



10<sup>TH</sup> INTERNATIONAL : GENOA, ITALY  
**CONGRESS 2019** : 31 MARCH - 3 APRIL  
FMF and Systemic Auto Inflammatory Diseases



Biannual Meeting of the  
International Society of Systemic  
Auto Inflammatory Diseases (ISSAID)

BRIDGING INFLAMMATION AND IMMUNE-DYSREGULATION

# FINAL PROGRAMME

website [www.issaid2019.org](http://www.issaid2019.org)

email [issaid@mci-group.com](mailto:issaid@mci-group.com)



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## WELCOME MESSAGE

Dear Colleagues,

We are delighted to welcome you to Genoa to the 10th Biannual Meeting of the International Society of Systemic Auto-Inflammatory Diseases (ISSAID). The objective of this congress is to discuss the following main topics: **classical autoinflammatory diseases, mechanisms of disease, new diseases, immune dysregulation, HSCT and gene therapy**, as well as **dermatology** as a special topic.

Most importantly, the goal of the congress is to foster discussion and knowledge exchange among colleagues from the ISSAID community.

The city, known as one of the most important ports on the Mediterranean Sea, welcomes you with its glorious past and impressive landmarks. The ISSAID 2019 Congress gathers over 600 participants, including **rheumatologists, immunologists and paediatricians, biologists** and others with an interest in **systemic auto-inflammatory diseases**.

We wish you a successful and inspiring congress!

**Marco Gattorno**  
(ISSAID President)

**Alberto Martini**  
(Honorary President – ISSAID 2019)

## LOCAL COMMITTEE

Alberto Martini  
*Honorary President*  
Luca Cantarini  
Isabella Ceccherini  
Fabrizio De Benedetti  
Raffaele Manna  
Angelo Marzano  
Angelo Ravelli  
Anna Rubartelli  
Nicola Ruperto

## LOCAL ORGANISING COMMITTEE

Roberta Caorsi  
Eugenia Mosci  
Elisa Patrone  
Paolo Picco  
Stefano Volpi

## ISSAID STEERING COMMITTEE

Marco Gattorno  
*President*  
Helen Lachmann  
*President-Elected*  
Paul Brogan  
*Treasurer*  
Erkan Demirkaya  
*Secretary*  
Daniel Kastner  
Isabelle Toutilou  
Seza Ozen  
Ivona Aksentijevich  
*Past-Presidents*

## CLINICAL COMMITTEE

Eldad Ben Chetrit  
Dirk Foell  
Joost Frenkel

## BASIC SCIENCE COMMITTEE

Hal Hoffman  
Fabio Martinon  
Seth Masters

## GENETIC COMMITTEE

Juan Arostegui  
Marielle van Gijn

## OTHER SUBSPECIALTIES

*Immune dysregulation:*  
Andrew Cant  
Isabelle Meyts

*Dermatology:*  
Dan Lipsker  
Angelo Marzano

# GENERAL INFORMATION

## CONGRESS VENUE

The 10<sup>th</sup> ISSAID International Congress takes place at Genoa's Conference Center located inside Porto Antico's historical Cotton Warehouses.

### **CENTRO CONGRESSI GENOVA**

Porto Antico di Genova – Centro Congressi  
Calata Molo Vecchio 15  
Modulo 5 – 1° Level  
16128 Genova – Italy



## DATE

The Congress opens on Sunday 31 March 2019 and closes on Wednesday 3 April 2019.

## REGISTRATION OPENING HOURS

Sunday 31 March 2019	07:30-20:00
Monday 01 April 2019	07:00-19:00
Tuesday 02 April 2019	07:00-19:00
Wednesday 03 April 2019	07:00-13:30

## SPEAKERS READY ROOM OPENING HOURS

Sunday 31 March 2019	07:30-20:00
Monday 01 April 2019	07:00-19:00
Tuesday 02 April 2019	07:00-19:00
Wednesday 03 April 2019	07:00-12:30



## OFFICIAL CONGRESS SOCIAL EVENTS

**Welcome Reception:** Sunday 31 March 2019 19:30-21:00  
**Congress Dinner\*:** Tuesday 2 April 2019 From 19:30

*\*Upon registration - limited tickets will be available at the registration desks on a "first come, first served" basis.*

## ORGANISING SECRETARIAT

### ISSAID 2019

c/o MCI Suisse SA  
9 Rue du Pré-Bouvier  
1242 Satigny  
Switzerland

### Registration & Accommodation

Tel: +41 22 33 99 702  
Email: [issaid.reghot@mci-group.com](mailto:issaid.reghot@mci-group.com)

### Exhibition & Sponsorship

Tel: + 41 22 33 99 673  
Email: [coralie.deleage@mci-group.com](mailto:coralie.deleage@mci-group.com)

### Speakers & Abstract handling

Tel: + 41 22 33 99 673  
Email: [issaid.abstract@mci-group.com](mailto:issaid.abstract@mci-group.com)

## CONGRESS DOCUMENTS & BADGES

Congress documents and badges must be collected at the registration area at the Congress venue. Name badges must be worn at all time during the Congress.

## FOOD & BEVERAGE

Welcome reception on Sunday, coffee and tea during official breaks are included in the registration fee and will be served in designated areas.

## LANGUAGE

The official Congress language is English. All sessions will be given in English. No simultaneous translation is provided.

## SERVICES FOR THE DISABLED

All the rooms at the Congress Centre are fully accessible to participants with disabilities.

## ATTENDANCE

Meeting 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

## CELLULAR PHONES & PAGERS

As a courtesy to all congress attendees and speakers, cellular phones, pagers, and other electronic devices must be operated in silent/vibrate mode during sessions. Devices that ring, beep, etc. are strictly prohibited. No cellular phone conversations are permitted during sessions/meetings.

## EXHIBITION

A commercial exhibition is presented to all delegates during the ISSAID 2019 Congress. Welcome reception and coffee breaks are served in the exhibition area.

### EXHIBITION OPENING HOURS

Sunday 31 March 2019	19:00-21:00
Monday 01 April 2019	09:30-16:30
Tuesday 02 April 2019	09:30-16:30
Wednesday 03 April 2019	09:30-11:00

# SCIENTIFIC INFORMATION

## SPEAKERS ROOM

Speakers and presenters should deliver and view / check their presentation at the Preview Room (level 1) at least 2 hours prior to the start of their respective Session.

### Presentation format

Power Point™ compatible with Microsoft Office 2013. Recommended format is 4:3.

### Electronic Support

USB only. Videos should be included in your PowerPoint Presentation or at least be in a format that is supported by Windows Media Player. Lecturers are asked to keep their presentations within the given time limits. This is of utmost importance to preserve the session and programme schedule!

## POSTER VIEWING

There will be two different poster viewing sessions. Each poster will be displayed for 1 day and a half.

### Poster Session 1

Monday 01 April 2019 15:00-16:00

### Poster Session 2

Tuesday 02 April 2019 15:00-16:00

Presenting authors will be available at their poster for informal discussions at the above hours.

## POSTER GUIDED TOURS

There will be a total of four poster guided tours taking place during the poster viewing times. Each poster will be displayed for 1 day and a half.

### Poster Guided Tours 1A & 1B

Monday 01 April 2019 15:00-16:00

### Poster Guided Tours 2A & 2B

Tuesday 02 April 2019 15:00-16:00

Each poster tour will be guided by a moderator and presenting authors will be available at their poster for informal discussions at the above hours.

# PROGRAMME OVERVIEW

## SUNDAY 31 MARCH 2019

	BORA	SCIROCCO	LIBECCIO	MAESTRALE
08:30 - 14:00	Patient organisations meeting			
14:00 - 17:00		Mechanisms of disease in autoinflammation	A clinical overview on autoinflammatory diseases	
17:00 - 18:00				ISSAID general assembly
18:00 - 19:30				Opening ceremony
19:30 - 21:00	Welcome reception			

## MONDAY 01 APRIL 2019

	SCIROCCO	LIBECCIO	MAESTRALE
07:30 - 08:30			Wake up early and find a new disease!
08:30 - 10:00			Molecular determinants of innate immunity
10:00 - 10:30	Coffee break and visit of the exhibition		
10:30 - 12:00	Multifactorial vs monogenic autoinflammatory diseases		Mechanisms of inflammasome activation
12:00 - 12:30	Break		
12:30 - 13:30			SOBI - sponsored symposium
13:30 - 15:00			Update of autoinflammatory diseases
15:00 - 16:00	Coffee break and poster guided tours		
16:00 - 17:00	Oral communications - immunology		Oral communications - clinical
17:00 - 18:30			Novel AID pathways and genes



# PROGRAMME OVERVIEW

## TUESDAY 02 APRIL 2019

	SCIROCCO	LIBECCIO	MAESTRALE
07:30 - 08:30			Wake up early and find a new disease!
08:30 - 10:00			Deficiencies in key regulator signals
10:00 - 10:30	Coffee break and visit of the exhibition		
10:30 - 12:00			Nucleid acid sensing and interferon
12:00 - 12:30	Break		
12:30 - 13:30			NOVARTIS - sponsored symposium
13:30 - 15:00			The skin as a target in autoinflammatory diseases
15:00 - 16:00	Coffee break and poster guided tours		
16:00 - 17:15	Genetic diagnosis of SAIDs: towards consensual large-scale approaches and guidelines		Complement and autoinflammation
17:15 - 18:30			Thief market
19:30 - 23:00	ISSAID networking dinner at the Villa Lo Zerbino		

## WEDNESDAY 03 APRIL 2019

	MAESTRALE
07:30 - 08:30	Wake up early and find a new disease!
08:30 - 10:00	New frontiers in the treatment of SAID
10:00 - 10:30	Coffee break and visit of the exhibition
10:30 - 12:00	Novel targets and therapies in autoinflammation
12:00 - 13:00	Oral communications - new diseases
13:00 - 13:30	Closing and young investigator award presentation

## SUNDAY 31 MARCH 2019

<b>08:30 - 14:00</b>	<b>PATIENT ORGANISATIONS MEETING</b> <i>Refer to page 41.</i>	<b>BORA ROOM</b>
<b>14:00 - 17:00</b>	<b>PRE-CONGRESS COURSE: MECHANISMS OF DISEASE IN AUTOINFLAMMATION</b> <i>Chairpersons: Seth Masters (AU) &amp; Stefano Volpi (IT)</i>	<b>SCIROCCO ROOM</b>
<b>14:00</b>	<b>Horror autoinflammaticus</b> Seth Masters (AU)	
<b>14:20</b>	<b>Mechanisms of NLRP3 inflammasome activation</b> Fabio Martinon (CH)	
<b>14:40</b>	<b>When neutrophils are unhappy, everyone is unhappy</b> Steven Holland (US)	
<b>15:00</b>	<b>General discussion</b>	
<b>15:15</b>	<b>Short break</b>	
<b>15:35</b>	<b>Type 1 interferonopathies</b> Yanick Crow (UK)	
<b>15:55</b>	<b>Biomarkers in SAID</b> Dirk Foell (DE)	
<b>16:25</b>	<b>Animal models in SAID</b> Hal Hoffman (US)	
<b>16:45</b>	<b>General discussion</b>	
<b>14:00 - 17:00</b>	<b>PRE-CONGRESS COURSE: A CLINICAL OVERVIEW ON AUTOINFLAMMATORY DISEASES</b> <i>Chairpersons: Roberta Caorsi (IT) &amp; Seza Ozen (TR)</i>	<b>LIBECCIO ROOM</b>
<b>14:00</b>	<b>Inherited periodic fever syndromes</b> Jordi Antón (ES)	
<b>14:20</b>	<b>Autoinflammatory diseases with vasculopathy</b> Seza Ozen (TR)	
<b>14:40</b>	<b>PFAPA and multifactorial SAID</b> Michael Hofer (CH)	
<b>15:00</b>	<b>General discussion</b>	
<b>15:15</b>	<b>Short break</b>	
<b>15:35</b>	<b>Making the diagnosis in SAID - role of clinics and genetics</b> Jasmine Kuemmerle-Deschner (DE)	
<b>15:55</b>	<b>How to measure disease activity and damage in SAID</b> Erkan Demirkaya (CA)	
<b>16:25</b>	<b>IL-1 as target for monogenic and multifactorial SAID</b> Isabelle Koné-Paut (FR)	
<b>16:45</b>	<b>General discussion</b>	
<b>17:00 - 18:00</b>	<b>GENERAL ASSEMBLY</b>	<b>MAESTRALE ROOM</b>

