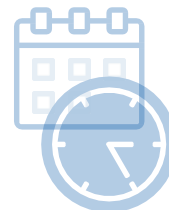
Mechanisms of inflammasome activation

Moderated by Andy Wullaert (BE) & Benedicte Py (FR)

- 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)

Programme Day 1

Tuesday 8 April 2025



12:30 - 13:30

📍 Galerie Wilson

Lunch break & Poster walks 1 - 5 - detailed poster walks on pages 22 - 24

12:30 - 13:30

📍 Salon Gulbenkian

Working parties: Updates from TARN, Eurofever Registry, CARRA & JIRCohort

13:30 - 15:00

📍 Espace Adenauer

Epidemiology/vasculitis of SAID

Moderated by Paul Brogan (UK) & Ovgu Kul Cinar

- 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 O01 - Systemic inflammation, lymphoproliferation and vasculopathy in a patient with ARHGAP10 mutation
Stefano Volpi (IT)
- 14:40 O02 - Biologics in the treatment of pediatric Behçet's disease
Fatma Gül Demirkan (TR)

13:30 - 15:00

📍 Salon Gulbenkian

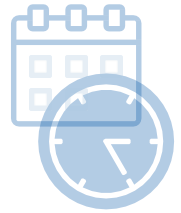
Molecular determinants of innate immunity

Moderated by Zhou Qing (CN) & Hal Hofmann (US)

- 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 O04 - COFILIN-1 is a Redox sensor regulating the NLRP3 inflammasome
Jae Jin Chae (US)
- 14:40 O05 - SGT1 controls NLRC4 inflammasome activation in auto-inflammatory diseases
Cyrielle Hou (CH)
- 14:50 O06 - GSDMD and GSDME amplify NLRP3 activation in autoinflammation
Janset Onyuru (US)

Programme Day 1

Tuesday 8 April 2025



15:00 - 16:00

📍 Espace Adenauer

Year in review & award presentation

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

- 15:00 Basic science
Ivona Aksentijevich (US)
- 15:20 Clinical science
Erkan Demirkaya (TR)
- 15:40 Dan Kastner award presentation
Alice Burleigh (UK) & Marcia Munoz (AU)

16:00 - 16:30

📍 Salon Honnorat

Coffee break, visit of the exhibition & poster viewing - detailed posters on pages 28 - 33

16:30 - 17:30

📍 Espace Adenauer

SAIDs across the ages

Moderated by Serdal Uğurlu (TR) & TBD

- 16:30 Late complications of Familial Mediterranean Fever
Sophie Georgin Lavalie (FR)
- 16:50 SAIDs and pregnancy
Huri Ozdogan (TR)
- 17:10 Debate: Does everybody need life-long treatment?
Pro: *Helen Lachmann (UK)*
Con: *Seza Ozen (TR)*

16:30 - 17:30

📍 Salon Gulbenkian

ISSAID general assembly

For members only

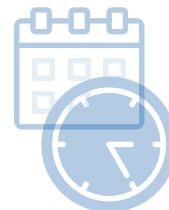
17:30 - 19:00

📍 Salon Honnorat

Welcome reception, visit of the exhibition & poster viewing - detailed posters on pages 28 - 33

Programme Day 2

Wednesday 9 April 2025



08:30 - 10:00

📍 Espace Adenauer

Contribution of algorithms and AI in SAIDs

Moderated by Rae Yeung (CA) & Marco Gattorno (IT)

- 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 O07 - Protein array profiling identifies distinct auto antibody signatures in SAIDs
Gabriel Alexander Vignolle (AT)
- 09:20 O08 - 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 O09 - 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 O10 - Incorporation of recent selection signals improves variant impact prediction in immunemediated genes
Brynja Matthiasardottir (US)
- 09:50 O11 - Improved machine learning models for predicting colchicine resistance in FmF
Admir Ozturk (TR)

10:00 - 10:30

📍 Salon Honorat

Coffee break, visit of the exhibition & poster viewing - detailed posters on pages 34 - 39

10:30 - 11:30

📍 Espace Adenauer

Neurologic inflammation and SAIDs

Moderated by Joost Frenkel (NL) & Jasmin Kuemmerle-Deschner (DE)

- 10:30 Neuroinflammation and SAIDs
Pierre Ellul (FR)
- 10:50 Functional MRI studies in neuroinflammation
Neil Basu (UK)
- 11:10 O12 - Diagnosis of cryopyrin-associated periodic syndrome (CAPS) in adulthood: Lessons from a french cohort
Yixiang Yves-Jean Zhu (FR)
- 11:20 O13 - Neutrophil extravasation and BBB disruption in murine NOMID: Insights into neuroinflammation
Hal Hoffman (US)

Programme Day 2

Wednesday 9 April 2025



11:30 - 12:30

📍 Espace Adenauer

Genetics

Moderated by Guislaine Boursier (FR) & Marielle van Jin (NL)

- 11:30 Whole genome sequencing and neonatal screening: The GUARDIAN experience
Alban Ziegler (US)
- 11:50 Which techniques for the diagnosis of SAIDs in 2025
Alexander Hoishen (NL)
- 12:10 O14 - IRAK2 deficiency causes a new immune dysregulation disorder
Yudie Fei (CN)
- 12:20 O15 - 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 Gül (TR)
- 12:10 AA amyloidosis complicating monogenic autoinflammatory diseases
Sophie Georgin Lavalie (FR)

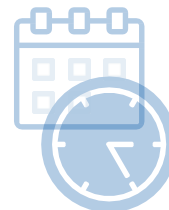
12:30 - 13:30

📍 Espace Adenauer

Lunch sponsored symposium supported by Sobi - detailed programme on page 41

Programme Day 2

Wednesday 9 April 2025



13:30 - 15:00

📍 Espace Adenauer

New clinical lessons in autoinflammation

Moderated by Jeroen van der Hilst (BE) & Seza Ozen (TR)

- 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 Novel insights and treatment for Blau syndrome
Carine Wouters (BE)
- 14:50 O16 - Rare TNFAIP3 hypomorphic variants are a massively underestimated driver of human autoinflammatory disease globally
Daniella Schwartz (US)

13:30 - 15:00

📍 Salon Gulbenkian

Selected oral communications 1

Moderated by Roberta Caorsi (IT) & Sefi Uziel (IL)

- 13:30 O19 - Pulmonary arterial hypertension with Still's disease: A new pulmonary manifestation associated with HLA-DRB1*15
Stéphane Mitrovic (FR)
- 13:40 O20 - 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 O21 - Current treatment of macrophage activation syndrome worldwide: The Metaphor project, a PREs/PRINTO real-life international survey
Francesca Minoia (IT)
- 14:00 O22 - 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
Helen Lachmann (UK)
- 14:10 O23 - Progress report on IL-6 inhibition in ROSAH autoinflammatory disease
Christina Kozycki (US)
- 14:20 O24 - Prospective follow up of 37 pregnancies in women receiving IL-1 inhibitors for systemic autoinflammatory diseases: A multicentric french study from the GR2 cohort
Marion Delplanque (FR)

Programme Day 2

Wednesday 9 April 2025



15:00 - 16:00

📍 Espace Adenauer

Thieves market

Moderated by Isabelle Koné-Paut (FR) & Raju Khubchandani (IN)

- 15:00 O25 - Forgotten but not gone
Adrien Schwartz (FR)
- 15:10 O26 - CNO: Voice of the people - unusual presentation
Toni Hospach (DE)
- 15:20 O27 - The bane of a vein
Archana Khan (IN)
- 15:30 O28 - CAPS: Beware a trap
Anna Agrusti (IT)
- 15:40 O29 - A male turner and the turning point
Federica Anselmi (FR)

15:00 - 16:00

📍 Salon Gulbenkian

Workshop: Autoinflammation and osteitis - Part I (Upon additional registration - Limited seats)

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

- 15:00 CRMO pathophysiology, diagnosis
Christian Hedrich (DE)
- 15:20 CRMO- associated diseases
Herman Girschick (DE)
- 15:40 O30 - OGFR1 gene mutations may link cherubism to chronic recurrent multifocal osteomyelitis (CRMO)
Linda Rossi-Semerano (FR)
- 15:50 O31 - 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)

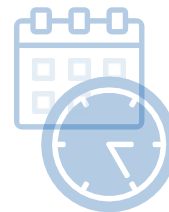
16:00 - 16:30

📍 Salon Honnorat

Coffee break, visit of the exhibition & poster viewing - detailed posters on pages 34 - 39

Programme Day 2

Wednesday 9 April 2025



16:30 - 18:00

📍 Espace Adenauer

Navigating autoinflammation through generations and resources

Moderated by Sophie Georgin-Lavialle (FR) & Helmut Witkowski (DE)

- 16:30 Summary of Early Career Group meeting
Fatma Dedeoglu (US)
- 16:40 27 years in autoinflammation
Seza Ozen (TR)
- 16:55 Autoinflammatory diseases in emerging countries: Challenges & perspectives
Rim Bourguiba (TN)
- 17:05 Navigating STING trafficking in autoinflammation: Insights from translational research in France
Marie-Louise Frémond (FR)
- 17:20 Access to genetic testing and tackling and securing treatment for an expanding spectrum of patients with autoinflammatory diseases: History - present obstacles - moving forward together
Raphaëla Goldbach-Mansky (US)
- 17:40 Panel discussion and Q&A

16:30 - 17:30

📍 Salon Gulbenkian

Workshop: Autoinflammation and osteitis - Part II (Upon additional registration - Limited seats)

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

- 16:30 Monogenic auto-inflammatory disease with bone osteitis
Polly Ferguson (US)
- 16:50 Radiologic findings of CRMO
Sandrine Chilloh (FR)
- 17:10 Debate session: Pro anti-TNF in CRMO versus pro bisphosphonates in CRMO?
Anja Schnabel (DE) and Perrine Dusser (FR)

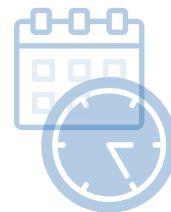
19:30 - 23:30

📍 Les Salons Hoches

Networking dinner (Upon additional registration only)

Programme Day 3

Thursday 10 April 2025



08:30 - 10:00

📍 Espace Adenauer

Beyond genetics of SAIDs

Moderated by Thomas Henry (FR) & Yael Shinar (IL)

- 08:30 Conceptualization of immune memory and trained immunity
Athanasios Ziogas (NL)
- 08:50 Epigenetic determinants in inflammasomopathies
Florence Apparailly (FR)
- 09:10 Cholesterol metabolism and inflammation
Sam Lockhart (UK)
- 09:30 O32 - Transcriptomic insights into PFAPA syndrome: RNA-sequencing analysis of flare relative to non-flare states
Sivia Kerry Lapidus (US)
- 09:40 O33 - Structure and function of pyrin inflammasome: Mechanistic link between FmF and NOCARH syndrome
Wonyong Lee (KR)
- 09:50 O34 - Mevalonate Kinase deficiency – An autoinflammatory disease of dysregulated NK cells
Michael Rogers (AU)

10:00 - 10:30

📍 Salon Honorat

Coffee break, visit of the exhibition & poster viewing - detailed posters on pages 34 - 39

10:30 - 11:30

📍 Espace Adenauer

Clonal haematopoiesis and autoinflammation 1

Moderated by Sinisa Savic (UK) & Arsene Mekinian (FR)

- 10:30 Clonal haematopoiesis and SAIDs
Arsene Mekinian (FR)
- 10:50 VEXAS syndrome in 2025
David Beck (US)
- 11:10 O35 - 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 O36 - Emerging treatment strategies for VEXAS syndrome: A systematic review and meta-analysis
Berkay Kilic (TR)