## SUNDAY 31 MARCH 2019

<b>18:00 - 19:30</b>	<b>OPENING CEREMONY</b>	<b>MAESTRALE ROOM</b>
<b>18:00</b>	<b>Welcome</b> Marco Gattorno (IT)	
<b>18:15</b>	<b>Autoinflammatory diseases: Lessons for the clinician</b> Alberto Martini (IT)	
<b>18:40</b>	<b>Targeting IL-1 Beta from gout to cancer</b> Charles Dinarello (US)	
<b>19:05</b>	<b>Autoinflammation at 20: Stepping through the looking glass</b> Daniel Kastner (US)	
<b>19:30 - 21:00</b>	<b>WELCOME RECEPTION</b>	<b>EXHIBITION AREA</b>

## MONDAY 01 APRIL 2019

- 07:30 - 08:30**    **WAKE UP EARLY AND FIND A NEW DISEASE!**    **MAESTRALE ROOM**  
*Discussion led by Fabrizio De Benedetti (IT) and Hal Hoffman (US)*
- 08:30 - 10:00**    **MOLECULAR DETERMINANTS OF INNATE IMMUNITY**    **MAESTRALE ROOM**  
*Chairpersons: Daniel Kastner (US) & Michael McDermott (UK)*
- 08:30**    **Cytosolic innate immunity: sensors and executioners**  
 Feng Shao (CN)
- 08:55**    **The role of protein ubiquitylation in inflammatory and autoimmune diseases**  
 Philip Cohen (UK)
- 09:20**    **The expanding role of cGAS in auto-inflammation**  
 Andreas Ablasser (CH)
- 09:45**    **O01 - New MVK mutant mouse avatars of mevalonate kinase deficiency mimic the underlying defect in protein prenylation in patients**  
 Marcia A Munoz (AU)
- 09:52**    **O02 - Generation and analysis of mice carrying a novel heterozygous missense mutation of a proteasome subunit, PSMB9, in patients with autoinflammation and immunodeficiency**  
 Tsuneyasu Kaisho (JP)
- 10:00 - 10:30**    **COFFEE BREAK & VISIT OF THE EXHIBITION**    **EXHIBITION AREA**
- 10:30 - 12:00**    **MECHANISMS OF INFLAMMASOME ACTIVATION**    **MAESTRALE ROOM**  
*Chairpersons: Fabrizio De Benedetti (IT) & Fabio Martinon (CH)*
- 10:30**    **Cell stress and inflammasome activation**  
 Anna Rubartelli (IT)
- 10:55**    **Loss of cell homeostasis and inflammasome activation**  
 Seth Masters (AU)
- 11:20**    **Familial Mediterranean Fever confers resistance to Yersinia pestis'**  
 Jae Jin Chae (US)
- 11:45**    **O03 - Cofilin-1 is an essential redox sensor for NLRP3 inflammasome activation**  
 Wonyong Lee (US)
- 11:52**    **O04 - Autoinflammatory mutation in NLRC4 reveals an LRR-LRR oligomerization interface**  
 Fiona Moghaddas (AU)
- 10:30 - 12:00**    **MULTIFACTORIAL VS MONOGENIC AUTOINFLAMMATORY DISEASES**    **SCIROCCO-LIBECCIO ROOM**  
*Chairpersons: Maurizio Cutolo (IT) & Huri Ozdogan (TR)*
- 10:30**    **Behçet disease: an update**  
 Luca Cantarini (IT)
- 10:55**    **NLRC4**  
 Scott Canna (US)
- 11:20**    **Systemic onset JIA and MAS**  
 Angelo Ravelli (IT)

## MONDAY 01 APRIL 2019

- 11:45 **O05 - Canakinumab, on a reduced dose or a prolonged dose interval without concomitant corticosteroids and methotrexate, maintains efficacy in systemic juvenile idiopathic arthritis patients in clinical remission**  
Ekaterina Alexeeva (RU)
- 11:52 **O06 - IL-18: CXCL9 ratio as a predictor of treatment response in patients with systemic juvenile idiopathic arthritis treated with Canakinumab**  
Tanja Hinze (DE)
- 12:30 - 13:30 **SPONSORED SYMPOSIUM** **MAESTRALE ROOM**  
*Refer to page 43.*
- 13:30 - 15:00 **UPDATE OF AUTOINFLAMMATORY DISEASES** **MAESTRALE ROOM**  
*Chairpersons: Marco Gattorno (IT) & Hal Hoffman (US)*
- 13:30 **A proposal for new taxonomy for AIDs**  
Eldad Ben Chetrit (IL)
- 13:55 **Year in review - Basic research in AIDs**  
Hal Hoffman (US)
- 14:20 **Year in review - Clinical aspects of AIDs**  
Joost Frenkel (NL)
- 14:45 **O07 - The NIH cohort study of DADA2 patients: Novel insights into pathophysiology and treatment with TNF inhibitors**  
Qing Zhou (CN)
- 14:52 **O08 - Recommendation on colchicine dosing and definition of colchicine resistance/intolerance in the management of FMF**  
Jasmin Kuemmerle-Deschner (DE)
- 15:00 - 16:00 **COFFEE BREAK & POSTER GUIDED TOURS** **POSTER AREA**
- POSTER GUIDED TOUR 1A**  
*Chairpersons: Phil Hashkes (IL) & Raju Khubchandani (IN)*
- PT1A01** **MULTI-OMICS ANALYSIS OF ADA2 DEFICIENCY IN JAPANESE COHORT** / H. Nihira\* - K. Izawa - T. Yasumi - M. Ito - S. Iwaki-Egawa - Y. Sasahara - H. Kanegane - T. Yasu - T. Kubota - S. Takei - D. Keino - E. Nanishi - H. Takada - S. Ohga - S. Kajikawa - M. Takahashi - N. Nakano - O. Ohara - T. Heike - J. Takita - R. Nishikomori
- PT1A02** **THE CLINICAL AND IMMUNOLOGICAL PROFILES OF HAPLOINSUFFICIENCY OF A20 IN JAPAN** / H. Ohnishi\* - T. Kadowaki - N. Kawamoto - T. Hori - K. Nishimura - C. Kobayashi - T. Shigemura - S. Ogata - Y. Inoue - T. Kawai - E. Hiejima - K. Izawa - T. Matsubayashi - K. Matsumoto - M. Takagi - K. Imai - R. Nishikomori - S. Ito - T. Heike - O. Ohara - T. Morio - H. Kanegane - T. Fukao
- PT1A03** **USE OF SIGLEC1/CD169 AS A BIOMARKER FOR MONOGENIC INTERFERONOPATHIES** / B. Orak\* - A. Panzer - M. Theophil - E. Krüger - F. Ebstein - B. Zieba - N. Unterwalder - C. Meisel - T. Kallinich
- PT1A04** **SCREENING OF PATIENTS WITH IDIOPATHIC POLYARTERITIS NODOSA, GRANULOMATOSIS WITH POLYANGIITIS, AND MICROSCOPIC POLYANGIITIS FOR DEFICIENCY OF ADENOSINE DEAMINASE 2 /** O. Schnappauf\* - M. Stoffels - I. Aksentijevich - A. Ombrello - N. Sampaio Moura - K. Barron - D. Kastner - P. Grayson - P. Merkel

## MONDAY 01 APRIL 2019

- PT1A05** **DIAGNOSIS AND LONG-TERM MANAGEMENT OF TYPE I INTERFERONOPATHIES IN A PEDIATRIC RHEUMATOLOGY CENTER** / S. Volpi\* - E. Santori - M. Ricci - P. Picco - A. Tesser - G. I. Rice - R. Caorsi - A. Grossi - I. Ceccherini - A. Martini - Y. J. Crow - A. Magnasco - A. Tommasini - F. Candotti - M. Gattorno
- PT1A06** **RESULTS OF INTERNATIONAL DELPHI SURVEY FOR THE DIAGNOSIS, INVESTIGATION AND MANAGEMENT OF DEFICIENCY OF ADA2** / T. A.-B. Youngstein\* - E. P. Chambers - H. J. Lachmann on behalf of DADA2 Delphi Study Participants
- PT1A07** **THE CLINICAL AND IMMUNOLOGICAL PROFILES OF HAPLOINSUFFICIENCY OF A20 IN JAPAN** / H. Ohnishi\* - T. Kadowaki - N. Kawamoto - T. Hori - K. Nishimura - C. Kobayashi - T. Shigemura - S. Ogata - Y. Inoue - T. Kawai - E. Hiejima - K. Izawa - T. Matsubayashi - K. Matsumoto - M. Takagi - K. Imai - R. Nishikomori - S. Ito - T. Heike - O. Ohara - T. Morio - H. Kanegane - T. Fukao
- PT1A08** **IDIOPATHIC RECURRENT PERICARDITIS: CLINICAL FINDINGS AND TREATMENT APPROACH** / C. Celani\* - S. Federici - A. Tulone - B. Bader Meunier - V. Messia - M. Pardeo - C. Bracaglia - P. Quartier Dit Maire - F. De Benedetti - A. Insalaco

### POSTER GUIDED TOUR 1B

*Chairpersons: Jordi Antón (ES) & Fabio Martinon (CH)*

- PT1B01** **MONOCYTES PROTEOMIC PROFILE OF PATIENTS WITH DIFFERENT AUTOINFLAMMATORY DISEASES: A NEW APPROACH TO CHARACTERIZE THESE DISEASES** / F. Penco\* - A. Petretto - C. Lavarello - I. Gueli - A. Bertoni - A. Omenetti - C. Pastorino - M. Gattorno
- PT1B02** **PRULIPOTENT STEM CELL DERIVED MYELOID CELL LINES FOR DISSECTING THE MECHANISM OF AUTOINFLAMMATION** / M. Saito\*
- PT1B03** **LOSS OF PROTEIN PRENYLATION IN HUMAN MONOCYTES PROMOTES THE FORMATION OF AN NLRP3-DEPENDENT INFLAMMASOME IN A MODEL OF MEVALONATE KINASE DEFICIENCY** / O. Skinner\* - J. Jurczyk - P. Baker - S. Masters - A. Robertson - K. Schroder - S. Mehr - M. Munoz - M. Rogers
- PT1B04** **PERFORMANCE OF TARGETED NGS FOR ROUTINE DIAGNOSIS OF AUTOINFLAMMATORY DISEASES** / G. Boursier\* - C. Rittore - D. Méchin - M. Gutierrez - F. Millharet - G. Sarrabay - I. Touitou
- PT1B05** **LATE-ONSET TRAPS WITH LOW-GRADE MOSAICISM IN TNFRSF1A** / B. P. Kant\* - M. J. Koudijs - R. van 't Slot - J. van Kuik - L. M. Sikkema - J. Frenkel - A. Simon - M. E. van Gijn
- PT1B06** **HETEROZYGOUS TNFAIP3 MUTATION AS THE CAUSE OF AN INTERFERON-MEDIATED NEUROINFLAMMATORY DISORDER** / C. M. Mulhern\* - Y. Hong - E. Omoyinmi - D. McCreary - M. Casimir - C. Hemingway - F. D'Arco - P. Brogan - D. Eleftheriou
- PT1B07** **SYSTEMIC EVALUATION OF GENETIC AND BIOCHEMICAL TESTING FOR DEFICIENCY OF ADA2 (DADA2) FROM THE NIH PATIENT COHORT** / N. Sampaio Moura\* - O. Schnappauf - N. Deutch - Q. Zhou - D. Kastner - I. Aksentjevich

### POSTER VIEWING - MONOGENIC AUTOINFLAMMATORY DISEASES (CLINICAL)

- P1001** **IS THERE ANY DIFFERENCE BETWEEN M694V HETEROZYGOTE AND NON-EXON 10 MUTATIONS ON SYMPTOMS ONSET AND RESPONSE TO COLCHICINE TREATMENT?** / H. Adiguzel Dundar\* - S. Turkucar - C. Acari - O. Altug Guvenmez - B. Makay - E. Unsal
- P1002** **A CASE REPORT OF AICARDI GOUTIERES SYNDROME TYPE 5 IN TWO SIBLINGS MIMICKING JUVENILE IDIOPATHIC ARTHRITIS** / B. Al Adba\* - H. Dauleh