Programme Day 3

Thursday 10 April 2025



10:30 - 11:30

📍 Salon Gulbenkian

Biomarkers & outcome measures in SAIDs

Moderated by Véronique Hentgen (FR) & Dirk Foell (DE)

- 10:30 Neutrophil activation markers in autoinflammation
Dirk Foell (DE)
- 10:50 Quality of life in SAIDs
Maryam Piram (CA)
- 11:10 O37 - Role of IL-18 as a biomarker in monitoring pediatric patients with Still's disease
Matteo Trevisan (IT)
- 11:20 O38 - 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

📍 Espace Adenauer

Clonal haematopoiesis and autoinflammation 2

Moderated by Juan Arostegui (ES) & Dorota Rowczenio (UK)

- 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)

Programme Day 3

Thursday 10 April 2025



11:30 - 12:30

📍 Salon Gulbenkian

Selected oral communications 2

Moderated by Helen Lachmann (UK) & Paul Brogan (UK)

- 11:30 O39 - Deep phenotyping identifies inflammatory pathways associated with disease activity of VEXAS syndrome
Kana Higashitani (JP)
- 11:40 O40 - 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 O41 - Phosphomevalonate kinase deficiency: Uncovering new dimensions of the disease phenotype
Ezgi Deniz Batu (US)
- 12:00 O42 - Characteristics and problems of japanese patients with UBA1 variant-negative VEXAS syndrome-like autoinflammatory disease
Ayaka Maeda (JP)
- 12:10 O43 - 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 O44 - 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

📍 Galerie Wilson

Lunch break & Poster walks 6-10 - detailed poster walks on pages 25 - 27

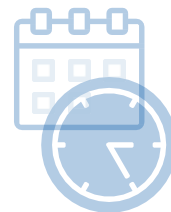
12:30 - 13:30

📍 Salon Gulbenkian

Working parties: Updates from TARN, Eurofever Registry, CARRA & JIRCohort

Programme Day 3

Thursday 10 April 2025



13:30 - 15:00

Espace Adenauer

New targets for the treatment of SAID

Moderated by Ahmet Gul (TR) & Jasmin Kuemmerle Deschner (DE)

- 13:30 Marrow transplant and stem cell transplant for SAID
Isabelle Meyts (BE)
- 13:50 What's new in DADA2 and monogenic vasculitis
Marco Gattorno (IT)
- 14:10 Debate: On demand or continuous treatment with IL-1 inhibition?
Veronique Hentgen (FR) & Ilan Ben-Zvi (IL)
- 14:40 O17 - JAK inhibitors are effective in pediatric refractory NLRC4 gain of function mutation
Jean Jacques De Bruycker (CA)
- 14:50 O18 - Dominant negative ADA2 mutations cause ADA2 deficiency in heterozygous carriers
Marjon Wouters (BE)

15:00 - 15:30

Salon Honnorat

Coffee break, visit of the exhibition & poster viewing - detailed posters on pages 34 - 39

15:30 - 16:30

Espace Adenauer

Equity and access to innovative therapeutics in rare diseases

Moderated by Kenza Bouayed (MA) & Leonardo Oliveira Mendonca (BR)

- 15:30 Patients' odyssey
Jean-Philippe Plancon (FR)
- 15:40 European public policies and rare diseases
Johanna Benesty (FR)
- 15:50 Overcoming the challenges of awareness availability and access
India: *Raju Khubchandani (IN)*
Kenya: *Angela Migowa (KE)*

16:30 - 17:00

Espace Adenauer

Closing ceremony

- 16:30 Closing remarks
Isabelle Koné-Paut (FR)
- 16:45 Announcement of ISSAID 2027
Erkan Demirkaya (CA)

Poster Walks

Tuesday 8 April 2025



Poster Walk 01

📍 Galerie Wilson

Moderated by Sophie Georgin-Lavialle (FR) & Helen Lachmann (UK)

Monogenic autoinflammatory diseases (clinical)

PT01 - Vexas syndrome in France: A multicenter case-series of 318 cases from the French Vexas study group (FRENVEX) / *Sophie Georgin-Lavialle (FR)*

PT02 - Progressive Glomerulonephritis in pediatric SAVI provides insights into pathogenesis and the role of type I IFN in renal outcomes / *Sara Alehashemi (US)*

PT03 - Phosphomevalonate Kinase deficiency expands the genetic spectrum of systemic autoinflammatory diseases / *Juergen Brunner (AT)*

PT04 - Pregnancy outcomes in women with Familial Mediterranean Fever treated with Anakinra: A retrospective study / *Ozgur Can Kilinc (TR)*

PT05 - Immunological insights into H Syndrome: A french national cohort study of 33 patients highlighting auto-inflammatory manifestations / *Maurine Jouret (FR)*

PT06 - Effects of Canakinumab dose adjustments on disease control of autoinflammatory periodic fever syndromes – Interim results of the reliance non-interventional study / *Jasmin Kümmeler-Deschner (DE)*

PT07 - Impact of autoinflammatory diseases: Insights from an interim analysis of the pro-aid study / *Tatjana Welzel (CH)*

PT08 - Prediction of the Colchicine response according to Familial Mediterranean Fever (FmF) 50 score in pediatric patients: Acute phase reactants or existing scoring systems? / *Eda Nur Dizman (TR)*

Poster Walk 02

📍 Galerie Wilson

Moderated by Bruno Fautrel (FR) & Jeroen Van der Hilst (BE)

Monogenic autoinflammatory diseases (clinical)

PT09 - Treatment outcomes in vexas syndrome: A retrospective study from the UK Vexas interest group (VEXNET-UK) / *Adam Al-Hakim (UK)*

PT10 - Infection burden in patients with genetic interferonopathies: A monocentric retrospective cohort study / *Quentin Deletang (FR)*

PT11 - Discontinuation of colchicine treatment in patients with Familial Mediterranean Fever / *Selen Duygu Arık (TR)*

Monogenic autoinflammatory diseases (basic science)

PT12 - Human ADA2 deficiency is characterized by the absence of an intracellular hypoglycosylated form of adenosine deaminase 2 / *Lisa Ehlers (DE)*

PT13 - Pyrin inflammasome activation leads to IL-18 secretion and perpetuates IFN-Gamma secretion in a novel culture-based Mevalonate Kinase deficiency model / *Niels Van Heusden (NL)*

Poster Walks

Tuesday 8 April 2025



PT14 - Autoinflammatory patients with golgi-trapped CDC42 exhibit intracellular trafficking defects leading to stinging hyperactivation and ER stress / *Alberto Iannuzzo (FR)*

PT15 - Gain-of-function human UNC93B1 variants as a novel cause of type I interferonopathy via enhanced TLR7 and TLR8 signalling / *Clémence David-Gabarre (FR)*

PT16 - Persistent IFN signature in patients with papa syndrome and its regulation by JAK inhibition / *Wonyong Lee (KR)*

Poster Walk 03

📍 Galerie Wilson

Moderated by Sarah Bindoli (IT) & David Saadoun (FR)

Monogenic autoinflammatory diseases (basic science)

PT17 - Somatic gain-of-function mutation in TLR7 causes early-onset systemic lupus erythematosus / *Qing Zhou (CN)*

PT18 - Assessment of ADA2 activity levels: Report from the Italian study group on DADA2 / *Roberta Caorsi (IT)*

PT19 - Novel CDC42 mutation reveals a mechanism of pyrin inflammasome activation / *Shouya Feng (AU)*

PT21 - Evidence for dysregulated erythropoiesis in mice and humans with mevalonate kinase deficiency / *Marcia Munoz (AU)*

PT22 - Comparison of immunological biomarkers and lung histology in patients with elevated IL18 - pulmonary alveolar proteinosis and recurrent macrophage activation syndrome (IL-18PAP-MAS) and other inflammatory lung diseases / *Alhanouf Alsaleem (SA)*

PT23 - Investigating NK cell deficiency and dysfunction in Familial Mediterranean Fever within the immunaid cohort: A multi-omics perspective / *Zhicheng Zhou (FR)*

PT24 - Generation of patient-derived iPSCs for hyperimmunoglobulin D syndrome / *Mehmet Emre Özkan (TR)*

Poster Walk 04

📍 Galerie Wilson

Moderated by Ezgi Deniz Batu (US) & Rae Yeung (CA)

Monogenic autoinflammatory diseases (genetics)

PT25 - Compound heterozygous variants in PIGO leading to a novel complement-mediated autoinflammatory disease / *Edan Itzkovitz (CA)*

PT27 - The genetic landscape of primary immune regulatory disorders in Poland / *Katarzyna Babol-Pokora (PL)*

PT28 - Analysis of clinical manifestations across the spectrum of UBA1 mutation burden / *Meghan Anderson (US)*

PT29 - A novel REXO2 variant in a patient with livedo reticularis, palmoplantar erythema and dental disease / *Sofia Torreggiani (US)*

Poster Walks

Tuesday 8 April 2025



PT30 - Mevalonate pathway in autoinflammation: Visualizing the biochemical impairments of mevalonate kinase deficiency / *Jack C. Drda (US)*

PT31 - Performance of targeted gene panel for routine diagnosis of autoinflammatory diseases at the national amyloidosis centre / *Ebun Omoyinmi (UK)*

Non-monogenic SAIDs (clinical)

PT32 - Still's disease associated lung disease: Data from the european registry / *Francesca Minoia (IT)*

Poster Walk 05

Galerie Wilson

Moderated by Susanne Benseler (CA) & Erika Nieuwenhove (NL)

Non-monogenic SAIDs (clinical)

PT33 - Extra-ocular involvement in children with a phenotype suggestive of ocular sarcoidosis / *Inga Turtsevich (UK)*

PT34 - Adult-onset Still's disease: A single-center review of clinical features, treatment, and outcomes / *Oguzhan Omer Kizilkaya (TR)*

PT35 - Anakinra derived amyloidosis detected in two patients, report from the UK national amyloidosis centre / *Dorota Monika Rowczenio (UK)*

PT36 - Unraveling the genetic and transcriptomic drivers of monogenic autoinflammatory diseases in Chile: Bridging gaps in diagnosis and targeted therapy / *Maria Cecilia Poli (CL)*

PT37 - Sitrame syndrome: Insights from 46 patients: The largest cohort study of a novel systemic autoinflammatory disease / *Sophie Georgin-Lavialle (FR)*

PT38 - The prevalence and spectrum of damage in patients with undifferentiated systemic autoinflammatory disease / *Abdurrahman Tufan (TR)*

Non-monogenic SAIDs (basic science)

PT39 - Relevance of pattern recognition receptor signaling in context of multi-mediator inflammation – towards understanding a role of TLR4-dependent damage associated molecular pattern signaling in autoinflammation / *Christoph Kessel (DE)*

PT40 - Phenotypic and functional characterization of innate lymphoid cells in systemic juvenile idiopathic arthritis patients / *Giusi Prencipe (IT)*

Poster Walks

Thursday 10 April 2025



Poster Walk 06

📍 Galerie Wilson

Moderated by TBD & Dirk Foell (DE)

Non-monogenic SAIDs (clinical)

PT41 - Tackling the diagnosis of HA20 in children: Challenges of a highly variable clinical and genetic spectrum / *Federica Anselmi (FR)*

PT42 - What can we learn from the drawing of children with autoinflammatory diseases? / *Noémie Lemoine (FR)*

PT43 - Using a T-cell directed approach in the treatment of DADA2-related neutropenia results in recovery of myeloid cell development pre-transplant and successful engraftment post-transplant / *Amanda Ombrello (US)*

PT44 - Clinical features and efficacy of different modalities of treatment in a patient with NEMO deleted exon 5 autoinflammatory syndrome (NDAS) / *Vasily Igorevich Burlakov (RU)*

PT45 - Renal involvement in autoinflammatory diseases: Data from the Eurofever registry (RIAD project) / *Šárka Fingerhutová (CZ)*

PT46 - Surfing on VUS: Experience in north-east Italy and description of a cohort of adult autoinflammatory patients through a validated next-generation sequencing panel of genes / *Sara Bindoli (IT)*

PT47 - Pregnancy outcomes after maternal and paternal anti-IL-1 treatment exposure in cryopyrin associated periodic syndromes (CAPS) / *Özlem Satirer (DE)*

PT48 - Interleukin-18 and interleukin-1B blockade to control inflammation in PAMI syndrome before and after HSCT / *Benjamin Fournier (FR)*

Poster Walk 07

📍 Galerie Wilson

Moderated by Sivia Lapidus (US) & Ilenia di Cola (IT)

Monogenic autoinflammatory diseases (clinical)

PT49 - Clinical, genetic, and imaging features of aicardi-goutières syndrome in a local cohort in Qatar / *Fatima Al-Maadid (QA)*

PT50 - Effects of canakinumab treatment on common long-term complications in autoinflammatory periodic fever syndromes – 60-month data from the reliance non-interventional study / *Martin Krusche (DE)*

PT51 - Autoinflammatory diseases in the Netherlands: Clinical and genetic insights from the Eurofever registry / *C.W.E. Dermer (NL)*

PT52 - EULAR/PreS endorsed recommendations for the management of Familial Mediterranean Fever (FmF): 2024 update / *Seza Ozen (TR)*

PT53 - A patient with a novel DOCK11 mutation managed with colchicine: A role for pyrin in DOCK11-associated disease? / *Ezgi Deniz Batu (US)*

Poster Walks

Thursday 10 April 2025



PT54 - Biomarker evaluation of disease activity and cardiovascular risk in Familial Mediterranean Fever / *Abdurrahman Tufan (TR)*

PT55 - Clinical practice strategies for the use of BDMARDS in colchicine resistant Familial Mediterranean Fever across the countries; a clips network interim analysis / *Rim Bourguiba (TN)*

PT56 - Compound heterozygosity for MEFV I692DEL and V726A pathogenic variants is associated with a severe phenotype of pyrin-associated autoinflammatory disease with elevated interleukin-18 / *Deborah L Stone (US)*

Poster Walk 08

 **Galerie Wilson**

Moderated by Francesca Minoia (IT) & Sefi Uziel (IL)

Monogenic autoinflammatory diseases (clinical)

PT57 - Clinical features of patients with Familial Mediterranean Fever over 50 years of age: a single-center experience / *Berkay Kilic (TR)*

PT58 - Canakinumab treatment in patients with Familial Mediterranean Fever: A tertiary center experience / *Ozkan Berke Simsek (TR)*

PT59 - Clinical practise strategies for the definition of colchicine resistance in Familial Mediterranean Fever across the countries; a CLIPS network analysis / *Nimet Öner (TR)*

PT60 - Epidemiological and economical factors influencing the colchicine resistance definitions for Familial Mediterranean Fever fever across the countries; a CLIPS network analysis / *Fatih Haşlak (TR)*

PT61 - Trisomy 8 mosaicism with multiple autoinflammatory manifestations including chronic non-bacterial osteomyelitis / *Elizabeth Twynam-Perkins (UK)*

PT62 - IL1 blockade in colchicine resistant Familial Mediterranean Fever - real world data / *Yosef Uziel (IL)*

PT63 - Clinical characteristics and treatment strategies for A20 haploinsufficiency in Japan: A national epidemiological survey / *Hidenori Ohnishi (JP)*

PT64 - AA amyloidosis complicating systemic autoinflammatory diseases: Data from the UK national amyloidosis centre / *Helen Lachmann (UK)*

Poster Walk 09

 **Galerie Wilson**

Moderated by Sulaiman Al-Mayouf (SA) & Jose Hernandez-Rodriguez (ES)

Monogenic autoinflammatory diseases (clinical)

PT65 - Unraveling genetic complexity: different diseases in siblings with shared clinical presentation / *Buthaina Al Adba (QA)*

PT66 - Neurological manifestations in cryopyrin-associated periodic syndromes (CAPS): A retrospective monocentric study / *Majdouline El Moussaoui (BE)*

Poster Walks

Thursday 10 April 2025



PT67 - Translational autoinflammatory research network (TARN): A global network approach to enhancing clinical trial readiness for rare autoinflammatory diseases / *Erkan Demirkaya (CA)*

PT68 - Three-year follow-up of canakinumab dose extension in children with colchicine-resistant FmF PIRA-RG experience / *Fatma Gül Demirkan (TR)*

PT69 - Beyond MEFV: How additional AID-associated mutations shape Familial Mediterranean Fever in children / *Ayşenur Doğru (TR)*

PT70 - Geranygeraniol supplementation leads to an improvement in inflammatory parameters and reversal of the disease specific protein and metabolic signature in patients with hyperIGD syndrome / *Anna Sediva (CZ)*

PT71 - Putting the predict-CRFMF score to the test: Prospective performance evaluation / *Fatma Gül Demirkan (TR)*

Poster Walk 10

📍 Galerie Wilson

Moderated by *Scott Canna (US)* & *Marie-Louise Fremont (FR)*

Monogenic autoinflammatory diseases (clinical)

PT73 - Disease presentation, response to treatment and outcome of pediatric and adult patients with DADA2 (deficiency of adenosine deaminase 2): Results from the Eurofever registry / *Chiara Conti (IT)*

Neurologic inflammation, psychiatry and SAIDs

PT74 - Clinical outcomes of baricitinib treatment in Aicardi-goutières syndrome: A retrospective cohort study at great ormond street hospital / *Afroditi Barmpakou (UK)*

PT75 - Paediatric autoimmune and autoinflammatory disease-related catatonia is associated with elevated CSF interferon- α titres and efficiently treated with immunoadsorption in severe cases / *Isabelle Melki (FR)*

Auto inflammation and vasculitis

PT76 - Loss of PSMD7 causes dysregulated protein degradation, enhanced inflammasome activation, and interferon responses / *Katariina Nurmi (FI)*

PT77 - Neonatal-onset vasculitis driven by pathogenic variants in the src family kinase haematopoietic cell kinase (HCK): a report of two families and a novel mutation / *Fiona Price-Kuehne (UK)*

Innovative therapeutics in Rare Diseases

PT78 - Pre-clinical characterization and clinical evaluation of MAS825, an anti-IL-1 β / anti-IL-18 bispecific antibody for the treatment of inflammasomopathies / *Jiri Kovarik (CH)*

PT79 - Preclinical evaluation of lentiviral gene therapy for the treatment of DADA2: Engraftment and biodistribution studies in humanised NBSGW mice / *Ying Hong (UK)*

Poster Session 1

Tuesday 8 April 2025



Monogenic autoinflammatory diseases (basic science)

Salon Honorat

PO001 - A NOVEL IKBKB VARIANT INCREASES PROTEIN STABILITY AND DRIVES PERSISTENT AUTOINFLAMMATION / *Jingyuan Zhang (CN)*

PO002 - PATHOPHYSIOLOGICAL MECHANISMS REGULATING THE PENETRANCE OF MEFV GENE VARIANTS / *Blandine Monjarret (CA)*

PO003 - UNRAVELING THE CELLULAR MECHANISMS UNDERLYING INFLAMMASOPATHIES USING GENETIC MOUSE MODELS / *Andy Wullaert (BE)*

PO005 - ELEVATED SERUM GASDERMIN D LEVELS IN PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER / *Serdal Ugurlu (TR)*

PO006 - CELL MIGRATION DEFECT IN HYPERIMMUNOGLOBULIN D SYNDROME PATIENT CELLS / *Banu Peynircioğlu (TR)*

PO007 - HUMAN PRIMARY MONOCYTES CELL DEATH AND IL-1B PRODUCTION IS DIFFERENTLY REGULATED IN FMF PATIENTS COMPARED TO HEALTHY CONTROLS / *Corlinda R.E. Kievit (NL)*

PO008 - COMPREHENSIVE ANALYSIS OF IMMUNE DYSREGULATION INDUCED BY A NOVEL GAIN-OF-FUNCTION UNC93B1 HOMOZYGOUS MUTATION IN LUPUS / *Panfeng Tao (CN)*

PO009 - CYP3A4 REGULATION BY MIR-505-5P: A NOVEL INSIGHT INTO COLCHICINE RESISTANCE IN FMF PATIENTS / *Tayfun Hilmi Akbaba (TR)*

PO010 - DISTINCT SERUM IMMUNOREACTIVITY PATTERNS IN MULTIPLE SCLEROSIS AND BEHÇET'S DISEASE: A COMPARATIVE ANALYSIS / *Tayfun Hilmi Akbaba (TR)*

PO011 - INVESTIGATION OF THE MECHANISMS UNDERLYING THE ALTERED EXPRESSION OF MIR-197-3P IN PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER / *Emre Nalbant (TR)*

PO012 - ASSESSMENT OF C26:0 LYSOPHOSPHATIDYLCHOLINE AND CHITOTRIOSIDASE LEVELS IN PATIENTS WITH DEFICIENCY OF ADENOSINE DEAMINASE 2 / *Fabiano Poswar (BR)*

PO015 - PSTPIP1 P.E250K VARIANT ATTENUATES PROTEIN EXPRESSION AND PODOSOME FORMATION IN PATIENT-DERIVED MACROPHAGES / *Nikita Kolchin (RU)*

PO016 - THE ROLE OF FATTY ACIDS IN FAMILIAL MEDITERRANEAN FEVER / *Jeroen van der Hilst (BE)*

PO017 - INVESTIGATION OF INNATE LYMPHOID CELLS IN CHILD PATIENTS DIAGNOSED WITH FAMILIAL MEDITERRANEAN FEVER WITH SPONDYLARTHROSITIS / *Ayşenur Paç Kisaarslan (TR)*

Monogenic autoinflammatory diseases (genetics)

Salon Honorat

PO019 - ASEPTIC ABSCESS SYNDROME: LINKS TO MONOGENIC AUTOINFLAMMATORY DISEASES / *Ludovic Trefond (FR)*

Poster Session 1

Tuesday 8 April 2025



PO020 - THE IMPACT OF NEXT GENERATION SEQUENCING: TEN YEARS' EXPERIENCE OF THE GREAT ORMOND STREET HOSPITAL AUTOINFLAMMATION CENTRE OF EXCELLENCE (GOSH-ACE) /

Fiona Price-Kuehne (UK)

PO021 - TWO NOVEL GAIN-OF-FUNCTION VARIANTS IN ELF4 IN PATIENTS WITH SYSTEMIC UNDEFINED AUTOINFLAMMATORY DISEASE / *Chiara Passarelli (IT)*

PO022 - A RARE AUTOINFLAMMATORY SYNDROME ASSOCIATED WITH A C2ORF69 FRAMESHIFT MUTATION: A CASE REPORT / *Betul Sozeri (TR)*

PO023 - A NOVEL STAT4 VARIANT AS THE POTENTIAL CAUSE OF A LONG-LASTING CASE OF DISABLING PANSCLEROTIC MORPHEA / *Juan I. Arostegui (ES)*

PO024 - A NOVEL NONSENSE MUTATION IN LPIN2 ASSOCIATED WITH MAJEED SYNDROME: CASE REPORT AND INSIGHTS INTO GENE EXPRESSION / *Abdulrahman Abdullah Alrasheed (SA)*