## MONDAY 01 APRIL 2019

- P1003**      **PHENOTYPIC AND GENOTYPIC CHARACTERISTICS AND DAMAGE ACCRUAL OF MONOGENIC AUTOINFLAMMATORY DISEASES OTHER THAN FAMILIAL MEDITERRANEAN FEVER FROM THE PEDIATRIC RHEUMATOLOGY ARAB GROUP (PRAG)** / S. Al-Mayouf\* - A. Almutairi - S. Albrawi - A. AlEnazi - B. Fatallah - A. Alsonbul - M. Abu-shukair - R. Alzyoud - A. Alwahadneh - M. Zlenti - E. Kawaja - K. Khawaja - Z. Almusawi - W. Madan - M. AlMutairi - N. Almuatiri on behalf of Pediatric Rheumatology Arab Group (PRAG)
- P1004**      **AUTO-INFLAMMATORY DISEASES: A RETROSPECTIVE SINGLE CENTER STUDY IN SAUDI ARABIA /**  
A. A. Alrasheed\* - A. Al Harthi - B. Al Rewaithi - W. Suwairi - J. Al Qanatish - F. Al Roqi
- P1005**      **CLINICAL AND GENETIC CHARACTERISTICS OF MYALGIA IN ARMENIAN CHILDREN WITH FAMILIAL MEDITERRANEAN FEVER /** G. G. Amaryan\* - T. F. Sarkisian - N. G. Mkrtchyan - M. M. Papazyan - A. E. Tadevosyan
- P1006**      **THE IMPACT OF AGING ON FAMILIAL MEDITERRINEAN FEVER PATIENTS /** O. Aydin\* - S. Ugurlu - B. Egeli - E. Soykut - D. Demir - H. Ozdogan
- P1007**      **COMORBIDITIES IN FAMILIAL MEDITERRANEAN FEVER /** U. Kaya Akca - B. Balci Peynircioglu\* - Z. S. Arici - E. Avci - Z. Y. Akkaya Ulum - E. Yilmaz - Y. Bilginer - S. Ozen
- P1008**      **BONE METABOLISM IN SYSTEMIC AUTOINFLAMMATORY DISEASES (SAIDS): A CASE - CONTROL STUDY FROM PADOVA COHORT /** S. Bindoli\* - G. Franceschet - P. Galozzi - M. Zaninotto - V. Camozzi - P. Sfriso
- P1010**      **IDENTIFICATION OF NOVEL NLRC4 AND IL2RA VARIANTS IN A FAMILY COHORT WITH JUVENILE-ONSET ARTHRITIS AND RASH /** J. L. Bloom\* - M. L. Curran - S. Canna - H. Hoffman - E. Hsieh
- P1011**      **DISCONTINUATION OF COLCHICINE THERAPY IN CHILDREN WITH FAMILIAL MEDITERRANEAN FEVER /** Y. Butbul\* - R. Silman - S. Fahoum - Y. Berkun
- P1012**      **LONG-TERM FOLLOW UP OF A MEVALONATE DEFICIENCY KINASE PATIENTS COHORT /**  
I. Calvo Penades\* - B. Lopez Montesinos - M. I. Gonzalez Fernandez - M. Marti Masanet - E. Fernandez de la Puebla
- P1013**      **NOVEL ASSAY TO DIAGNOSE AND MONITOR CRYOPYRIN ASSOCIATED PERIODIC SYNDROMES (CAPS) /** F. Carbone\* - L. Cantarini - T. Micillo - M. Alessio - A. N. Olivieri - M. F. Gicchino - A. Insalaco - M. C. Maggio - O. M. Lucherini - R. Scarpioni - M. Piga - M. M. Angioni - L. Obici - A. Simpatico - P. Leccese - R. Consolini - R. Manna - P. Sfriso - S. Bindoli - P. Galozzi - I. Orlando - S. Chiesa - M. Gattorno - G. Matarese
- P1014**      **AN ITALIAN FAMILY WITH FCAS /** M. Carrabba\* - M. Zarantonello - I. Ceccherini - G. Fabio
- P1015**      **SCORES ASSESSMENT FOR CLINICAL MANAGEMENT OF FAMILIAL MEDITERRANEAN FEVER /**  
M. Carrabba\* - M. Zarantonello - G. Fabio
- P1016**      **SINGLE CENTER EXPERIENCE WITH 402 FAMILIAL MEDITERRANEAN FEVER PATIENTS /**  
O. Ozdemir Isik - S. Tekeoglu - D. Temiz Karadag - A. Yazici - A. Cefle\*
- P1017**      **TESTICULAR ISCHEMIA IN DEFICIENCY OF ADENOSINE DEAMINASE 2 (DADA2) /** K. Clarke\* - C. Campbell - E. Eibun Omoyinmi - Y. Hong - M. Al Obaidi - N. Sebire - P. Brogan
- P1018**      **A NOVEL DUPLICATION IN THE X-LINKED INHIBITOR OF APOPTOSIS PROTEIN GENE LEADING TO RECURRENT HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS THAT IS RESPONSIVE TO INTERLEUKIN-1 BLOCKADE /** D. Dissanayake\* - R. Marsh - A. Naqvi - M. Jordan - R. Yeung - R. Laxer
- P1019**      **10 YEAR PROGNOSIS OF PATIENTS DIAGNOSED WITH FAMILIAL MEDITERRANEAN FEVER /** S. Ugurlu - B. H. Egeli\* - A. E. Soykut - B. Ergezen

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- P1020** **IN A FAMILIAL MEDITERRANEAN FEVER PREVALENT REGION, ARE FAMILIAL MEDITERRANEAN FEVER AND BEHÇET'S DISEASE ASSOCIATED?** / O. Alparslan - B. H. Egeli\* - Y. Demirel - S. Ugurlu
- P1021** **DOES TESTING FOR SAA IS MORE BENEFICIAL THAN CRP FOR THE FOLLOW-UP OF PATIENTS WITH FMF?** / O. Selvi - S. Ugurlu - B. Ergezen\* - H. Ozdogan
- P1022** **THE PREGNANCY OUTCOMES IN FMF PATIENTS WHO ARE EXPOSED TO IL-1 BLOCKADE WITH ANAKINRA** / B. Ergezen\* - S. Ugurlu - H. Ozdogan
- P1023** **CLINICAL PICTURE OF 7 PAPA PATIENTS FOLLOWED IN A SINGLE PEDIATRIC RHEUMATOLOGIC CENTER** / S. Federici\* - C. Celani - V. Messina - G. Marucci - C. Kessel - F. De Benedetti - A. Insalaco
- P1024** **CLINICAL PRESENTATION, GENETIC ANALYSIS AND IFN-SCORE IN PATIENTS WITH UNDEFINED INTERFERONOPATHIES** / S. Federici\* - G. Moneta - C. Passarelli - C. Bracaglia - L. Raffaele - F. De Benedetti - A. Insalaco
- P1025** **LESSON FROM EUROFEVER REGISTRY AFTER THE FIRST TEN YEARS OF ENROLLMENT** / M. Finetti\* - I. Gueli - J. Frenkel - S. Ozen - H. Lachmann - F. De Benedetti - I. Koné-Paut - C. Wouters - P. Brogan - H. Girschick - B. Neven - A. Martini - N. Ruperto - M. Gattorno
- P1026** **CLINICAL STUDY OF JAPANESE PATIENTS WITH FEVER OF UNKNOWN ORIGIN: INVESTIGATION OF MUTATIONS IN 22 GENES RELATED TO AUTOINFLAMMATORY DISEASES** / K. Fujimoto\* - Y. Hidaka - Y. Yoshida - M. Hayashi - T. Koga - S. Kaieda - S. Yamasaki - T. Hoshino - H. Ida
- P1027** **A CASE OF ADENOSINE DEAMINASE 2 DEFICIENCY (DADA2) WITH AN UNCOMMON CLINICAL PRESENTATION AND RESPONSE TO IV IG** / F. Garbarino\* - R. Caorsi - S. Volpi - A. Grossi - I. Ceccherini - M. Gattorno
- P1028** **NON-AMYLOID KIDNEY DISEASES AND AUTOINFLAMMATORY DISEASES: REPORT OF 20 CASES AND LITERATURE REVIEW** / C. Borocco - I. Kone-Paut - A. Belot - M. Desjonqueres - A. Karras - C. Miceli-Richard - B. Moulin - T. Ulinski - J.-J. Boffa - D. Buob - G. Grateau - S. Georgin-Lavialle\*
- P1029** **THE TRANSITION FROM PEDIATRICS TO ADULTHOOD IN AUTO-INFLAMMATORY DISEASES: COMPARISON OF 2 TRANSITION STRATEGIES AMONG 72 PATIENTS** / S. Georgin-Lavialle\* - P. Quartier - B. Bader-Meunier - K. Stankovic Stojanovic - V. Avellino - S. Deshayes - I. Melki - A. Belot - I. Kone-Paut - G. Grateau - V. Hentgen on behalf of Transition working group, National reference center for autoinflammatory diseases and AA amyloidosis
- P1030** **THE LONGITUDINAL EUROFEVER PROJECT: AN UPDATE ON ENROLLMENT** / I. Gueli\* - M. Finetti - F. De Benedetti - J. Anton Lopez - M. Alessio - J. Frenkel - L. Cantarini - R. Gallizzi - J. Sanchez Manubens - M. Cattalini - E. Papadopoulou-Alataki - R. Cimaz - D. Rigante - A. N. Olivieri - P. Dolezalova - A. Martini - N. Ruperto - M. Gattorno
- P1031** **DIGITAL BRACHIAL INDEX TESTING AS A NONINVASIVE TOOL IN DIAGNOSING PERIPHERAL VASCULAR DISEASE IN DADA2** / P. M. Hoffmann\* - D. L. Stone - K. Barron - C. Cudrici - A. Brofferio - A. Jones - T. Romeo - I. Aksentijevich - D. L. Kastner - A. K. Ombrello
- P1032** **THE EXPANDING CLINICAL AND LABORATORY SPECTRUM OF PAPA SYNDROME: THE NIH COHORT** / P. Hoffmann\* - A. K. Ombrello - D. L. Stone - K. Barron - A. Jones - T. Romeo - M. Nehrebecky - J. Chae - I. Aksentijevich - D. L. Kastner
- P1033** **PSEUDODOMINANT INHERITANCE OF BEHÇET-LIKE AUTOINFLAMMATORY DISEASE ASSOCIATED WITH TNFAIP3 (A20) AND MEFV MUTATIONS IN A TURKISH FAMILY WITH FAMILIAL MEDITERRANEAN FEVER** / N. Horita\* - A. Gul - I. Aksentijevich - D. Kastner - E. Remmers

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- P1034**      **SYSTEMATIC REVIEW OF BIOLOGICAL TREATMENT OF DEFICIENCY OF INTERLEUKIN-36 RECEPTOR ANTAGONIST (DITRA) IN CHILDREN AND ADOLESCENTS** / T. Hospach\* - F. Glowatzki - F. Blankenburg - D. Conzelmann - C. Stirnkorb - S. Müllerschön - P. von den Driesch - L. Koehler - M. Rohlf - C. Klein - F. Hauck
- P1035**      **FAMILIAL MEDITERRANEAN FEVER IN SLOVAKIA – CLINICAL AND GENETIC CHARACTERISTICS OF SLOVAK COHORT** / M. Jesenak\* - L. Kapustova - K. Hrubiskova - T. Dallos - P. Banovcin
- P1036**      **DIAGNOSTIC CRITERIA FOR PROTEASOME-ASSOCIATED AUTOINFLAMMATORY SYNDROMES (PRAASS) INCLUDING NAKAJO-NISHIMURA SYNDROME, JMP SYNDROME AND CANDLE SYNDROME** / N. Kanazawa\* - H. Ida - N. Kinjo - T. Ishikawa - R. Nishikomori
- P1037**      **SHORT TERM FOLLOW-UP RESULTS OF CHILDREN WITH FAMILIAL MEDITERRANEAN FEVER AFTER CESSATION OF COLCHICINE: IS IT POSSIBLE TO QUIT?** / A. Tanatar - H. E. Sönmez - S. G. Karadağ\* - M. Çakan - N. Aktay Ayaz
- P1038**      **PERIODIC FEVER SYNDROMES: A YEAR FOLLOW-UP OF A TERTIARY PEDIATRIC RHEUMATOLOGY OUTPATIENT CLINIC IN TURKEY** / N. Aktay Ayaz - S. G. Karadağ\* - H. E. Sönmez - A. Tanatar
- P1039**      **ASSESSING FRENCH LIBERAL PEDIATRICIANS AWARENESS AND REFERRAL FOR RECURRENT FEVER SYNDROMES: THE FIREVILLE SURVEY** / V. Koskas\* - R. Assathiany - S. Hubinois - C. Levy - M. Koskas - I. Koné-Paut
- P1040**      **CRIMEAN TATARS IS NEW TARGET NATIONALITY FOR THE FAMILIAL MEDITERRANEAN FEVER** / O. Zhogova - N. Lagunova - S. Ivanovskiy - E. Suspitsin - M. Kostik\*
- P1041**      **NEW VARIANT IN THE IL1RN-GENE ASSOCIATED WITH LATE ONSET AND ATYPICAL PRESENTATION OF DIRA – FOLLOW-UP** / J. B. Kuemmerle-Deschner\* - K. Reicherter - S. Schlipf - S. Hansmann - A. Hospach - I. Tsifikas - X. Liu - S. Benseler - A. Weber
- P1042**      **EFFICACY OF ANTI-IL-1 TREATMENT IN FAMILIAL MEDITERRANEAN FEVER: A SINGLE-CENTER EXPERIENCE** / T. Kurt\* - H. O. Basaran - F. Aydın - N. Uncu - B. Celikel Acar
- P1043**      **ERYSIPELAS-LIKE ERYTHEMA AS THE PRIMARY GRIPE OF FMF** / O. Kuru\* - A. K. Cengiz
- P1044**      **ANALYSIS OF NEW REFERRALS TO A SPECIALIST UK ADULT AUTOINFLAMMATORY DISEASE SERVICE** / S. Ugurlu - P. N. Hawkins - C. Papadopoulou - T. Rezk - D. Rowczenio - H. J. Lachmann\*
- P1045**      **APPLICATION OF AUTOINFLAMMATORY DISEASE DAMAGE INDEX (ADDI) TO OTHER AUTOINFLAMMATORY DISEASES IN A TERTIARY REFERRAL HOSPITAL** / M. Lopez\* - E. Moreno
- P1046**      **VASCULITIS IN AUTOINFLAMMATORY DISEASES IN A TERTIARY HOSPITAL** / R. M. Alcobendas - S. M. Loza\* - P. F. Fraga - C. U. Gascon - C. Fervenza - A. Remesal
- P1047**      **DIAGNOSIS AND TREATMENT OF PERIODIC FEVERS: A SINGLE CENTRE EXPERIENCE** / P. McNaughton\* - J. Peake - B. Whitehead - S. H. Lum - K. Preece
- P1048**      **MULTIFOCAL OSTEOMYELITIS REVEALING A PSTPIP1- ASSOCIATED MYELOID-RELATED PROTEINEMIA INFLAMMATORY (PAMI) SYNDROME: CASE REPORT AND REVIEW OF THE LITERATURE** / M. Mejbrí\* - K. Theodoropoulou - M. Hofer