PO025 - WHOLE EXOME SEQUENCING IN PAEDIATRIC-ONSET COGAN'S SYNDROME / *Kirsty McLellan (UK)*

PO026 - LATE ONSET OF AN AUTOINFLAMMATORY DISEASE: IDENTIFICATION AND FUNCTIONAL CHARACTERIZATION OF A MOSAIC VARIATION OF NLRC4 / *Farah Diab (FR)*

PO027 - EVALUATING GENETIC VARIANTS AND THEIR CLINICAL CORRELATIONS IN UNDIFFERENTIATED SYSTEMIC AUTOINFLAMMATORY DISEASES (USAIDS) / *Abdurrahman Tufan (TR)*

PO028 - CLINICAL UTILITY OF EXOME SEQUENCING IN ADULTS WITH AUTOINFLAMMATORY DISORDERS: A PROSPECTIVE STUDY ON 138 PATIENTS / *Antoine Fayand (FR)*

PO029 - MEFV MUTATIONAL SPECTRUM AND CLINICAL MANIFESTATIONS IN GEORGIAN FMF PATIENTS / *Christian Oberkanins (AT)*

PO031 - HARDY-WEINBERG DISEQUILIBRIUM OF MEFV DISEASE ASSOCIATED GENOTYPES IN A LARGE ISRAELI COHORT OF INDIVIDUALS TESTED FOR PRENATAL CARRIER STATE OF GENETIC DISEASES / *Yael Shinar (IL)*

Non-monogenic SAIDs (clinical)

📍 Salon Honnorat

PO033 - DEFINITION OF DISEASE PHENOTYPES IN PEDIATRIC SAPHO SYNDROME: A NATIONAL MULTICENTRIC STUDY / *Caterina Matucci Cerinic (IT)*

PO034 - MACROPHAGIC ACTIVATION SYNDROME IN STILL'S DISEASE: A MULTICENTER OBSERVATIONAL COHORT STUDY / *Joséphine Masserey (CH)*

PO035 - USE AND SAFETY OF DIFFERENT BISPHOSPHONATES IN CHRONIC RECURRENT MULTIFOCAL OSTEOMYELITIS AND A COMPARISON OF THE SIDE EFFECT PROFILE BETWEEN CHRONIC RECURRENT MULTIFOCAL OSTEOMYELITIS AND OTHER UNDERLYING DIAGNOSES / *Ovgu Kul Cinar (UK)*

PO036 - ARE BLOOD MONOCYTES USEFUL IN DIFFERENTIATING PFAPA FROM FMF? INSIGHTS INTO THEIR LIMITS AND COMPLEMENTARY MARKERS / *Emmanuelle QUESTE (FR)*

Poster Session 1

Tuesday 8 April 2025



PO037 - WORLDWIDE EVALUATION OF CLINICAL PRACTICE STRATEGIES (CLIPS) FOR LUNG INVOLVEMENT IN STILL'S DISEASE WITHIN THE JIR-CLIPS NETWORK: A COST ACTION / *Maurine Jouret (FR)*

PO038 - SYNDROME OF UNDIFFERENTIATED RECURRENT FEVER (SURF): CLINICAL AND GENETIC INSIGHTS FROM A MONOCENTRIC HOMOGENEOUS COHORT OF 101 PATIENTS / *Serena Palmeri (IT)*

PO039 - DECODING THE SYNDROME OF UNDIFFERENTIATED RECURRENT FEVER: CLINICAL INSIGHTS, BIOMARKERS, AND TREATMENT OUTCOMES FROM A NATIONAL UK AUTOINFLAMMATORY CENTRE / *Charalampia Papadopoulou (UK)*

PO040 - THE ASSESSMENT OF IL-15 IN PATIENTS WITH STILL'S DISEASE, IN VITRO AND EX-VIVO FINDINGS / *Ilenia Di Cola (IT)*

PO041 - PREDICTIVE FACTORS FOR RELAPSE IN ADULT-ONSET STILL'S DISEASE: A RETROSPECTIVE COHORT STUDY / *Abdurrahman Tufan (TR)*

PO042 - EVALUATION OF THE PATIENTS WITH CHRONIC NON-BACTERIAL OSTEOMYELITIS BASED ON MAGNETIC RESONANCE IMAGING / *Ayşenur Paç Kısaarslan (TR)*

PO043 - STILL'S DISEASE OVER 2 DECADES: LEARNING FROM THE PAST / *Anna Kozáková (CZ)*

PO046 - ADULT PATIENTS WITH UNCLASSIFIED SYSTEMIC AUTOINFLAMMATORY DISEASE - A SINGLE CENTER CASE SERIES / *Jason An (CA)*

PO047 - CUTANEOUS MANIFESTATIONS IN A CHILEAN COHORT WITH SYSTEMIC AUTOINFLAMMATORY DISEASES / *Maria Cecilia Poli (CL)*

PO048 - WORLDWIDE ASSESSMENT OF CLINICAL PRACTICE STRATEGIES (CLIPS) IN STILL'S DISEASE TREATMENT THROUGH THE JIR-CLIPS NETWORK: A COST ACTION / *Sophie Georgin-Lavialle (FR)*

PO049 - CHRONIC RECURRENT MULTIFOCAL OSTEOMYELITIS AND LUNG INVOLVEMENT: REPORT OF TWO CASES / *Chiara Conti (IT)*

PO050 - MACROPHAGE ACTIVATION SYNDROME IN A PATIENT WITH CLERICUZIO-TYPE POIKILODERMA NEUTROPENIA SYNDROME / *Ayşenur Paç Kısaarslan (TR)*

PO051 - CLINICAL AND RADIOLOGICAL FEATURES OF MANDIBULAR CHRONIC NONBACTERIAL OSTEOMYELITIS (CNO): A RETROSPECTIVE CASE SERIES / *Sodality Sutnga (IN)*

PO052 - DISPENSED PRESCRIPTIONS OF ADHD MEDICATIONS TO CHILDREN WITH PFAPA / *Karin Rydenman (SE)*

PO053 - MONITORING COLCHICINE EFFECTIVITY IN CHILDREN WITH PFAPA BY USING AUTOINFLAMMATORY DISEASES ACTIVITY INDEX (AIDAI) SCORES / *Lutfiye Koru (TR)*

PO054 - ATTITUDES TOWARD GENETIC TESTING IN PFAPA SYNDROME: UNVEILING CLINICAL TRENDS FROM THE JIR-CLIPS SURVEY / *Hafize Emine Sönmez (TR)*

PO056 - CARD8-FS VARIANT IN SLOVAK COHORT OF PFAPA PATIENTS / *Lenka Kapustova (SK)*

Poster Session 1

Tuesday 8 April 2025



PO057 - EVALUATION OF SECOND-LINE TREATMENTS IN CHRONIC RECURRENT MULTIFOCAL OSTEOMYELITIS / *Salomé Pacaud (FR)*

PO058 - USE AND SAFETY OF DIFFERENT BISPHOSPHONATES IN CHRONIC RECURRENT MULTIFOCAL OSTEOMYELITIS AND A COMPARISON OF THE SAFETY PROFILE: GOSH EXPERIENCE / *Ovgu Kul Cinar (UK)*

PO059 - YAO SYNDROME IN CHILDREN: A PEDIATRIC CASE SERIES FROM AUTOINFLAMMATION REFERENCE CENTER TÜBINGEN / *Fehime Kara Eroglu (DE)*

PO060 - CHRONIC MACROPHAGE ACTIVATION SYNDROME IN STILLS DISEASE: A CASE REPORT OF SUCCESSFUL TREATMENT WITH JAK AND INTERLEUKIN-1 INHIBITORS / *Raghad Bin Salman (SA)*

PO061 - DIAGNOSIS, TREATMENT AND MONITORING OF PEDIATRIC BEHCET'S DISEASE (BD) AND BD-RELATED PHENOTYPES ON IDENTIFIED MONOGENIC MIMICS: A SYSTEMATIC REVIEW PROTOCOL / *Micol Romano (CA)*

PO062 - NEUROLOGIC PRESENTATIONS IN ELDERLY PATIENTS WITH YAO SYNDROME / *Qingping Yao (US)*

Unusual or unsolved case reports

📍 Salon Honnorat

PO063 - DE NOVO TGFBR1 MUTATION ASSOCIATED WITH ATYPICAL AUTOINFLAMMATORY AND PERIOSTEAL INVOLVEMENT: DIAGNOSTIC CHALLENGES AND THERAPEUTIC APPROACHES / *Martina Rossano (IT)*

PO064 - A NOVEL CASE OF P2XR7 ASSOCIATED AUTOINFLAMMATORY DISEASE SUCCESSFULLY TREATED WITH ANTI-IL1 THERAPY / *Selen Duygu Arık (TR)*

PO065 - A PATIENT WITH VERY EARLY ONSET INFLAMMATORY BOWEL DISEASE (VEO-IBD), HEPATITIS, SPECIFIC ANTIBODY DEFICIENCY AND A VARIANT OF UNCERTAIN SIGNIFICANCE IN PIK3CD / *Shang Ming Samuel Lee (SG)*

PO066 - TRNT1 RELATED AUTOINFLAMMATORY SYNDROME IN A PATIENT WITH PRIMARY CILIARY DYSKINESIA / *Simona Di Gennaro (IT)*

PO067 - NEMO-NDAS: CLINICAL DIVERSITY AND THERAPEUTIC CHALLENGES IN PEDIATRIC CASES / *Fanny Faron (FR)*

PO068 - HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS DURING A RELAPSE OF A VEXAS SYNDROME / *Baptiste Farrulo (FR)*

PO071 - A RETROSPECTIVE ANALYSIS OF THREE PATIENTS WITH A VEXAS-LIKE SYNDROME LACKING DETECTABLE UBA1 MUTATIONS / *Kohei Tsujimoto (JP)*

PO072 - CHRONIC RECURRENT WHEELS AND APHTHOUS ULCERS ASSOCIATED WITH AN MEFV K695R MUTATION / *Leonie S. Herzog (DE)*

PO073 - CHALLENGING DIAGNOSTIC AND THERAPEUTIC JOURNEY IN VEXAS SYNDROME: A CASE REPORT / *Xianghong Jin (CN)*

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PO074 - AN UNUSUAL CASE OF WRISTS AND ANKLES "BOGGY SYNOVITIS": AUTOINFLAMMATION LINKING AUTOIMMUNITY? / *Alessia Omenetti (IT)*

PO075 - CASE REPORT: IDENTIFICATION OF POST-ZYGOTIC MOSAICISM WITH A PATHOGENIC TNFRSF1A VARIANT IN A PATIENT WITH ELEVATED INFLAMMATORY MARKERS / *Barbara Bangol (DE)*

PO076 - AN UNUSUAL KIDNEY PRESENTATION IN GENETICALLY-CONFIRMED FAMILIAL MEDITERRANEAN FEVER / *Alessia Omenetti (IT)*

PO077 - EVALUATION OF A DOMINANTLY INHERITED MEFV VARIANT IN A FAMILY WITH FMF-LIKE PHENOTYPE / *Carl Esperanzate (US)*

PO078 - FATAL RHEUMATOID VASCULITIS ASSOCIATED WITH ANTIPHOSPHOLIPID ANTIBODY POSITIVITY LEADING TO CRITICAL LIMB ISCHEMIA / *Jinsu Park (KR)*

PO079 - EFFICACY OF HIGH DOSES INTRAVENOUS ANAKINRA IN TWO PAEDIATRIC CASES OF TAFRO SYNDROME / *Serena Palmeri (IT)*

PO080 - A RARE CONDITION THAT CAN BE MISTAKEN FOR VASCULITIS: PROLIDASE ENZYME DEFICIENCY / *Abdurrahman Tufan (TR)*

PO081 - A RARE PRESENTATION OF OPTIC DISC EDEMA DIAGNOSIS OF CRYOPYRIN-ASSOCIATED PERIODIC SYNDROMES (CAPS): A CASE REPORT / *Betul Sozeri (TR)*

PO082 - RECURRENT MAS-HLH IN A 68-YEAR-OLD WOMAN WITH ADULT-ONSET STILL'S DISEASE AND STXBP2 MUTATION / *Shaye Kivity (IL)*

PO083 - AN ATYPICAL PRESENTATION OF ANTISYNTHEASE SYNDROME / *Ryan Lethem (UK)*

PO084 - CASE REPORT OF A FEMALE PATIENT WITH OVER 25 YEARS OF RECURRENT FEVER - STILL MORE QUESTIONS THAN ANSWERS / *Ewa Wiesik-Szewczyk (PL)*

PO085 - A PATIENT WITH A VARIANT OF UNKNOWN SIGNIFICANCE (VUS) ON AUTOINFLAMMATORY PHOSPHOLIPASE CG2 (PLCG2) GENE: NEW VARIANT WITH A RARE CLINICAL PRESENTATION? / *Alessia Omenetti (IT)*

PO086 - DEFICIENCY OF ADENOSINE DEAMINASE 2: A TALE OF TWO PATIENTS, ONE MUTATION / *Selen Duygu Arık (TR)*

PO087 - DIFFICULT-TO-TREAT RARE DISEASES: A CHALLENGING CASE OF PASH SYNDROME REFRACTORY TO CONVENTIONAL THERAPY / *Sara Bindoli (IT)*

PO089 - PYODERMA GANGRENOSUM (PG) PRECEDING TAKAYASU ARTERITIS (TA) IN A PEDIATRIC PATIENT: AN ONGOING UNSOLVED SAGA - A CASE REPORT AND LITERATURE REVIEW / *Sodality Sutnga (IN)*

PO090 - PERINATAL ENCEPHALOPATHY IN INFANT WITH TREX1 VARIANT / *Ivanna Romankevych (US)*

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Neurologic inflammation, psychiatry and SAIDs

Salon Honorat

PO091 - MEVALONATE KINASE DEFICIENCY: AN UNDERDIAGNOSED CAUSE OF INFLAMMATION-RELATED ISCHEMIC STROKE – CASE REPORT AND NOVEL GENE MUTATION / *Lyna-Nour Hamidi (CA)*

PO092 - NEUROLOGICAL PHENOTYPES OF SOCS1 HAPLOINSUFFICIENCY: INSIGHTS FROM FUNCTIONAL AND HISTOLOGICAL INVESTIGATIONS / *Serena Palmeri (IT)*

PO093 - STEROID-SENSITIVE NEUROINFLAMMATION IN 2 SIBLINGS WITH AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME / *Raffaele Manna (IT)*

Auto inflammation and vasculitis

Salon Honorat

PO094 - (MONO-) GENETIC MIMICS OF BEHÇET'S DISEASE / *Pascale Kurt (CH)*

PO096 - CENTRAL NERVOUS SYSTEM (CNS) VASCULITIS IN ACTIVATED PHOSPHOINOSITIDE 3-KINASE DELTA SYNDROME 1 (APDS1) TREATED WITH HEMATOPOIETIC STEM CELL TRANSPLANTATION (HSCT): A 39-MONTH FOLLOW-UP AND LITERATURE REVIEW / *Archana Khan (IN)*

PO097 - AUTOINFLAMMATORY MANIFESTATIONS IN A PATIENT WITH TYPE II D2-HYDROXYGLUTARIC ACIDURIA / *Sharon Bout-Tabaku (QA)*

PO098 - A CROSS-SECTIONAL OVERVIEW OF BEHÇET'S DISEASE MANAGEMENT: A TUNISIAN EXPERIENCE / *Rim Bourguiba (TN)*

PO100 - THE PATHOLOGY OF THE SKIN, LYMPH NODES, LIVER, AND BONE MARROW AND RELATED CLINIC-BIOLOGICAL FEATURES IN PATIENTS WITH SEVERE SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS : A CASE SERIES OF 11 PATIENTS / *Elise Sun (FR)*

Poster Session 2

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Monogenic autoinflammatory diseases (clinical)

Salon Honorat

PO101 - CLINICAL OUTCOME AND QUALITY OF LIFE IN PATIENTS WITH ARPC1B DEFICIENCY MANAGED CONSERVATIVELY OR WITH ALLOGENEIC HEMATOPOIETIC STEM CELL TRANSPLANTATION - ON BEHALF OF THE ESID/EBMT INBORN ERRORS WORKING PARTY / *Enrico Drago (IT)*

PO102 - AN UPDATE ON THE CLINGEN MONOGENIC AUTOINFLAMMATORY DISEASES EXPERT CURATION PANELS: A FRAMEWORK FOR INTERPRETING GENETIC FINDINGS IN AUTOINFLAMMATORY DISEASES / *Natalie Deutch (US)*

PO103 - PREGNANCY OUTCOMES IN AUTOINFLAMMATORY DISEASES: A PROSPECTIVE STUDY OF 117 CASES, INCLUDING 79 WITH FAMILIAL MEDITERRANEAN FEVER. / *Lea Savey (FR)*

PO104 - TRACKING COLCHICINE COMPLIANCE IN CHILDREN WITH FMF: CAN HAIR COLCHICINE DOSING PROVIDE USEFUL INFORMATION? / *Veronique Hentgen (FR)*

PO105 - BREAKING THE CYCLE: IMPROVED OUTCOMES IN THE FIRST COHORT OF SECOND-GENERATION CAPS PATIENTS THROUGH EARLY DIAGNOSIS AND TREATMENT / *Senne Cuyx (UK)*

PO106 - LONG-TERM SAFETY AND EFFICACY OF COLCHICINE AND ANTI-IL-1 BLOCKERS IN FMF: RESULTS FROM THE EUROFEVER MULTICENTER OBSERVATIONAL STUDY / *Marco Gattorno (IT)*

PO107 - COMBINATION OF BIOLOGICS AND JAK INHIBITORS IN THE TREATMENT OF REFRACTORY SYSTEMIC AUTOINFLAMMATORY DISEASES / *Abdurrahman Tufan (TR)*

PO108 - AA AMYLOIDOSIS RELATED TO MONOGENIC AUTOINFLAMMATORY DISEASES IN FRANCE: A COHORT STUDY OF 77 CASES / *Majdouline El Moussaoui (BE)*

PO110 - "BALANCING IL-1 BLOCKADE IN DIRA: A TALE OF REMISSION, FLARE, AND THERAPEUTIC PRECISION" / *Reem Abdwani (OM)*

PO111 - EVALUATION OF PULMONARY INVOLVEMENT IN COLCHICINE RESISTANT FMF PATIENTS / *Nihal Şahin (TR)*

PO112 - GLOBAL MORTALITY OF FRENCH PATIENTS WITH SYSTEMIC AUTOINFLAMMATORY DISEASES / *Julie Teillet (FR)*

PO113 - BURDEN OF FATIGUE IN CRYOPYRIN-ASSOCIATED PERIODIC SYNDROMES (CAPS) / *Özlem Satirer (DE)*

PO114 - TYPE I INTERFERON SCORE AS A BIOMARKER OF DISEASE ACTIVITY IN ADA2 DEFICIENCY / *Silvia Federici (IT)*

PO115 - CLINICAL PHENOTYPE AND LABORATORY MARKERS IN PATIENTS AFFECTED BY A20 HAPLOINSUFFICIENCY (HA20): A CASE SERIES FROM TWO ITALIAN CENTERS / *Laura De Nardi (IT)*

PO116 - GENETICS, CLINICAL CHARACTERISTICS, AND MANAGEMENT OF FAMILIAL MEDITERRANEAN FEVER IN DIVERSE POPULATIONS: A COMPARATIVE STUDY OF BARI AND ISTANBUL / *Lara Yagci (TR)*

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PO117 - DIAGNOSTIC DELAY IN SYSTEMIC AUTOINFLAMMATORY DISEASE: PRELIMINARY RESULTS FROM THE EUROFEVER REGISTRY / *Erkan Demirkaya (CA)*

PO118 - ATTACK TRIGGERS IN CHILDHOOD FAMILIAL MEDITERRANEAN FEVER / *Elif Kucuk (TR)*

PO119 - IFIH1 GENE MUTATION AS A CAUSE OF SEVERE HYPERINFLAMMATION – CASE REPORT / *Mudr. Dušana Genšor, Mph. (SK)*

PO120 - THE ROLE OF DEMOGRAPHICS IN THE NATURE OF FAMILIAL MEDITERRANEAN FEVER AND ITS OUTCOMES: A COMPARATIVE INTERNATIONAL STUDY / *Merav Heshin-Bekenstein (IL)*

PO122 - NEUROLOGICAL MANIFESTATIONS AND VASCULOPATHY IN PLCG2–ASSOCIATED ANTIBODY DEFICIENCY AND IMMUNE DYSREGULATION (APLAD): REPORT OF TWO CASES / *Fatma Dedeoglu (US)*

PO123 - GENOTYPE-PHENOTYPE CORRELATIONS IN CRYOPYRIN-ASSOCIATED PERIODIC SYNDROME AMONG TURKISH PATIENTS IN GERMANY AND TURKEY: BEYOND BORDERS / *Yagmur Bayindir (DE)*

PO124 - EVERYDAY LIFE OF PATIENTS WITH AUTOINFLAMMATORY PERIODIC FEVER SYNDROMES DURING LONG-TERM TREATMENT WITH CANAKINUMAB – 5-YEAR DATA FROM THE RELIANCE NON-INTERVENTIONAL STUDY / *Juergen Rech (DE)*

PO125 - CLINICAL FEATURES AND OUTCOMES OF A SMALL VEXAS SYNDROME PATIENTS' COHORT / *Raquel Faria (PT)*

PO126 - FAMILIAL MEDITERRANEAN FEVER AND PAPASH: IL-1 BLOCKERS RECOVER COMPLEX HIDRADENITIS / *Maria Carrabba (IT)*

PO127 - EVOLVING PHENOTYPIC AND GENOTYPIC SPECTRUM OF HUMAN ISG15 AND USP18 DEFICIENCIES / *Alhanouf Alsaleem (SA)*

PO128 - IMPORTANCE OF POTENTIAL PROTHROMBOTIC STATE MARKERS IN BEHÇET'S DISEASE AND CORRELATION WITH VASCULAR ULTRASOUND / *Abdurrahman Tufan (TR)*

PO129 - NEUTRALIZING ANTIBODIES AND ANTI-TUMOR NECROSIS FACTOR (TNF) MONOCLONAL ANTIBODY MEDICATIONS / *Patrycja Hoffmann (US)*

PO130 - CARDIOVASCULAR COMORBIDITIES IN PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER / *Serdal Ugurlu (TR)*

PO132 - AN UNEXPECTEDLY HIGH PREVALENCE OF FAMILIAL MEDITERRANEAN FEVER IN SLOVAKIA – RESULT FROM NATIONAL AWARENESS CAMPAIGN / *Milos Jesenak (SK)*

PO133 - CUTANEOUS MANIFESTATIONS OF AUTOINFLAMMATORY DISEASES IN PATIENTS WITH GENETIC TESTING / *Mariia Beketova (RU)*

PO134 - DADA2: THE FIRST REPORT OF A MULTICENTER EGYPTIAN EXPERIENCE / *Yasmine El Chazli (EG)*

PO135 - PERIODONTITIS IN DADA2 PATIENTS WITH SEVERE NEUTROPENIA / *Lorena Wilson (US)*

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PO136 - DETERMINATION OF THE RELATIONSHIP BETWEEN SERUM PROTEIN 14-3-3 ETA LEVELS AND CLINICAL FEATURES OF THE DISEASE IN PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER /