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## POSTER VIEWING - MONOGENIC AUTOINFLAMMATORY DISEASES (BASIC SCIENCE)

- P1049** INVESTIGATION OF INFLAMMASOME COMPONENTS IN THE PROCESS OF CELL MIGRATION IN FMF PATIENTS / T. H. Akbaba\* - Z. Y. Akkaya-Ulum - S. Demir - Z. Tavukcuoglu - S. Ozen - B. Balci-Peynircioglu
- P1050** CELL MIGRATION DEFECT IN HYPERIMMUNOGLOBULIN D SYNDROME / T. H. Akbaba\* - S. Demir - Z. Y. Akkaya-Ulum - S. Ozen - B. Balci-Peynircioglu
- P1051** POSSIBLE REGULATORY EFFECTS OF MIRNAS IN THE PATHOGENESIS OF SYSTEMIC AUTO INFLAMMATORY DISEASES, FROM THE PERSPECTIVE OF FAMILIAL MEDITERRANEAN FEVER / Z. Y. Akkaya-Ulum - Z. Tavukcuoglu - E. D. Batu-Akal - T. H. Akbaba\* - H. E. Sonmez - B. Balci-Peynircioglu - S. Ozen
- P1052** MIR-197 REGULATES INFLAMMATION IN MONOCYTES AND SYNOVIAL FIBROBLASTS BY TARGETING IL1R1 / Y. Z. Akkaya Ulum\* - Z. Tavukcuoglu - T. H. Akbaba - E. Yilmaz - B. Balci-Peynircioglu
- P1053** COMPARISON OF FMF PATIENTS WITH AGE OF ONSET BEFORE 20 VERSUS 40 YEARS AND OVER / O. Aydin\* - S. Ugurlu - H. Ozdogan
- P1054** WHAT HISTORY OF APPENDECTOMY WILL TELL US ABOUT THE COURSE OF FAMILIAL MEDITERRANEAN FEVER IN ADULTHOOD? / E. Bodakçi\* - N. S. A. BİLGE - N. Ataş - B. Armağan - H. Satiş - A. Sarı - H. Babaoğlu - G. K. Yardımcı - R. B. Salman - L. Kılıç - M. A. Öztürk - B. Göker - S. Haznedaroğlu - U. Kalyoncu - A. Tufan - T. Kaşifoğlu
- P1055** COMPLEMENT ENDORSE THE PATHOGENESIS IN AUTOINFLAMMATION / J. Brunner\* - W. Posch - D. Wilflingseder
- P1056** GENERATION OF ADA2 GENETIC KNOCKOUT IN A MYELOID CELL LINE USING CRISPR/CAS9 GENOME EDITING: AN IN VITRO CELL LINE MODEL TO STUDY DADA2 / M. S. Casimir\* - Y. Hong - P. Brogan - D. Eleftheriou
- P1057** PROMISING BLOOD TEST FOR DIAGNOSIS AND TREATMENT OPTIONS IN PATIENTS WITH SUSPECTED CHRONIC AUTO-INFLAMMATORY SYNDROMES / A.-L. N. Chetaille\* - N. Pagé - M.-P. Longchamps - L. Bessette - L. Michou - P. Fortin - P. Tessier - M. Pelletier
- P1058** SPECIFIC DYSBIOSIS ASSOCIATED WITH FAMILIAL MEDITERRANEAN FEVER COMPLICATED OR NOT WITH AA AMYLOIDOSIS / S. Deshayes\* - S. Fellahi - J.-P. Bastard - J.-M. Launay - J. Callebert - T. Fraisse - D. Buob - J.-J. Boffa - I. Giurgea - C. Dupont - S. Jegou - M. Straube - A. Karras - A. Aouba - G. Grateau - H. Sokol - S. Georgin-Lavialle on behalf of AA amyloidosis Study Group
- P1059** PROINFLAMMATORY CYTOKINES INDUCED BY PBMCS FROM A PATIENT WITH A NLRP1 VARIANT SHOWING SEVERE CORNEAL INTRAEPITHELIAL DYSKERATOSIS / T. Herlin\* - S. E. Jørgensen - C. Høst - M. Christiansen - D. A. Larsen - T. H. Mogensen
- P1060** GAIN-OF-FUNCTION MUTATION OF NOD2 IMPAIRS ITS LIGAND SPECIFIC IMMUNE RESPONSES IN BLAU SYNDROME PATIENT-DERIVED IPS CELLS / N. Kambe\* - N. T. M. Ly - M. K. Saito - H. Okamoto
- P1061** THE ROLE OF DNA METHYLATION FOR DISEASE SEVERITY IN PATIENTS WITH HETEROZYGOUS MUTATIONS IN THE MEDITERRANEAN FEVER GENE MEFV / J. Krainer\* - W. Pulverer - D. Foell - S. Özen - A. Weinhäusel
- P1062** DIFFERENTIAL KINETIC OF ACTIN POLYMERIZATION IN NEUTROPHIL FROM PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER / D. Poghosyan - A. Martirosyan - N. Mkrtychyan - S. Margaryan - S. Ghonyan - G. Amaryan - G. Manukyan\*

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- P1063** **CARD15 MUTATIONS IN AN INDIAN COHORT OF BLAU SYNDROME** / A. Rawat\* - D. Suri - R. Kumrah - S. Bhattad - S. Guleria - V. Pandiarajan - A. Jindal - A. Gupta - I. Ceccherini - M. Gattorno - S. Singh
- P1064** **DIFFERENTIAL ACTIVATION OF THE PYRIN INFLAMMASOME IN MONOCYTES AND MACROPHAGES PREDICTS THE PATHOLOGICAL SIGNIFICANCE OF MEFV VARIANTS IN FAMILIAL MEDITERRANEAN FEVER (FMF) PATIENTS** / T. Shiba\* - T. Tanaka - H. Ida - M. Watanabe - H. Nakaseko - M. Osawa - H. Shibata - K. Izawa - T. Yasumi - Y. Kawasaki - M. K. Saito - J. Takita - T. Heike - R. Nishikomori
- P1065** **CROSS-TALK BETWEEN TYPE I IFN PATHWAY AND TLR SENSING IN DNASE2 MUTATED FIBROBLASTS** / A. Tesser\* - E. Piscianz - V. Boz - G. Piperno - F. Benvenuti - A. Tommasini
- P1066** **AUTO-INFLAMMATORY DISEASES (SAIDS) IN WESTERN SWITZERLAND: A DESCRIPTIVE STUDY THROUGH THE JIRCOHORTE PLATFORM** / L. Tosetti\* - M. Mejbri - A. Theodoropoulou - M. Hofer
- P1067** **FAMILIAL MEDITERRANEAN FEVER ASSOCIATED INFERTILITY AND UNDERLYING FACTORS** / N. Atas - B. Armagan - E. Bodakci - T. Kasifoglu - H. Satis - A. Sari - N. S. Y. Bilge - H. Babaoglu - G. K. Yardimci - R. B. Salman - L. Kilic - M. A. Ozturk - B. Goker - S. Haznedaroglu - U. Kalyoncu - A. Tufan\*
- P1068** **MEFV-MUTANT INDUCED PLURIPOTENT STEM-CELLS HAVE REDUCED ANTI-INFLAMMATORY PROPERTIES** / K. Kessel - C. Kessel - N. Ludwig - T. Weinhage - D. Foell - H. Wittkowski\*

### POSTER VIEWING - MONOGENIC AUTOINFLAMMATORY DISEASES (CLINICAL)

- P1069** **THREE NOVEL CASES OF LATE-ONSET CRYOPYRIN-ASSOCIATED PERIODIC SYNDROMES DUE TO SOMATIC NLRP3 MOSAICISM** / A. Mensa-Vilaró - M. T. Bosque - E. Gómez de la Fuente - N. Palmou - L. Martín-Penagos - C. Delgado - S. Plaza - R. Lara - M. C. Antón - H. Martínez-Banaclocha - J. J. Martínez-García - J. Yagüe - M. Á. González-Gay - P. Pelegrin - J. I. Arostegui\*
- P1070** **THE NLRP3 PA441V MUTATION IN CRYOPYRIN-ASSOCIATED PERIODIC SYNDROME PATHOGENESIS: FUNCTIONAL CONSEQUENCES, PHENOTYPE-GENOTYPE CORRELATIONS AND EVIDENCE FOR A FOUNDER EFFECT** / E. Assrawi\* - F. Awad - C. Jumeau - S. Odent - V. Despert - G. Cam - A. Perdriger - C. Louvrier - L. Cobret - B. Copin - P. Duquesnoy - W. Piterboth - C. Le Jeune - S. Georgin-Lavialle - G. Gâteau - M. Legendre - I. Giurgea - S. A. Karabina - S. Amselem
- P1071** **THE ANALYSIS OF IL-36RA STRUCTURAL DYNAMICS IMPROVES PATHOGENICITY PREDICTIONS FOR IL36RN VARIANTS OBSERVED IN GENERALISED PUSTULAR PSORIASIS** / C. Davan-Wetton - N. K. Hassi - J. C. Ng - F. Fraternali - F. Capon\*
- P1072** **SPONDYLOENCHONDRODYSPLASIA AND SYSTEMIC LUPUS ERYTHEMATOSUS: CASE REPORT OF TWO SIBLING** / B. Kara - A. Cefle\* - O. Kasapcopur - A. Sakarya Gunes
- P1073** **ADA2 DEFICIENCY WITHOUT ADA2 MUTATIONS EXPLAINED BY A STRUCTURAL HOMOZYGOUS VARIATION IN 22Q11.1** / A. Grossi\* - F. Garbarino - R. Caorsi - R. Cusano - M. Rusmini - F. Penco - F. Schena - R. A. Podda - P. Uva - I. Ceccherini - M. Gattorno
- P1074** **EVALUATION OF THE ROLE OF SAA1, SAA2 AND SAA4 GENES IN AA AMYLOIDOSIS** / C. Jumeau\* - C. Louvrier - E. Assrawi - F. Awad - L. Cobret - S. Georgin-Lavialle - C. Cazorla - P. Duquesnoy - J.-S. Rech - W. Piterboth - F. Dastot-Le Moal - B. Copin - D. Buob - G. Gâteau - J.-F. Bernaudin - M. Legendre - S.-A. Karabina - I. Giurgea - S. Amselem
- P1075** **AN INTEGRATED DATA SHARING AND ANALYSIS APPLICATION FOR SYSTEMIC AUTOINFLAMMATORY DISEASES** / B. P. Kant\* - K. J. van der Velde - M. K. Slofstra - J. Frenkel - A. Weinhäusel - I. Touitou - D. Föll - J. I. Aróstegui - S. Ozen - M. Gattorno - M. E. van Gijn - M. A. Swertz on behalf of the INSAID consortium