Abdurrahman Tufan (TR)

PO138 - ST2 AS AN INFLAMMATORY AND CARDIOVASCULAR MARKER IN FMF-ASSOCIATED AMYLOIDOSIS

/ Oguzhan Kizilkaya (TR)

PO140 - REAL-LIFE DATA ON TAPERING OF ANTI-IL-1 THERAPY IN PATIENTS WITH MEVALONATE KINASE DEFICIENCY (MKD) /

Betul Sozeri (TR)

PO141 - INFLAMMATORY LINEAR VERRUCOUS EPIDERMAL NEVUS (ILVEN) - A NEW SKIN MANIFESTATION IN A CHILD WITH BLAU SYNDROME /

Pallavi Pimpale Chavan (US)

PO142 - A TIGHT ROPE (D) WALK IN A CHILD WITH ACUTE ABDOMEN /

Ahmed Jamal (IN)

PO143 - VEXAS SYNDROME AND INFECTIONS: ANALYSIS OF A MULTICENTRIC COHORT AND BRIEF LITERATURE REVIEW /

Sara Bindoli (IT)

PO144 - VALIDATION OF THE COLCHICINE RESISTANCE PREDICTION CRITERIA FROM THE TURPAID COHORT TO THE JIR COHORT: A MULTICENTER DESCRIPTIVE ANALYSIS /

Philippe Mertz (FR)

PO145 - EXPANDING THE GENETIC AND CLINICAL SPECTRUM OF NFKB1 VARIANTS IN CHINESE PATIENTS WITH PRIMARY IMMUNODEFICIENCY /

Jingyuan Zhang (CN)

PO146 - A CASE OF CANDLE SYNDROME PRESENTING AS SERONEGATIVE POLYARTHRITIS /

Samantha Cooray (UK)

PO147 - FAMILIAL MEDITERRANEAN FEVER: DISEASE SEVERITY AND AMYLOIDOSIS IN AN EGYPTIAN COHORT /

Walaa Ibrahim Shoman (EG)

PO149 - A CASE WITH DIAGNOSIS OF MACROPHAGE ACTIVATION SYNDROME AND FAMILIAL MEDITERRANEAN FEVER /

Eda Kayhan (TR)

PO150 - A PARTICULAR PHENOTYPE IN TWO PATIENTS WITH SIDEROBLASTIC ANEMIA, B-CELL IMMUNODEFICIENCY, PERIODIC FEVERS AND DEVELOPMENTAL DELAY /

Mihaela Tatiana Bataneant (RO)

PO151 - INFLAMMATORY BOWEL DISEASE IN NEONATAL-ONSET MULTISYSTEM INFLAMMATORY DISEASE: A GENETIC RISK ASSESSMENT /

Sara Alehashemi (US)

PO152 - DIAGNOSTIC CHALLENGE: TACKLING VEXAS SYNDROME IN LOW RESOURCE COUNTRIES /

Rim Bourguiba (TN)

PO153 - ADULT PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER - A SINGLE CENTER CASE SERIES /

Jason An (CA)

PO154 - PATH TO DIAGNOSIS IN FAMILIAL MEDITERRANEAN FEVER (FMF) /

Serdal Ugurlu (TR)

PO155 - TNF-RECEPTOR ASSOCIATED PERIODIC SYNDROME: AN ANALYSIS OF SLOVAKIAN COHORT OF TRAPS PATIENTS /

Branislav Šlenker (SK)

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PO157 - FAMILIAL MEDITERRANEAN FEVER AMONG ADULT PATIENTS: A MULTICENTRIC STUDY IN TUNISIA / *Rim Bourguiba (TN)*

PO158 - NEMO-NDAS: DIVERSE CLINICAL PRESENTATIONS AND PENETRANCE IN THREE PEDIATRIC PATIENTS / *Abdulrahman Abdullah Alrasheed (SA)*

PO159 - A CASE OF MUCKLE-WELLS SYNDROME WITH HYPERTROPHIC PACHYMEMINGITIS IN WHICH A NOVEL NLRP3 GENE VARIANT WAS IDENTIFIED / *Hiroaki Ida (JP)*

PO160 - VEXAS SYNDROME IN TUNISIA: A RARE DIAGNOSIS, A FIRST REPORT / *Rim Bourguiba (TN)*

PO161 - ANAKINRA IS A FAST AND EFFECTIVE TREATMENT OPTION IN SUBSIDING FMF EPISODES AND THEREBY, IN DECREASING HOSPITALIZATION RATES : FUTURE PROSPECTS FOR READILY AVAILABLE, HOME-USE ANAKINRA INJECTORS / *Sezgin Sahin (TR)*

PO162 - CLINICAL SPECTRUM AND GENETIC PROFILE OF MEVALONATE KINASE DEFICIENCY: OUR EXPERIENCE FROM NORTH-WEST INDIA / *Deepti Suri (IN)*

PO164 - PROLONGED URTICARIA AND FEVER IN A LIBYAN TODDLER BOY / *Awatif Abid Abushhaiwia (LY)*

PO165 - CLINICAL AND GENETIC PROFILE OF ROMANIAN PATIENTS DIAGNOSED WITH MEFV-ASSOCIATED AUTOINFLAMMATORY DISEASE: PRELIMINARY RESULTS OF THE RO_FMF STUDY / *Alexandru Stieber (RO)*

PO166 - CLINICAL PRESENTATION AND GENETICS OF SUSPECTED MONOGENIC AUTOINFLAMMATORY DISEASES IN A SINGLE CENTRE AUSTRALIAN PAEDIATRIC RHEUMATOLOGY COHORT / *Georgina Tiller (AU)*

PO167 - PRESENTATION OF PATIENTS WITH NOD2 GENE VARIANTS - A CASE SERIES / *Horea Cioran (RO)*

PO168 - INFLAMMATORY BIOMARKER ANALYSIS CONFIRMS REDUCED DISEASE SEVERITY IN HETEROZYGOUS PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER / *Inès Elhani (FR)*

PO169 - PERIPHERAL ANEURYSMS IN A PATIENT WITH DEFICIENCY OF ADENOSINE DEAMINASE 2 (DADA2) / *Abdurrahman Tufan (TR)*

PO170 - SUSPECTED NLRC4 IN LIBYAN PATIENTS WITH PERSISTENT SKIN RASH / *Awatif Abid Abushhaiwia (LY)*

PO171 - UNRAVELING INFLAMMASOMOPATHIES: INSIGHTS FROM A SINGLE -CENTER EXPERIENCE IN ARGENTINE / *Maria Katsicas (AR)*

Non-monogenic SAIDs (basic science)

Salon Honorat

PO172 - MULTISYSTEM INFLAMMATORY SYNDROME IN CHILDREN IS ASSOCIATED WITH TGF-B-INDUCED EPSTEIN-BARR VIRUS REACTIVATION / *Tilman Kallinich (DE)*

PO173 - LONG-TERM VIRAL PRESENCE IN MONOCYTES CORRELATES WITH DYSREGULATION OF INNATE IMMUNITY IN PATIENTS WITH MIS-C / *Enrico Drago (IT)*

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PO174 - UNDERSTANDING HOST-MICROBIOME RELATIONSHIPS IN THE PATHOPHYSIOLOGY OF BEHÇET SYNDROME: ANALYSIS OF SALIVARY CYTOKINES, SERUM CYTOKINES, AND TRYPTOPHAN METABOLITES CONCENTRATIONS IN PATIENTS FROM THE BEHCETBIOT STUDY. / *Morgane Joubert (FR)*

PO175 - DISSECTING THE HLH IMMUNE SYNAPSE (IS): CRITICAL ROLES FOR IS TERMINATION, CYTOKINE INTENSITY, AND TARGET CELL DEATH / *Scott Canna (US)*

PO176 - NATURAL KILLER CELL EXHAUSTION AND DYSFUNCTION AS A HALLMARK OF THE INFLAMMATION IN STILL'S DISEASE / *Jarne Beliën (BE)*

PO177 - MULTI-OMIC STUDY IN PATIENTS WITH SITRAME SYNDROME IDENTIFIES DIFFERENCES IN SYSTEMIC IMMUNE RESPONSES / *Yixiang Yves-Jean Zhu (FR)*

PO178 - EVALUATION OF TYPE I INTERFERON SIGNATURE AS A BIOMARKER FOR DISEASE ACTIVITY IN JUVENILE DERMATOMYOSITIS / *Anna Mensa-Vilaro (ES)*

PO179 - PLASMA PROTEOMIC PROFILES SEPARATE SURF PATIENTS FROM FMF AND PFAPA: PRELIMINARY DATA FROM THE PERSAIDS PROJECT. / *Federica Penco (IT)*

PO180 - PERSONALIZED MEDICINE FOR SYSTEMIC AUTOINFLAMMATORY DISEASES: THE EUROPEAN MULTICENTER "PERSAIDS" PROJECT / *Marco Gattorno (IT)*

PO181 - UNVEILING THE UNIQUE IMMUNOPATHOGENESIS OF CHRONIC RECURRENT MULTIFOCAL OSTEOMYELITIS THROUGH SINGLE-CELL RNA SEQUENCING / *Ye Won Moon (KR)*

Allied Health Professionals

📍 Salon Honorat

PO182 - BRIDGING THE GAP: DISPARITIES IN GENETIC TESTING AND TRAINING FOR AUTOINFLAMMATORY DISEASES BETWEEN EMERGING AND DEVELOPED COUNTRIES: A SURVEY ON 258 DOCTORS FROM 10 FRENCH SPEAKING COUNTRIES / *Rim Bourguiba (TN)*

PO183 - TO ASSESS HEALTH-RELATED QUALITY OF LIFE (HRQOL) IN PEDIATRIC PATIENTS WITH HEREDITARY AUTOINFLAMMATORY DISEASES (HAID) IN INDIA USING KIDSCREEN - 52 QUESTIONNAIRE - PILOT STUDY / *Archana Khan (IN)*

PO184 - INSIGHTS FROM A ONE-YEAR INTERNATIONAL MONTHLY QUIZ ON AA AMYLOIDOSIS CAUSES: ENGAGING 2,567 VOTERS ACROSS FRENCH-SPEAKING COUNTRIES / *Rim Bourguiba (TN)*

Clonal haematopoiesis and auto inflammation

📍 Salon Honorat

PO185 - CLINICAL FEATURES AND TREATMENT OUTCOMES IN VEXAS SYNDROME: A RETROSPECTIVE SINGLE CENTER EXPERIENCE / *Ozgur Can Kilinc (TR)*

Innovative therapeutics in rare diseases

📍 Salon Honorat

PO186 - IW-601, A FIRST-IN-CLASS CLINICAL-STAGE MONOCLONAL ANTIBODY TARGETING A NOVEL ADHESION CHECKPOINT ON MYELOID CELLS: POTENTIAL FOR TREATMENT OF AUTOINFLAMMATORY AND AUTOIMMUNE INDICATIONS / *Tamar Rachmilewitz Minei (US)*

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Miscellaneous and other

Salon Honorat

PO187 - KIKUCHI-FUJIMOTO DISEASE AS PRESENTING SIGN OF INBORN ERRORS OF IMMUNITY: A SINGLE CENTER EXPERIENCE FROM A COUNTRY AT LOW DISEASE PREVALENCE / *Francesca Baudi (IT)*

PO188 - A QUANTITATIVE STANDARDIZED STRATEGY FOR CLINICAL APPLICATION OF TYPE I INTERFERON SIGNATURE / *Stefano Volpi (IT)*