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- P1076** **COMPARATIVE ANALYSIS OF REPORTED 54 HA20 CASES AND 520 JAPANESE BEHÇET'S DISEASE PATIENTS** / Y. Kirino\* - N. Tsuchida - Y. Soejima - H. Nakajima
- P1077** **NOVEL MISSENSE MUTATION OF TNFAIP3 GENE IN A FAMILY WITH A20 HAPLOINSUFFICIENCY** / C. Borocco - G. Sarabay - G. Boursier - S. Artru - H. Palluy - I. Touitou - I. Kone-Paut\*
- P1078** **DEVELOPMENT AND CLINICAL APPLICATION OF A TARGETED NEXT GENERATION SEQUENCING GENE PANEL FOR MONOGENIC AUTOINFLAMMATORY DISEASES OF THE CNS** / D. McCreary\* - E. O. Omoyinmi - Y. Hong - C. Mulhern - C. Papadopoulou - M. Al Obaidi - K. Mankad - K. Gilmour - C. Hemingway - P. Brogan - D. Eleftheriou
- P1079** **NEXT GENERATION SEQUENCING ANALYSIS OF AN NLRP3 MUTATION NEGATIVE CAPS COHORT** / S. Melo Gomes\* - E. Omoyinmi - J. Arostegui - Y. Hong - N. Klein - P. Brogan
- P1080** **A RECESSIVE MUTATION IN NLRC4 (A160T) CAUSES AUTOINFLAMMATION AND IMMUNEDYSREGULATION, AND PREDISPOSES TO ULCERATIVE COLITIS IN HETEROZYGOUS CARRIERS** / L. O. Mendonca\* - A. Steiner - A. Pontillo - I. Ceccherini - F. Caroli - A. Grossi - J. Moecking - F. Moghaddas - M. Gattorno - S. Masters
- P1081** **THE AUSTRALIAN AUTOINFLAMMATORY DISEASES REGISTRY (AADRY): A STREAMLINED APPROACH TO THE GENETIC AND IMMUNOLOGICAL EVALUATION OF MONOGENIC AUTOINFLAMMATORY DISEASES** / F. Moghaddas\* - J. Harris - D. Vekic - H. Cassandra - D. J. Calleja - N. Adib - J. Akikusa - R. Allen - D. Andrews - V. Bryant - G. Cains - J. Chaitow - B. Christensen - D. Coman - M. Cook - A. Cox - J. A. Douglass - P. Gowdie - L. Hosking - M. F. Hunter - P. Kubler - S. J. Lee - J. Munro - S. Ojaimi - W. Renton - D. Singh-Grewal - C. Slade - J. Smart - G. Tiller - B. Whitehead - J. Woods - P. Wu - S. Mehr - I. P. Wicks - S. L. Masters on behalf of AADRY
- P1082** **GENOTYPIC DIVERSITY OBSERVED WITHIN A LARGE COHORT OF ARMENIAN PATIENTS WITH LATE-ONSET FAMILIAL MEDITERRANEAN FEVER** / G. Kriegshäuser - H. Hayrapetyan - S. Atoyan - S. Nemeth - C. Oberkanins\* - T. Sarkisian
- P1083** **GENETIC POLYMORPHISM OF FAMILIAL MEDITERRANEAN FEVER IN GEORGIA - A SNAPSHOT** / K. Pagava\* - H. Phagava - D. Tatoshvili
- P1084** **EXPLORATORY STUDY OF MYD88 L265P AND CLONAL HAEMATOPOIESIS IN 30 PATIENTS WITH SCHNITZLER SYNDROME, AN ACQUIRED SYSTEMIC AUTOINFLAMMATORY DISORDER** / S. Pathak\* - D. Rowczenio - R. Owen - G. D. Doody - D. Newton - C. Taylor - C. Cargo - P. Hawkins - K. Krause - H. Lachmann - S. Savic
- P1085** **E148Q MUTATION IN FAMILY WITH FAMILIAL MEDITERRANEAN FEVER** / T. F. Sarkisian\* - H. Hayrapetyan - A. Yeghiazaryan - R. Karakhanyan
- P1086** **FAMILIAL MEDITERRANEAN FEVER: CLINICAL SIGNIFICANCE OF E148Q AND P369S** / T. F. Sarkisian\* - H. Hayrapetyan - S. Atoyan - A. Yeghiazaryan - G. Shahsuvaryan
- P1087** **SEVERE PROGNOSIS OF LATE ONSET CAPS ASSOCIATED WITH AGE RELATED CLONAL HEMATOPOIESIS** / Y. Shinar\* - R. Cohen - V. Marcu - I. Goldstein - M. Nagar - G. Slobodin - I. Ben-Zvi - M. Rozenbaum
- P1088** **CNV ANALYSIS FROM TARGETED NGS DATA: CASE REPORT ON A PATIENT WITH A LARGE DELETION IN THE NLRP12 GENE** / B. Bangol - K. Starz\* - M. Ziegler - D. Becker - J. Schiller - S. Eilitz - H.-G. Klein
- P1089** **POPULATION BASED STUDY OF FREQUENCY OF FMF GENE MUTATION CARRYING IN ARMENIA** / A. Tadevosyan\* - T. Avagyan - A. Budumyan - H. Hayrapetyan - G. Amaryan



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- P1090** **WHOLE GENOME LINKAGE AND EXOME SEQUENCING ANALYSIS IN A CRMO FAMILY** / D. Yavuz - S. Yuksel - E. Tahir Turanli\*
- P1091** **DETECTION OF A RARE VARIANT ON PSTPI1 GENE THROUGH THREE GENERATIONS IN A TURKISH FAMILY WITH SYSTEMIC AUTOINFLAMMATORY DISEASE** / M. Ö. Önen - S. Ugurlu - A. Balamir - E. Tahir Turanli\* - H. Ozdogan

### POSTER VIEWING - DERMATOLOGY AND AUTOINFLAMMATION

- P1092** **LONG-TERM TREATMENT WITH USTEKINUMAB FOR GENERALIZED PUSTULAR PSORIASIS IN A CHILD WITH HETEROZYGOUS MUTATION IN THE IL36RN AND TYK2 GENE** / T. Herlin\* - M. Christiansen - M. Glerup - S. E. Jørgensen - C. Høst - J. E. Veirum - D. A. Larsen - L. Iversen - M. Glerup - B. Mahler - T. H. Mogensen

### NON-MONOGENIC SAIDS (CLINICAL)

- P1093** **PERFORMANCE OF NEWLY PROPOSED PERIODIC FEVER, APHTHOUS STOMATITIS, PHARYNGITIS AND CERVICAL ADENITIS (PFAPA) SYNDROME CRITERIA IN REGIONS ENDEMIC FOR FAMILIAL MEDITERRANEAN FEVER (FMF)** / A. Adrovic\* - M. Yildiz - N. Gucuyener - I. Ulkersoy - M. Kanber - S. Sahin - O. Koker - K. Barut - O. Kasapcopur
- P1094** **SPINE INVOLVEMENT IN PATIENTS WITH NON-BACTERIAL MULTIFOCAL OSTEOMYELITIS** / D. Alekseev\* - I. Nikishina - D. Zubkov - A. Smirnov - O. Kostareva
- P1095** **DNASE1L3 VARIANT IN HYPOCOMPLEMENTEMIC URTICARIAL VASCULITIS SYNDROME IDENTIFIES A DIFFERENT CLINICAL PHENOTYPE** / M. Ranalli - C. Passarelli - V. Messina - M. Pardeo - E. Sacco - A. Insalaco - M. Vivarelli - F. De Benedetti - C. Bracaglia\*
- P1096** **A CASE OF IDIOPATHIC RECURRENT PERICARDITIS TREATED WITH ANAKINRA BUT UNRESPONSIVE TO CANAKINUMAB** / F. Ricci - S. Ghetti - R. Razza - C. Dallavilla - G. Verzura - A. Manerba - M. Cattalini\*
- P1097** **EFFICACY OF CANAKINUMAB AS FIRST-LINE BIOLOGIC AGENT IN ADULT-ONSET STILL'S DISEASE** / G. Cavalli\* - A. Tomelleri - C. Campochiaro - E. Baldissera - G. De Luca - L. Dagna
- P1098** **CARRA AUTOINFLAMMATORY DISEASE NETWORK DEVELOPMENT- IMPROVING CARE AND FOSTERING RESEARCH IN SYSTEMIC AUTOINFLAMMATORY DISEASES (SAIDS)** / J. Cherian\* - T. L. Wampler Muskardin - M. Twilt - R. M. Laxer - J. S. Hausmann - C. Yildirim-Toruner - M. J. Gutierrez - N. Saad - G. S. Schulert - E. J. Propst - G. Licameli - A. Thatayatikom - G. Amarilyo - L. Harel - I. C. Michelow - P. F. Wright - Y. Stepanovskiy - L. N. Moorthy - K. Durrant - P. J. Ferguson - L. B. Tucker - F. Dedeoglu - S. K. Lapidus on behalf of CARRA Autoinflammatory Network Development Work Group
- P1099** **THE HELIOS (HACETTEPE UNIVERSITY ELECTRONIC RESEARCH FORMS) REGISTRY: USE OF BIOLOGIC DRUGS IN AUTOINFLAMMATORY DISEASES** / S. Demir\* - E. D. Batu - F. Akal - E. Sağ - U. Kaya Akca - E. Arslanoğlu Aydın - E. Aliyev - K. Yüksel - A. Keskin - Y. Bilginer - S. Ozen
- P1100** **PEDIATRIC BEHÇET'S DISEASE WITH SINUS VENOUS THROMBOSIS: THREE CENTER EXPERIENCE FROM TURKEY** / S. Demir\* - C. Açarı - Ö. Basaran - E. Sağ - Y. Bilginer - S. E. Ünsal - S. Ozen
- P1101** **PFAPA - THE IRISH EXPERIENCE IN A TERTIARY AUTOINFLAMMATORY CLINIC** / A.-L. Hawke\* - J. MacMahon - E. J. MacDermott - K. Butler - R. Leahy - P. Gavin - O. Killeen

## MONDAY 01 APRIL 2019

### POSTER VIEWING - SYSTEMIC-ONSET JIA AND AOSD

- P1102** HEPATITIS A VIRUS VACCINATION IN AUTOINFLAMMATORY DISEASES UNDER CANAKINUMAB AND TOCILIZUMAB TREATMENT / K. Barut\* - A. Adrovic - S. Sahin - M. Yıldız - O. Koker - G. Yalcin - O. F. Beser - B. Kocazeybek - P. Yuksel - O. Kasapcopur
- P1103** RISK SCORE OF MACROPHAGE ACTIVATION SYNDROME IN PATIENTS WITH SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS / S. Carbogno - D. Pires Marafon - G. Marucci - M. Pardeo - A. Insalaco - V. Messina - E. Sacco - F. De Benedetti - C. Bracaglia\*
- P1104** ADULT ONSET STILL'S DISEASE COMPLICATED WITH HAEMOPHAGOCYtic SYNDROME: SUCCESSFUL TREATMENT WITH IVIG AND TOCILIZUMAB / A. Cefle\* - F. Tuncer - A. Yazici
- P1105** DRUG RETENTION RATE AND PREDICTIVE FACTORS OF DRUG SURVIVAL FOR INTERLEUKIN-1 INHIBITORS IN SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS / C. Gaggiano\* - J. Sota - A. Insalaco - R. Cimaz - M. Alessio - M. Cattalini - R. Gallizzi - M. C. Maggio - G. Lopalco - F. La Torre - C. Fabiani - M. Pardeo - A. N. Olivieri - P. Sfriso - C. Salvarani - S. Grosso - C. Bracaglia - F. De Benedetti - D. Rigante - L. Cantarini
- P1106** CLINIC-LABORATORY PROFILE OF MACROPHAGE ACTIVATION SYNDROME IN CHILDREN WITH SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS / S. Guleria\* - J. Nameirakpam - A. Gummadi - A. Gupta - A. Rawat - P. Bhatia - D. Suri - S. Singh
- P1107** SERUM SOLUBLE CD25: AN USEFUL BIOMARKER OF MACROPHAGE ACTIVATION SYNDROME IN SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS / S. Guleria\* - A. Gupta - A. Rawat - P. Bhatia - A. Sharma - D. Suri - S. Singh
- P1108** PATIENT WITH CASP1 VARIANT WITH SEVERE SYSTEMIC JIA AND RECURRENT MAS TREATED BY DUAL BLOCKADE WITH ANAKINRA AND TOCILIZUMAB / T. Herlin\* - S. E. Jørgensen - C. Høst - M. Christiansen - C. Høst - D. A. Larsen - M. Glerup - T. H. Mogensen - B. Mahler
- P1109** VALIDATION AND CLINICAL APPLICATION OF MRP8/14 SERUM LEVELS AS BIOMARKER FOR THE DIAGNOSIS OF SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS IN FEVER OF UNKNOWN ORIGIN / C. Pretzer - M. Miranda-Garcia - R. Berendes - G. Horneff - J. Kuemmerle-Deschner - G. Ganser - H.-I. Huppertz - K. Minden - J.-P. Haas - A. Jansson - M. Borte - C. Schuetz - P. Oommen - M. Frosch - C. Kessel - B. Schlueter - A. Richter-Unruh - H. Wittkowski - J. Roth - D. Foell - D. Holzinger\*
- P1110** DO JUVENILE IDIOPATHIC ARTHRITIS PATIENTS WITH SYSTEMIC ONSET IN THE RISK OF LOW ANTI-VACCINE ANTIBODY: THE PRELIMINARY REPORT? / N. Lubimova - O. Goleva - I. Fridman - M. Dubko - V. Masalova - L. Snegireva - E. Isupova - E. Gaidar - A. Santimov - O. Kalashnikova - S. Kharit - M. Kostik\*
- P1111** HAVE PATIENTS WITH JUVENILE IDIOPATHIC ARTHRITIS PATIENTS WITH SYSTEMIC ONSET PARTICULAR FEATURES FOR MONOCYCLIC COURSE? / M. Kostik\* - M. Romyantseva - E. Isupova - I. Chikova - M. Dubko - V. Masalova - L. Snegireva - T. Kornishina - T. Likhacheva - E. Gaidar - A. Santimov - O. Kalashnikova
- P1112** SINGLE CENTER EXPERIENCE OF BIOLOGICAL THERAPY IN PATIENTS WITH SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS ASSOCIATED WITH MACROPHAGE ACTIVATION SYNDROME / M. I. Kaleda - I. P. Nikishina\*

### POSTER VIEWING - UNUSUAL OR UNSOLVED CASE REPORTS

- P1113** BEHÇET DISEASE IN PEDIATRICS; A SOLITARY SIGN CAN BE ENOUGH / H. M. Abd El-Lateef\*

## MONDAY 01 APRIL 2019

- P1114** CHILDHOOD PYODERMA GANGRENOSUM: DIAGNOSTIC CHALLENGES AND MANAGERMENTS OF TWO CASES FROM LIBYA / A. Abushhaiwia\* - N. Abushhiwa - M. Kamel - L. Sabei - M. Zletni - H. Dawi
- P1115** A GIRL WITH A DEVELOPMENTAL DELAY AND LUPUS-LIKE PHENOTYPE ASSOCIATED WITH BOTH NEUROBLASTOMA AND AN MDA5 GAIN-OF-FUNCTION MUTATION (AICARDI-GOUTIÈRES SYNDROME 7, AGS7) / S. Berg\* - D. McCreary - G. Drake af Hagelsrum - N. Björkander - D. Eleftheriou
- P1116** COLD URTICARIA ASSOCIATED WITH A NOVEL PHOSPHOLIPASE-GAMMA-C2 (PLCY2) MUTATION / H. Bonnekoh\* - N. A. Mahnke - J. Scheffel - M. Maurer - K. Krause
- P1117** MISSENSE VARIANTS IN SERPINF1 ARE ASSOCIATED WITH A NOVEL AUTO-INFLAMMATORY DISEASE PRESENTING WITH PERSISTENT FEVER AND MULTIFOCAL NEUTROPHILIC PERIOSTITIS / M. Bustaffa\* - M. Rusmini - M. Cattalini - S. Signa - R. Papa - A. Grossi - R. Caorsi - S. Volpi - P. Picco - A. Plebani - A. Ravelli - N. Zoppi - M. Ritelli - M. Colombi - M. Di Rocco - I. Ceccherini - M. Gattorno
- P1118** LATE-ONSET SAPHO SYNDROME REVEALING CHRONIC MYELOMONOCYtic LEUKEMIA / L. Damian\* - S. Grad - F. Mustatea - B. Balan - G. Cobzac - C. Bolosiu - L. Bene - A. Trifa - D. Dima - A. Albu
- P1119** BLOODY STOOLS AND CRYOPYRIN-ASSOCIATED PERIODIC SYNDROME (CAPS) - ASSOCIATION OR COINCIDENCE? / M. Correia Marques - J. S. Hausmann - E. Anderson - F. Dedeoglu\*
- P1120** THE CHALLENGE OF TREATING PULMONARY VASCULITIS IN BEHÇET'S DISEASE: TWO PEDIATRIC CASES AND LITERATURE REVIEW / S. Demir\* - E. Sağ - Ü. Kaya Akca - T. Hazırolan - Y. Bilginer - S. Ozen
- P1121** LATE DIAGNOSIS OF CRYOPYRIN-ASSOCIATED PERIODIC SYNDROME - WITHOUT GENE CONFIRMATION - IS THERE ANY OTHER EXPLANATION? / R. Faria\* - A. Lamas - A. Campar - G. Carvalheiras
- P1122** LATE ONSET OF TUMOR NECROSIS FACTOR RECEPTOR ASSOCIATED PERIODIC SYNDROME (TRAPS) DUE TO SOMATIC MOSAICISM / R. Faria\* - L. Magalhães - D. Oliveira - A. Marinho - A. Campar
- P1123** HYPERZINCAEMIA AND HYPERCALPROTECTINEMIA SYNDROME: MORE THAN JUST AUTOINFLAMMATION? / A. Uva - C. Bracaglia - S. Federici\* - C. Celani - M. Pardeo - C. Kessel - F. De Benedetti - A. Insalaco
- P1124** INHERITED DEFICIT OF PROTEOGLYCAN MIMICKING SEPTIC ARTHRITIS / A. Florio\* - R. Papa - R. Caorsi - A. Consolaro - T. Rinne - R. Gastaldi - A. Ravelli - M. Gattorno - P. Picco
- P1125** LRBA DEFICIENCY IN A PATIENT WITH CHRONIC ARTHRITIS / S. G. Karadağ\* - A. Tanatar - M. Çakan - H. E. Sönmez - F. Çakmak - N. Aktay Ayaz
- P1126** UNUSUAL CASE OF RECURRENT PERIODIC MACROPHAGE ACTIVATION / K. Khawaja\* - T. T. Hen
- P1127** AA AMYLOIDOSIS IN RECIPIENTS OF CADAVERIC DERIVED PITUITARY GROWTH HORMONE - POTENTIAL EFFECT OF AN AMYLOID ENHANCING FACTOR? / H. J. Lachmann\* - D. Rowczenio - J. A. Gilbertson - H. Trojer - I. Noor - T. Lane - J. D. Gillmore - A. D. Wechalekar - P. N. Hawkins
- P1128** PROGRESSIVE NECROTIC ULCERATIVE LESIONS: CHALLENGING DIAGNOSIS? / A. Mauro\* - F. Orlando - M. Tardi - R. Rega - M. Luigi - R. Sottile
- P1129** AN UNDEFINED SYSTEMIC AUTOINFLAMMATORY DISEASE COMPLICATED BY MAS OR AN UNIDENTIFIED PRIMARY HLH? / R. Naddei\* - F. Orlando - C. Porfito - T. Lastella - M. Amico - M. Alessio

## MONDAY 01 APRIL 2019

### POSTER VIEWING - ALLIED HEALTH PROFESSIONALS

**P1130** TOWARDS EUROPEAN HARMONISATION OF HEALTH CARE FOR PATIENTS WITH RARE IMMUNE DISORDERS: ERN RITA RESULT FROM THE REGISTRIES SURVEY / R. Papa\* - A. Cant - C. Klein - M. Little - N. M. Wulfraat - M. Gattorno - N. Ruperto on behalf of RITA ERN Council

**16:00 - 17:00** **ORAL COMMUNICATIONS – CLINICAL** **MAESTRALE ROOM**  
*Chairpersons: Véronique Hentgen (FR) & Seza Ozen (TR)*

**16:00** **O09 - The clinical spectrum of the deficiency of adenosine deaminase 2 (DADA2) continues to expand**  
 Karyl Barron (US)

**16:08** **O10 - Serum S100A8/A9 (Calprotectin) in Familial Mediterranean Fever and carriers of MEFV mutations does not correlate with disease activity**  
 Helen Lachmann (UK)

**16:16** **O11 - PAPA syndrome: novelties from the Eurofever registry**  
 Roberta Caorsi (IT)

**16:24** **O12 - New classification criteria for recurrent autoinflammatory diseases applied to an independent cohort: experience from the JIR cohort database**  
 Glory Dingulu (FR)

**16:32** **O13 - Impaired platelet functions in patients treated with colchicine**  
 Selcan Demir (TR)

**16:40** **O14 - Autoinflammatory disorders in patients with myelodysplastic syndrome: the role of distinctive karyotypes and somatic mutations**  
 Mark Kacar (UK)

**16:48** **O15 - A novel MEFV mutation associated with an autosomal dominant FMF complicated by AA amyloidosis in a large British kindred**  
 Dorota Rowczenio (UK)

**16:00 - 17:00** **ORAL COMMUNICATIONS – IMMUNOLOGY** **SCIROCCO-LIBECCIO ROOM**  
*Chairpersons: Fabio Martinon (CH) & Seth Masters (AU)*

**16:00** **O16 - Pysin inflammasome dysregulation in FMF patients: implication for a fast diagnostic test**  
 Thomas Henry (FR)

**16:08** **O17 - Unraveling the molecular pathogenesis of proteasome-associated autoinflammatory syndromes**  
 Frédéric Ebstein (DE)

**16:16** **O18 - A novel knock-in mouse model of CAPS that develops amyloidosis: therapeutic efficacy of proton pump inhibitors**  
 Arinna Bertoni (IT)

**16:24** **O19 - T cell defects in patients with ARPC1B germline mutations account for combined immunodeficiency**  
 Immacolata Brigida (IT)

**16:32** **O20 - B cell defect in ADA2 deficiency patients**  
 Francesca Schena (IT)

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- 16:40 **O21 - Periodic fever, aphthous stomatitis, pharyngitis and adenitis (PFAPA) syndrome and obstructive sleep apnea are distinct inflammatory disorders of oropharyngeal lymphoid tissue**  
Kalpana Manthiram (US)
- 16:48 **O22 - PFAPA syndrome: NK cells infiltrating tonsils support the crucial role of innate immunity in the pathogenesis**  
Sabrina Chiesa (IT)
- 17:00 - 18:30 **NOVEL AID PATHWAYS AND GENES** **MAESTRALE ROOM**  
*Chairpersons: Isabella Ceccherini (IT) & Marielle van Gijn (NL)*
- 17:00 **Epigenetics for clinicians**  
Seza Ozen (TR)
- 17:25 **The genetic journey of autoinflammatory diseases: From linkage analysis to WGS**  
Isabelle Touitou (FR)
- 17:50 **Ubiquitination defects: The evolving story**  
Ivona Aksentijevich (US)
- 18:15 **O23 - Identification of rare coding variants in IL-1-related pathways in patients with adult-onset still's disease**  
Giulio Cavalli (IT)
- 18:22 **O24 - Trisomy 8-associated autoinflammatory disease (TRIAD) is characterized by increased monocyte activation**  
Kalpana Manthiram (US)