PO189 - BRIDGING THE GAP: WHEN INSTAGRAM BECOMES A TOOL FOR PATIENT EDUCATION IN FAMILIAL MEDITERRANEAN FEVER / *Zohra Aknouche (FR)*

PO190 - SYSTEMIC AUTOINFLAMMATORY DISEASES IN UKRAINE: CHALLENGES, ACHIEVEMENTS, AND FUTURE PROSPECTS / *Yuriy Stepanovskyy (UA)*

PO192 - DATA FROM THE EUROFEVER REGISTRY FOR CENTRAL AND EASTERN EUROPEAN COUNTRIES: AN UPDATE / *Nataša Toplak (SI)*

PO193 - AA AMYLOIDOSIS IN A FRENCH COHORT OF 312 PATIENTS: A COMPREHENSIVE STUDY ON ETIOLOGIES, DISEASE PROGRESSION AND MORTALITY / *Sophie Georgin-Lavialle (FR)*

PO194 - EMPOWERING PATIENTS WITH RARE AUTOINFLAMMATORY DISEASES: A PIONEER THERAPEUTIC EDUCATION PROGRAM FOR AMYLOIDOSIS, FMF, AND CAPS IN FRANCE / *Sofia Hassina Aloui (FR)*

PO195 - AUTOIMMUNE-LYMPHOPROLIFERATIVE IMMUNODEFICIENCIES: A MONOCENTRIC EXPERIENCE / *Silvia Federici (IT)*

PO196 - EFFECTIVENESS OF IL1 INHIBITION WITH ANAKINRA IN ACUTE AND RECURRENT MYOCARDITIS IN CHILDREN / *Camilla Celani (IT)*

PO197 - CANADIAN AUTOINFLAMMATORY CASE ROUNDS: AN EDUCATION INITIATIVE TO FOSTER A COMMUNITY OF PRACTICE IN AUTOINFLAMMATION THROUGH CONNECTIVISM / *Herman Tam (CA)*

PO198 - SERUM CYTOKINE PROFILES IN NEUTROPHILIC DERMATOSES / *Hanna Bonnekoh (DE)*

PO199 - MUTATION OF X-LINKED INHIBITOR OF APOPTOSIS (XIAP) IN A 14-YEAR-OLD GIRL: CHALLENGING MANAGEMENT OF INFLAMMATORY BOWEL DISEASE (IBD) IN XIAP-DEFICIENT FEMALE PATIENTS / *Chiara Conti (IT)*

PO201 - WHOLE GENOME SEQUENCING OF RECURRENT UNEXPLAINED FEVER IN BRAZIL: A SCREENSHOT OF THE BRAZILIAN NETWORK FOR AUTO INFLAMMATORY AND IMMUNE DYSREGULATORY DISEASES (A CIGEN INITIATIVE) / *Leonardo Mendonça (BR)*

**The Essential
Source**

research
essential
results
impact
hypothesis
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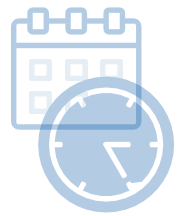
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Sponsored Symposium

Wednesday 9 April 2025



12:30 - 13:30

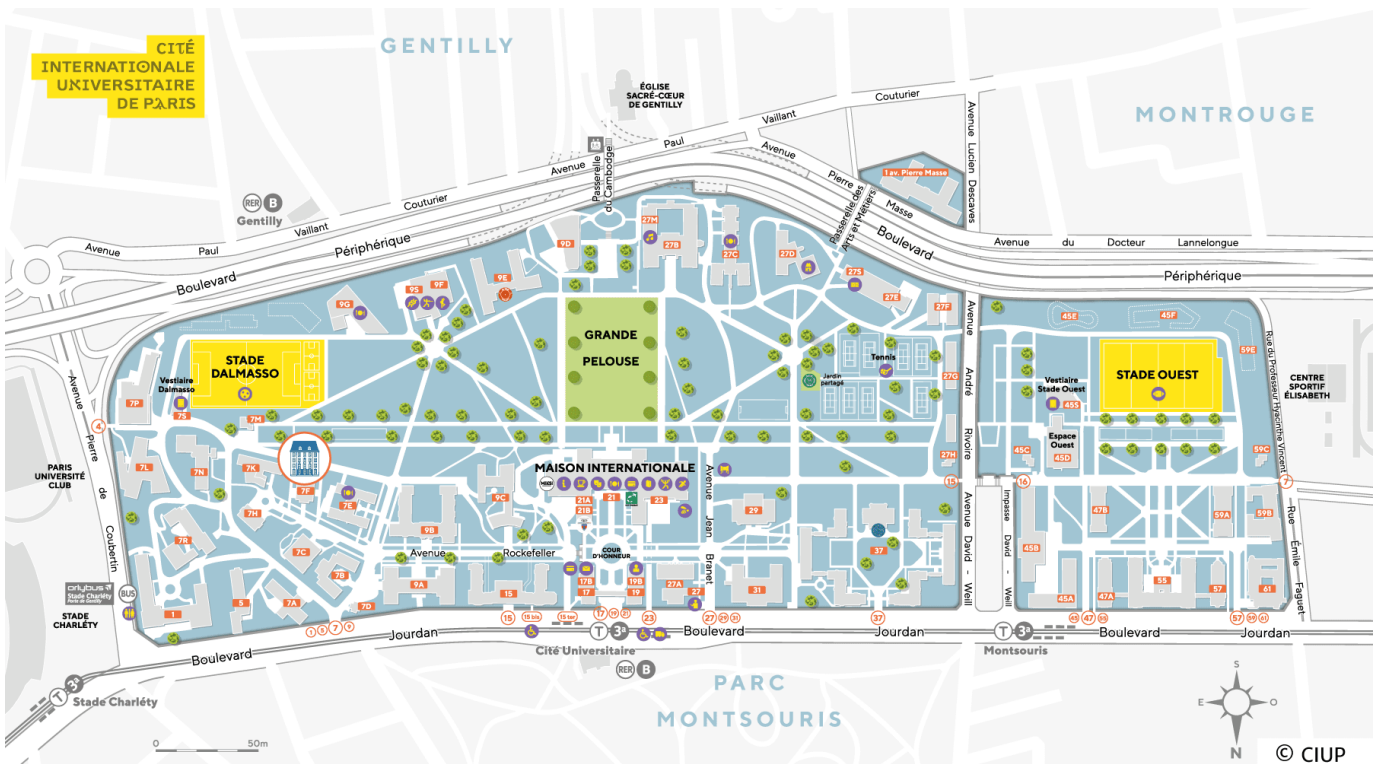
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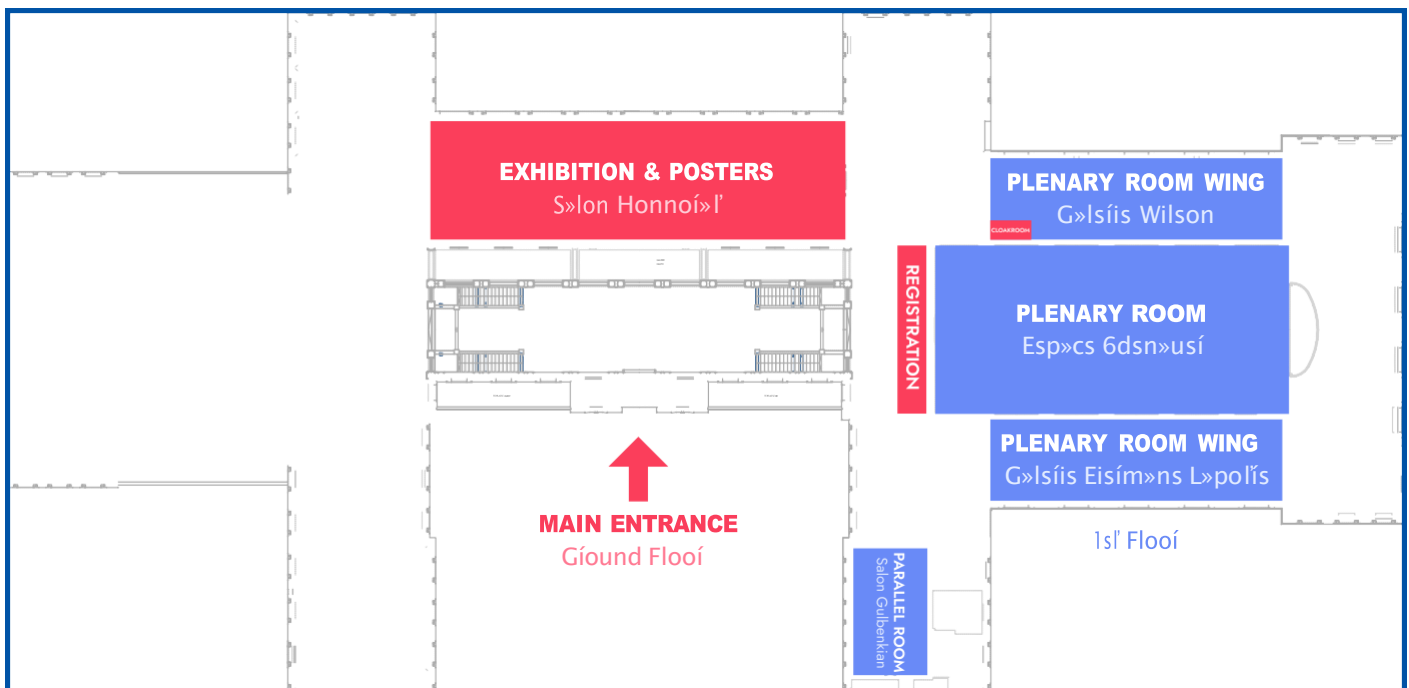
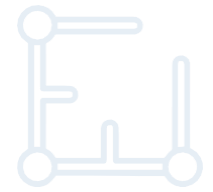
When autoinflammation touches the heart – A case-based discussion

Moderated by Rae Yeung (CA)

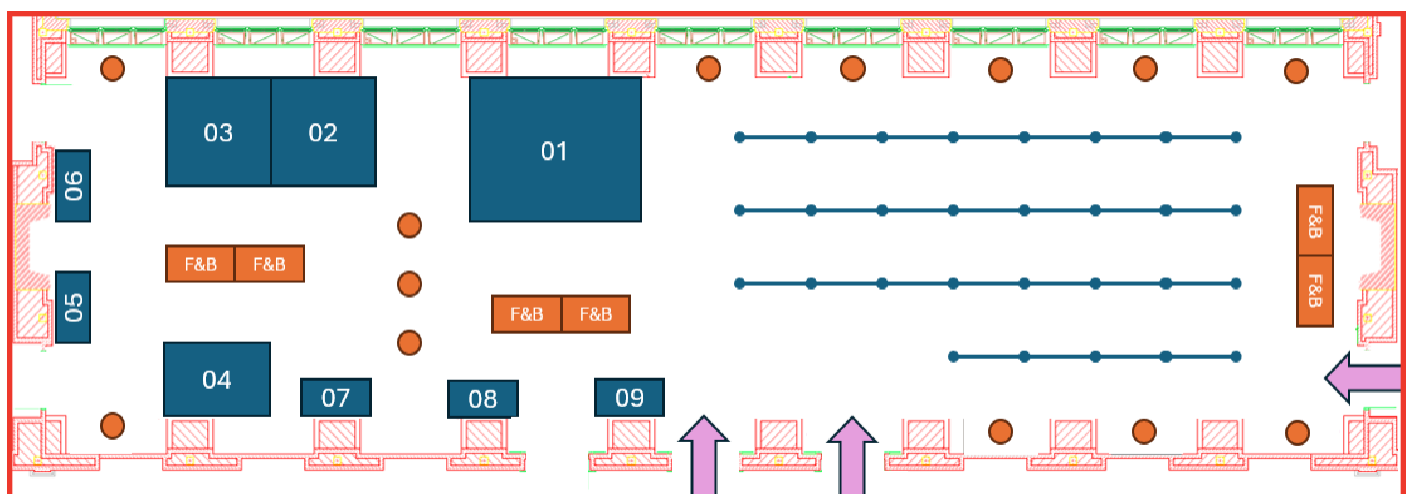
- 12:30 Chair's welcome
Rae Yeung (CA)
- 12:35 Pathophysiology of cardiac inflammation: The example of Still's disease
Yvan Jamilloux (FR)
- 12:50 Cardiac inflammation in Still's disease: Lessons learned from Kawasaki disease
Rae Yeung (CA)
- 13:05 Autoinflammation and recurrent pericarditis
Antonio Brucato (IT)
- 13:20 Q&A and closing remarks
All



Venue Layout



Salon Honnorat



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Acknowledgements



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Company Profiles



Amyloidosis Alliance

Booth N°02

The Amyloidosis Alliance is an international association that supports the fight against amyloidosis worldwide and accompanies patient organizations in different countries.