## TUESDAY 02 APRIL 2019

- 07:30 - 08:30**    **WAKE UP EARLY AND FIND A NEW DISEASE!**    **MAESTRALE ROOM**  
*Discussion led by Paul Brogan (UK) and Raphaela Goldbach-Mansky (US)*
- 08:30 - 10:00**    **DEFICIENCIES IN KEY REGULATORY SIGNALS**    **MAESTRALE ROOM**  
*Chairpersons: Angela Rösen-Wolff (DE) & Carine Wouters (BE)*
- 08:30**    **DADA2: The hematological perspective**  
 Isabelle Meyts (BE)
- 08:55**    **LRBA Deficiency, CTLA-4 haploinsufficiency and STAT3-GOF**  
 Bodo Grimbacher (DE)
- 09:20**    **When actin is not acting right**  
 Steven Holland (US)
- 09:45**    **O25 - A combined immunodeficiency with severe infections, inflammation and allergy caused by ARPC1B deficiency**  
 Stefano Volpi (IT)
- 09:52**    **O26 - ARPC1B-deficiency causes defective cell migration, loss of actin polymerization and hyperresponsiveness in zebrafish and patient-derived cells**  
 Gabriella Leung (CA)
- 10:00 - 10:30**    **COFFEE BREAK & VISIT OF THE EXHIBITION**    **EXHIBITION AREA**
- 10:30 - 12:00**    **NUCLEIC ACID SENSING AND INTERFERON**    **MAESTRALE ROOM**  
*Chairpersons: Andrea Ablasser (CH) & Yanick Crow (UK)*
- 10:30**    **Extracellular DNA in autoinflammation**  
 Boris Reizis (US)
- 10:55**    **Nucleic acid sensing in disease**  
 Sun Hur (US)
- 11:20**    **What's new on interferonopathies**  
 Yanick Crow (UK)
- 11:45**    **O27 - Heterozygous mutations in COPA are associated with enhanced type I interferon signaling**  
 Marie-Louise Frémond (FR)
- 11:52**    **O28 - Sting-associated vasculopathy in mice requires adaptive immunity but not CGAS or type I interferon**  
 Angela Rösen-Wolff (DE)
- 12:30 - 13:30**    **SPONSORED SYMPOSIUM**    **MAESTRALE ROOM**  
*Refer to page 43.*
- 13:30 - 15:00**    **THE SKIN AS A TARGET IN AUTOINFLAMMATORY DISEASES**    **MAESTRALE ROOM**  
*Chairpersons: Sulaiman Al-Mayouf (SA) & Aurora Parodi (IT)*
- 13:30**    **Hidradenitis suppurativa. A focus on autoinflammation**  
 Barbara Horvath (NL)



## TUESDAY 02 APRIL 2019

- 13:50**      **PAPA spectrum disorders**  
Angelo Marzano (IT)
- 14:10**      **Neutrophilic urticarial dermatosis: An entity bridging monogenic and polygenic auto inflammatory disorders, and beyond**  
Dan Lipsker (FR)
- 14:30**      **Dermatologic clues for the diagnosis of auto inflammatory diseases in children**  
Antonio Torello (ES)
- 14:50**      **General discussion**

**15:00 - 16:00**      **COFFEE BREAK & POSTER GUIDED TOURS**

**POSTER AREA**

**POSTER GUIDED TOUR 2A**

*Chairpersons: Erkan Demirkaya (TR) & Yosef Uziel (IL)*

- PT2A01**      **THE FRENCH PEDIATRIC COHORT OF CASTLEMAN DISEASE** / C. Borocco - C. Ballot-Schmit - O. Ackermann - N. Aladjidi - J. Delaleu - V. Giacobbi-Milet - S. Jannier - E. Jeziorski - Y. Perel - F. Maurier - C. Piguët - E. Oksenhendler - I. Kone-Paut - C. Galeotti\*
- PT2A02**      **USE OF WHOLE-BODY MAGNETIC RESONANCE TO IDENTIFY POTENTIAL DIAGNOSTIC CLUES IN CHILDREN WITH FEVER OF UNKNOWN ORIGIN (FUO)** / S. Signa\* - R. Caorsi - G. Stagnaro - F. Minoia - P. Picco - A. Ravelli - G. Magnano - M. B. Damasio - M. Gattorno
- PT2A03**      **A BIOMARKER PROFILE INCLUDING S100A12 AS DIAGNOSTIC TOOL FOR THE DIFFERENTIAL DIAGNOSIS OF SJIA-ASSOCIATED MAS VS. PRIMARY OR SECONDARY HLH** / C. Kessel - N. Fall - A. Grom - W. de Jager - S. Vastert - R. Strippoli - C. Bracaglia - E. Sundberg - A. Horne - S. Ehl - S. Ammann - K. Lehmborg - F. De Benedetti - H. Wittkowski - K. Kessel - K. Beutel - D. Foell - D. Holzinger\*
- PT2A04**      **EARLY TREATMENT WITH ANAKINRA IN SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS** / M. Pardeo\* - C. Bracaglia - A. Tulone - A. Insalaco - G. Marucci - R. Nicolai - V. Messina - E. Sacco - F. De Benedetti
- PT2A05**      **ANTIBODIES TO COLLAGEN V AND K-A 1 TUBULIN IN A PATIENT WITH SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS ASSOCIATED LUNG DISEASE** / E. Tronconi\* - T. Mohanakumar - L. Danziger-Isakov - G. Schulert - A. Grom
- PT2A06**      **RECURRENT FEVER, RECURRENT PYODERMA GANGRENOSUM-LIKE AND BACTERIAL INFECTIONS DUE TO A NOVEL HOMOZYGOUS WDR1 MUTATION IN A CHILD TREATED WITH ALLOGENEIC HEMATOPOIETIC STEM CELL TRANSPLANTATION** / E. Iglesias - M. Roldan - A. Vlaga - M. Solís-Moruno - J. M. Mosquera - V. Bitterman - J. Calzada-Hernández - L. Alsina - A. Deyà-Martinez - R. Lara - M. C. Anton - S. Plaza - H. Martínez-Banaclocha - M. Trabazo - G. Morón - M. Juan - J. Yagüe - F. Casals - I. Badell - C. Fortuny - A. Vicente - J. Anton - P. Pelegrin - J. I. Aróstegui - A. Mensa-Vilaro\*

**POSTER GUIDED TOUR 2B**

*Chairpersons: Dirk Foell (DE) & Michael Hofer (CH)*

- PT2B01**      **ADENOSINE DEAMINASE 2 DEFICIENCY: CORRECTION OF THE IMMUNE DEFECTS BY GENE THERAPY** / I. Brigida - M. Zoccolillo - R. J. Hernandez - L. Sergi Sergi - G. Milardi - F. Barzaghi - L. Naldini - M. P. Cicalese - A. Aiuti - A. Mortellaro\*
- PT2B02**      **ANALYSIS OF THE EFFICACY OF TREATMENT ON 45 PATIENTS WITH DEFICIENCY OF ADENOSINE DEAMINASE 2** / A. K. Ombrello\* - D. L. Stone - K. Barron - P. M. Hoffmann - C. Cudrici - A. Jones - T. Romeo - D. Dimitrova - A. Dotan - D. Wall - M. Thakar - J. Kanakry - D. Kastner

## TUESDAY 02 APRIL 2019

- PT2B03**      **LONG-TERM RETENTION RATE OF ANAKINRA IN ADULT ONSET STILL'S DISEASE AND PREDICTIVE FACTORS FOR TREATMENT RESPONSE** / A. Vitale\* - G. Cavalli - S. Colafrancesco - R. Priori - G. Valesini - L. M. Argolini - E. Baldissera - E. Bartoloni - D. Cammelli - G. Canestrari - J. Sota - E. Cavallaro - M. G. Massaro - P. Ruscitti - P. Cipriani - G. De Marchi - S. De Vita - G. Emmi - G. Ferraccioli - M. Frassi - R. Gerli - E. Gremese - F. Iannone - G. Lapadula - G. Lopalco - R. Manna - A. Mathieu - C. Montecucco - M. Mosca - I. Piazza - M. Piga - I. Pontikaki - M. Romano - S. Rossi - M. Rossini - E. Silvestri - C. Stagnaro - R. Talarico - A. Tincani - O. Viapiana - G. Vitiello - P. Galozzi - P. Sfriso - C. Gaggiano - D. Rigante - L. Dagna - R. Giacomelli - L. Cantarini on behalf of "Working Group" of Systemic Autoinflammatory Diseases of SIR (Italian Society of Rheumatology)
- PT2B04**      **THE ANTI-INFLAMMATORY CYTOKINE INTERLEUKIN 37 IS AN ENDOGENOUS INHIBITOR OF TRAINED IMMUNITY** / G. Cavalli\* - M. Gresnigt - T. Nemkov - R. Arts - A. D'Alessandro - S. Giugliano - E. Eisenmenger - L. Dagna - L. Joosten - M. Netea - C. Dinarello
- PT2B05**      **INTERLEUKIN 18 GENE EXPRESSION BY HUMAN MONOCYTES IS CONTROLLED BY TYPE I INTERFERON** / E. Verweyen - D. Holzinger - H. Wittkowski - P. Pickkers - D. Foell - C. Kessel\*
- PT2B06**      **GLYCOGEN SYNTHASE KINASE 3B REGULATES TLR3-MEDIATED ANTIVIRAL RESPONSE** / R. Ko\* - S. Y. Lee
- PT2B07**      **ANTICYTOKINE THERAPY IS SUCCESSFUL IN PREVENTION AND TREATMENT OF BCG-ASSOCIATED INFLAMMATORY SYNDROME (IS) AFTER HEMATOPOIETIC STEM CELL TRANSPLANTATION (HSCT) IN PATIENTS WITH SEVERE COMBINED IMMUNODEFICIENCIES (SCID)** / A. Laberko - I. Shipitsina - S. Rodigina - S. Blagov - D. Yukhacheva - E. Deripapa - A. Kozlova - Y. Rodina - L. Shelikhova - D. Balashov - A. Shcherbina\*
- PT2B08**      **ALLOGENEIC HAEMATOPOIETIC STEM CELL TRANSPLANTATION (HSCT) OUTCOME IN IMMUNEDYSREGULATION AND RHEUMATOLOGIC PATIENTS: A SINGLE CENTER 19 YEARS EXPERIENCE** / S. Signa\* - A. Gennery - S. Hambleton - T. Flood - A. Cant - M. Abinun - M. Slatter
- PT2B09**      **GLOBAL PROFILING AND MOLECULAR CHARACTERIZATION OF THE AUTOANTIBODY REPERTOIRE IN APECED** / J. M. Lindner - A. Krämer - X. Leber - V. Caballero - F. Kolbinger - M. S. Lionakis - S. Holland - E. Traggiai\*

### POSTER VIEWING - MONOGENIC AUTOINFLAMMATORY DISEASES (CLINICAL)

- P2001**      **IN VITRO NLRP3 INFLAMMASOME ACTIVATION ASSAY ASSISTS DIAGNOSIS OF GENETICALLY NEGATIVE CAPS PATIENTS AND GUIDES ANTI-IL1 THERAPY** / L. O. Mendonca\* - M. T. Barros - T. Robazzi - P. F. Giavina-Bianchi - F. Caroli - A. Grossi - J. Kalil - F. F. Morato Castro - I. Ceccherini - M. Gattorno - A. Pontillo
- P2002**      **VACCINATION SAFETY AND COVERAGE IN A SINGLE COHORT OF AUTOINFLAMMATORY SYNDROMES IN ITALY** / L. O. Mendonca\* - E. Toniolo - S. Volpi - R. Caorsi - M. Bustaffa - M. D'Alessandro - S. Signa - M. Gattorno
- P2003**      **PERIPHERAL T REGULATORY LYMPHOCYTES AND APOPTOSIS EXPRESSION IN A LARGE GROUP OF AUTOIMMUNE, MULTIFACTORIAL AND AUTOINFLAMMATORY DISEASES REVEALS SPECIFIC CLINICAL MANIFESTATIONS** / L. O. Mendonca\* - M. Bustaffa - C. Maticci-Cerinic - I. Prigione - S. Volpi - R. Caorsi - S. Chiesa - F. Schena - A. Grossi - P. Terranova - I. Ceccherini - M. Miano - M. Gattorno
- P2004**      **ASSOCIATION OF GENE POLYMORPHISM WITH AUTOINFLAMMATORY SYNDROMES IN PATIENTS WITH PALINDROMIC RHEUMATISM AND INTERMITTENT HYDRARTHROSIS** / T. Miyamae\* - Y. Tani - T. Kishi - M. Kawamoto - Y. Kawaguchi - A. Taniguchi - H. Yamanaka
- P2005**      **REPORT OF 65 PATIENTS WITH SAID IN ARGENTINA** / I. Moreira\* - A. G. Seminario - A. Llarens - L. R. Riaño Cardozo - L. Regairaz - L. Bezrodnik