Communication and mutual support between patients organizations

With the Amyloidosis Alliance, whether you are an association, a patient organization somewhere in the world, by speaking out together we help each other and advance the fight against the disease worldwide.

Amyloidosis Alliance: advocacy at an international level

Through the Alliance, we share our knowledge about the disease. Together, we make sure that awareness, diagnosis, treatment and care, but also research, advance on an international level.

Fondation Arthritis

Booth N°06

The compagny Arthritis R&D, founded in 2014, is a SASU fully owned by the Fondation Arthritis.

Supporting the next generation of researchers is a top priority for Arthritis. Through tailored funding opportunities, we empower young scientists, helping them launch their careers and become future leaders in rheumatic and musculoskeletal diseases (RMD) research.

Our goal is to attract talented researchers to RMD, provide them with the training and resources they need to thrive, and foster bold, high-impact research with strong scientific , clinical and societal value.

CEREMAIA

The Reference Centre for Autoinflammatory Diseases and Inflammatory Amyloidosis (CEREMAIA) is a centre specialising in the treatment of rare autoinflammatory diseases. It has seven sites across France, including the hospitals of Le Kremlin-Bicêtre, La Pitié-Salpêtrière, Tenon, Versailles, Saint Antoine, Lyon and Montpellier.

The main tasks of a centre of reference are (i) to coordinate the structures it brings together; (ii) to provide specialised expertise and act as a referral centre for complex cases; (iii) to initiate and participate in research to improve the understanding and treatment of rare diseases; (iv) to provide training for health professionals and education for patients and their families; and (v) to provide care and follow-up for patients with rare diseases.

CEREMAIA focuses on systemic auto-inflammatory diseases such as monogenic auto-inflammatory diseases, Still's disease and other rare auto-inflammatory syndromes. The centre is part of the FAI2R network, a member of the ERN RITA and also participates in the Île-de-France Paediatric Inflammatory Rheumatism Network.

Company Profiles



Chugai Pharma

Established in 1925, Chugai Pharmaceutical Co., Ltd. (Chugai) is one of Japan's leading research-based pharmaceutical companies.

Boasting a rich 100-year history, Chugai is driven by a mission to add value by creating and delivering innovative products and services for the medical community and human health around the world. Adopting a unique technology-driven approach to drug discovery, its scientists in Japan use proprietary technologies and expertise in disease biology to target antibodies and small/mid-size molecules against disease-causing targets. Seeking to discover first- or best-in-class treatments in areas of high unmet need, Chugai possess a diverse pipeline including candidates in oncology, immunology and rare diseases, with a vision to be a top innovator for advanced and sustainable patient-centric healthcare by 2030.

In 2024, Chugai's European entities united to form one regional organization, combining regional flexibility with a robust quality culture inherited from long-established roots in Japan. Therefore, in Europe, Chugai Pharma Europe is the regional coordinator for commercial, medical, and business activities for Chugai products, as well as on agreements with other external partners, along with supporting clinical research programs from its headquarters in London.

DADA2

The DADA2 Foundation is the world's only organization dedicated to convening a global group of researchers, physicians, and patients to cure DADA2.

Since 2016, we have focused our efforts on gathering 150+ researchers and physicians at one of our four international conferences, working to launch an IRB-approved Patient Registry and now developing the first FDA approved enzyme replacement therapy. At the same time, we continue to be a critical resource for the growing number of DADA2 patients worldwide (approximately 630+) and their physicians.

For more information, visit www.dada2.org

EuroFever Registry

 Booth N°08

The **EuroFever project** was initially promoted in 2008 by the work group of autoinflammatory diseases of the Paediatric Rheumatology European Society (PRES) and was supported by the Executive Agency for Health and Consumers (EAHC), with the main objective of creating a registry of autoinflammatory diseases.

EuroFever currently collects **information on more than 30 monogenic and multifactorial auto-inflammatory diseases**, and more than 135 centres from over 40 countries are contributing with their cases.

The Registry is coordinated by Dr Marco Gattorno and his team, based at IRCCS Istituto Giannina Gaslini in Genoa, Italy.

Company Profiles



Eurofins Biomnis

Booth N°04

Eurofins Biomnis is a major player in specialised clinical diagnostics in Europe offering a variety of over 2,000 tests enabling physicians to make clear and reliable decisions. Clinical diagnostics is at the heart of therapeutic decisions thanks to screening, diagnosis, treatment response prediction and patient monitoring.

Building on its 125 years of expertise, we continue to innovate and use progress in molecular biology to offer tests with high medical added value for accurate and personalised patient management, particularly in auto-inflammatory diseases. Learn more at www.eurofins-biomnis.com

FAI2R

Booth N°05

FAI²R is a national healthcare network for rare autoimmune and autoinflammatory diseases, supported by the Ministry of Health. It facilitates patient care pathways, diagnosis, and treatment for both adults and children. It covers diseases such as lupus, scleroderma, juvenile arthritis, and systemic autoinflammatory diseases.

Its network includes 33 reference centers, 93 expert centers, 18 patient associations, 43 laboratories, and 9 scientific societies. Labeled since 2014, FAI²R is part of the National Rare Diseases Plan 3 (PNMR3), aiming to strengthen expertise and care coordination.

Journal of Rheumatology

The Journal of Rheumatology is a monthly international serial edited by Earl D. Silverman featuring peer-reviewed research articles on clinical subjects from scientists working in rheumatology and related fields, with the support of our esteemed Editorial Committee and Editorial Board.

With over 1600 submissions a year from over 50 countries, each issue of The Journal of Rheumatology includes original clinical research articles, meta-analyses, editorials, historical vignettes, case reports, and letters by world-renowned experts studying drug therapy, patient-reported outcomes, discovery of therapeutic targets in the treatment of rheumatic diseases such as rheumatoid arthritis, ankylosing spondylitis, psoriatic arthritis, osteoarthritis, and other rheumatic and musculoskeletal diseases.

Novartis France

Booth N°03

For over 200 years, Novartis has used scientific research and technological innovation to develop treatments that revolutionize the fight against certain diseases and improve patients' lives.

Novartis is a world leader in healthcare: from research and development (R&D) to manufacturing and distribution of treatments, Novartis contributes to advancing research and reversing disease.

Our mission is to bridge the gap between therapeutic innovations and those who need them: improving patient access to innovations and accelerating the adoption of new treatments by healthcare professionals.

Company Profiles



Recherche et Enseignement en Médecine Interne (REMI)

R.E.M.I (Research and Teaching in Internal Medicine) is an association founded in 1996 by Professor Georges Crémer (1927-2013), President of Paris Descartes University and Head of the Internal Medicine Department at Cochin Hospital. REMI is a non-profit organization under the French Law of 1901; it benefits from the status of a general interest association.

It may receive donations, grants, and legacies. Our goal is to promote clinical research by participating in and funding research activities. We also support and participate in the training of internists and their colleagues.

RESRIP

Booth N°07

RESRIP (RESeau Rhumatismes Inflammatoires Pédiatriques) is a regional network focused on providing expertise and care for children and adolescents with chronic rheumatic and inflammatory diseases in the Paris area. The rarity of these conditions in children, the lack of awareness leading to delayed diagnoses, and the challenges of coordinating care from a variety of trained healthcare professionals close to patients' homes led to the establishment of this network.

RESRIP supports patients in integrating into the school environment, assists with the transition from pediatric to adult care, and follows up with patients for up to two years post-transition. Additionally, the therapeutic education program MIRAJE (Maladies Inflammatoires Rhumatismales Adolescent Jeune Enfant), accredited by the ARS (Agence Régionale de Santé) in 2016, helps patients manage their condition by enhancing their understanding of their disease.

For more information, visit: www.resrip.fr.

Sandoz

Booth N°09

As a global leader in generic drugs and biosimilars, Sandoz is driven by its goal of pioneering patient access to medication.

Sandoz develops, manufactures, and distributes quality medicine at affordable prices, generating savings to contribute to the sustainability of healthcare systems and offer more treatment options to healthcare professionals and patients. Its industry-leading portfolio of more than 1,500 products treats conditions ranging from the common cold to cancer.

The history of Sandoz, headquartered in Basel, Switzerland, dates back to 1886. It is marked by Calcium Sandoz in 1929, the world's first oral penicillin in 1951 and the world's first biosimilar in 2006. Thus, as an independent player, Sandoz intends to confirm its vision and objective for the years to come: to become the leader in generic and biosimilar medicines, recognized worldwide for its impact on patients and society.

Company Profiles



Société Nationale Française de Médecine Interne (SNFMI)

The French National Society of Internal Medicine (S.N.F.M.I.)'s mission is "to study all questions relating to Internal Medicine, to promote its dissemination among doctors and future doctors and to ensure its representation at the national and international level" and for means of action: the organization of congresses, conferences or any other scientific meeting; the publication of a Bulletin; the institution of study missions or prizes.

Initially composed of 20 members elected for four consecutive years by the General Assembly, the Board of Directors of the S.N.F.M.I would later include 24 members (3 per inter-region and 6 for the Ile-de-France, of which 2/3 practiced in C.H.U and 1/3 outside the C.H.U); each member being elected for six consecutive years, the board of directors being renewed by thirds every two years.

SOBI

Booth N°01

Within the therapeutic area of Immunology, Sobi is dedicated to the treatment of rare, ultra-rare and severe diseases. This dedication combined with a rare expertise has resulted in a long history of developing transformative treatments for patients with high unmet medical needs.

Our ability to identify unmet medical needs, develop treatments and secure evidence has been instrumental in the success of our current therapies. Our track record in getting innovative treatments to the people who need them positions us well to take a leading role in Immunology.

We keep challenging the status quo and are dedicated to the continuous discovery of high-potential therapies that we can develop to make a lasting impact and create sustainable value, sooner rather than later. Through close collaboration with rare disease communities, we continue to improve access to care for those otherwise overlooked.

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This image shows a full page of blank, lined paper. It features approximately 20 horizontal blue lines spaced evenly across the page, typical of notebook or legal stationery. The lines are thin and extend from the left edge to the right edge of the page. There is no handwriting, printed text, or other markings on the paper.

A rare expertise in systemic
autoinflammatory diseases



Meet us at ISSAID 2025

We look forward to ISSAID 2025 in Paris and we hope to meet you there. We warmly welcome you to step in to the Sobi exhibition booth where you will be able to meet with Sobi representatives from both the medical and commercial team.