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- P2006**      **MULTISYSTEMIC INFLAMMATORY DISEASE DUE TO DNASE2 MUTATIONS: FROM PHYSIOPATHOLOGY TO NEW THERAPEUTIC APPROACHES** / V. Moressa\* - A. Tesser - A. Trombetta - S. Ghirardo - M. Bobbo - E. Piscianz - F. Faletra - S. Volpi - A. Tommasini - S. Pastore - A. Taddio
- P2007**      **PREGNANCY AND OTHER GYNECOLOGICAL ASPECTS IN MEVALONATE KINASE DEFICIENCY** / C. Mulders-Manders\* - A. Simon
- P2008**      **EARLY ONSET SARCOIDOSIS ASSOCIATED WITH NOD2 MUTATION: FAMILIAL CASES REPORT** / R. Naddei\* - F. Orlando - C. Porfito - T. Lastella - M. Amico - M. Alessio
- P2009**      **EXTENDING THE CLINICAL SPECTRUM OF BLAU SYNDROME, A CASE REPORT WITH LIVER CIRRHOSIS** / M. Nashawi\* - H. Brennenstuhl - B. Bangol - T. Lutz
- P2010**      **DIAGNOSIS AND MANAGEMENT OF HEREDITARY RECURRENT FEVERS: 20-YEAR EXPERIENCE AT A SINGLE ITALIAN REFERRAL CENTRE** / L. Obici\* - G. Bossi - R. Caporali - R. Mussinelli - S. Casarini - S. Monti - I. Ceccherini - F. Caroli - A. Vergori - E. Di Buduo - C. Sforzini - M. Gattorno - G. Merlini
- P2011**      **RNF213 GENE VARIANTS IN PATIENTS WITH TAKAYASU ARTERITIS OR VASCULOPATHY** / E. Omoyinmi\* - A. Keylock - D. McCreary - S. Fingerhutova - P. Dolezalova - S. Ozen - S. Kamphuis - C. Hemingway - D. Eleftheriou - P. Brogan
- P2012**      **CLINICAL AND MOLECULAR FEATURES OF 3 CHILDREN WITH NEONATAL ONSET MULTISYSTEM INFLAMMATORY DISEASE: AN EXPERIENCE FROM A SETTING WHERE THERE IS LACK OF ACCESS TO ANTI-IL1 AGENTS** / V. Pandiarajan\* - D. Suri - S. Bhattad - A. Gupta - A. Rawat - R. Goldbach-Mansky - M. Gattorno - S. Singh
- P2013**      **LONG-TERM OUTCOMES AND TREATMENT EFFICACY IN PATIENTS WITH TNF RECEPTOR-ASSOCIATED AUTOINFLAMMATORY SYNDROME (TRAPS): A SERIES OF 290 CASES FROM THE EUROFEVER/EUROTRAPS INTERNATIONAL REGISTRY** / R. Papa\* - T. Lane - T. Youngstein - T. Rezk - C. Papadopoulou - N. Ruperto - P. A. Brogan - P. N. Hawkins - P. Woo - M. Gattorno - H. J. Lachmann
- P2014**      **A CHILD WITH MAJEED SYNDROME FROM INDIA** / P. Pimpale Chavan\* - R. P. Khubchandani
- P2015**      **SPONDYLOENCHONDRODYSPLASIA – A CASE FROM INDIA** / P. Pimpale Chavan\* - M. Moreews - S. Ranade - A. Belot - R. P. Khubchandani
- P2016**      **CLINICAL PROFILE OF SEVEN CHILDREN WITH HEREDITARY COMPLEMENT DEFICIENCY FROM A CENTER IN MUMBAI, INDIA** / P. Ranade - Y. J. Crow - P. Pimpale Chavan\* - L. Seabra - G. Rice - R. P. Khubchandani
- P2017**      **PRELIMINARY RESULTS OF THE LATIN-AMERICAN COHORT OF AUTO-INFLAMMATORY SYNDROMES** / D. G. P. Piotto\* - C. A. Silva - K. Kozu - A. L. Cunha - F. P. Santos - A. L. Tolin - S. K. Oliveira - S. Appenzeller - C. S. Magalhães - M. Bandeira - F. Sztajn bok - C. N. Rabelo Jr - C. L. de Cunto - C. De Battagliotti - B. Bica - P. Munitis - L. Bezrodnik - M. F. Rodrigues - T. C. Robazzi - E. N. Matos - R. Russo - M. T. Terreri
- P2018**      **CHRONIC NONBACTERIAL OSTEOMYELITIS: REPORT OF SIX CASES** / D. G. P. Piotto\* - A. Aihara - A. Fernandes - G. Balbi - C. Len - M. T. Terreri
- P2019**      **FAMILIAL MEDITERRANEAN FEVER: STRONG EVIDENCE FOR A FUNCTIONAL EFFECT OF E148Q WHEN COMBINED WITH M694V** / E. Pras\* - O. Eyal - Y. Shinar - M. Pras
- P2020**      **TREATMENT OF TYPE 1 INTERFERONOPATHY WITH CICLOSPORIN A AND BARICITINIB IN A 5-YEAR-OLD BOY WITH HETEROZYGOUS PSMB-8 MUTATION** / C. Rietschel\* - E. Salamano - M. A. Lee-Kirsch - K. Latta

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- P2021**      **PROTEIN PRENYLATION IS DEFECTIVE IN MEVALONATE KINASE DEFICIENCY AND IS AN ACCURATE BIOMARKER IN PERIPHERAL BLOOD MONONUCLEAR CELLS** / M. Rogers\* - J. Jurczyk - O. Skinner - A. Simon - S. Mehr - P. Hissaria - R. Arts - D. Coman - M. Munoz
- P2022**      **ENCEPHALOPATHY IN EARLY-ONSET SARCOIDOSIS: IS IT NEUROSARCOIDOSIS OR AUTOIMMUNE ENCEPHALITIS?** / N. Sahin\* - S. O. Cicek - A. P. Kisaarslan - Z. Gündüz - M. H. Poyrazoglu - R. Düşünel
- P2023**      **RISK FACTORS OF COLCHICINE RESISTANCE IN PEDIATRIC PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER: A SINGLE CENTER EXPERIENCE** / N. Sahin\* - A. P. Kisaarslan - S. O. Cicek - Z. Gunduz - M. H. Poyrazoglu - R. Düşünel
- P2024**      **DISTINCTIVE FEATURES OF FAMILIAL MEDITERRANEAN FEVER WITH JUVENILE SPONDYLOARTHRITIS PATIENTS FROM FAMILIAL MEDITERRANEAN FEVER AND JUVENILE SPONDYLOARTHRITIS PATIENTS** / A. Paç Kisaarslan - N. Şahin\* - S. Özdemir Çiçek - Z. Gündüz - M. H. Poyrazoğlu - R. Düşünel
- P2025**      **TYPE 1 INTERFERONOPATHY PRESENTING WITH FEVER, FATIGUE, CHRONIC URTICARIA, ARTHRITIS, ELEVATED LIVER ENZYMES AND HYPERFERRITINEMIA IN A 13-YEAR-OLD GIRL – AN IMPORTANT DIFFERENTIAL DIAGNOSIS TO SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS** / E. Salamano\* - M. A. Lee-Kirsch - C. Rietschel
- P2026**      **CRYOPYRIN-ASSOCIATED PERIODIC SYNDROMES IN ADULTS: DIAGNOSIS, THERAPY IN CLINICAL PRACTICE** / S. Salugina\* - E. Fedorov - T. Dubinina - S. Palshina
- P2027**      **PULMONARY SCREENING OF FMF PATIENTS DURING PERIODIC FOLLOW-UP: CLINICAL AND PATHOGENETIC CONSIDERATIONS** / A. Sargsyan\*
- P2028**      **IDENTIFICATION OF NOVEL LOSS-OF-FUNCTION MUTATIONS IN TWO INDEPENDENT PATIENTS WITH DEFICIENCY OF ADENOSINE DEAMINASE 2** / O. Schnappauf\* - N. Sampaio Moura - Q. Zhou - N. Deutch - D. Kastner - I. Aksentjevich
- P2029**      **HUMORAL IMMUNE COMPROMISE WITH AUTOINFLAMMATORY DISEASE DUE TO MUTATION IN NLRP12** / A. G. Seminario\* - M. S. Caldirola - I. Moreira - L. Regairaz - L. Bezrodnik
- P2030**      **GENETIC SCREENING IN PATIENTS WITH UNDIFFERENTIATED PERIODIC FEVER SYNDROME** / F. Demir - O. Akgun Dogan - Y. Kendir Demirkol - K. Ermis Tekkus - S. Canbek - N. Aktay Ayaz - L. Doganay - B. Sozeri\*
- P2031**      **DEFICIENCY OF ADA2 FROM CHILDHOOD TO ADULT; THE SAME MUTATION IN A FAMILY** / B. Sozeri\* - G. Ercan - O. Akgun Dogan - J. Yildiz - F. Demir - L. Doğanay
- P2032**      **AUTOINFLAMMATORY DISEASES IN UKRAINE** / Y. Stepanovskyy\* - Y. Boyko
- P2033**      **THE SPECTRUM OF AUTOINFLAMMATORY SYNDROMES AT POSTGRADUATE INSTITUTE OF MEDICAL EDUCATION AND RESEARCH, CHANDIGARH, INDIA** / D. Suri\* - A. Rawat - A. Gupta - P. Vignesh - A. Jindal - S. Bhattad - M. Gattorno - A. A de Jesus - R. Goldbach-Mansky - S. Singh
- P2034**      **PATIENT WITH FMF PRESENTED BY ISOLATED MYOSITIS** / S. N. Taskin\* - A. Kisaarslan - S. Ozdemir Çiçek - N. Sahin - M. H. Poyrazoğlu - R. Dusunsel
- P2035**      **INTERSTITIAL LUNG DISEASE IN A NEWBORN AFFECTED BY MEVALONIC ACIDURIA** / S. Torreggiani\* - C. Pietrasanta - F. Minoia - G. Filocamo - A. Ronchi - S. Volpi - R. Caorsi - M. Gattorno - F. Caroli - A. Grossi - I. Ceccherini - L. Pugni - F. Mosca

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- P2036** **FAMILIAL MEDITERRANEAN FEVER RELATED DAMAGE ASSESSED BY AUTO-INFLAMMATORY DISEASE DAMAGE INDEX (ADDI) AND ASSOCIATED FACTORS WITH DAMAGE** / H. Babaoglu - B. Armagan - E. Bodakci - T. Kasifoglu - H. Satis - N. Atas - A. Sari - G. K. Yardimci - N. S. Y. Bilge - R. B. Salman - L. Kilic - M. A. Ozturk - S. Haznedaroglu - B. Goker - U. Kalyoncu - A. Tufan\*
- P2037** **FAMILIAL MEDITERRANEAN FEVER ASSOCIATED INFLAMMATORY DISORDERS** / N. Atas - B. Armagan - E. Bodakci - T. Kasifoglu - H. Satis - A. Sari - N. S. Y. Bilge - H. Babaoglu - G. K. Yardimci - R. B. Salman - L. Kilic - M. A. Ozturk - S. Haznedaroglu - B. Goker - U. Kalyoncu - A. Tufan\*
- P2038** **WORK PRODUCTIVITY AND ACTIVITY IMPAIRMENT IN FAMILIAL MEDITERRANEAN FEVER PATIENTS** / E. Suticen - N. Atas - H. Babaoglu - H. Satis - R. B. Salman - O. Varan - M. A. Ozturk - S. Haznedaroglu - B. Goker - A. Tufan\*
- P2039** **HUMAN PHENOTYPE ONTOLOGY FOR SYSTEMIC AUTOINFLAMMATORY DISORDERS** / M. Van Gijn\* - J. Pazmandi - P. Brogan - M. Gattorno - P. van Daele - K. Boztug - P. Robinson - M. Haimel - A. Simon
- P2040** **DIAGNOSIS AND STRATIFICATION OF FAMILIAL MEDITERRANEAN FEVER BY A SIMPLE FUNCTIONAL ASSAY** / O. Zhogova - N. Lagunova - S. Ivanovskiy - E. Suspitsin - M. Kostik\*
- P2041** **EVALUATION OF COEXISTING DISEASES IN CHILDREN WITH FAMILIAL MEDITERRANEAN FEVER** / M. Yildiz\* - A. Adrovic - E. Tasdemir - K. Baba-Zada - M. Aydin - O. Koker - S. Sahin - K. Barut - O. Kasapcopur
- P2042** **RESULTS OF INTERNATIONAL DELPHI SURVEY ON THE INVESTIGATION AND MANAGEMENT OF FIRST-DEGREE RELATIVES OF THOSE WITH DEFICIENCY OF ADA2** / T. A.-B. Youngstein\* - E. P. Chambers - H. J. Lachmann on behalf of DADA2 Delphi Study Participants

### POSTER VIEWING - NON-MONOGENIC SAIDS (CLINICAL)

- P2043** **WE NEED MORE TESTS FOR DISCRIMINATION NON-BACTERIAL OSTEOMYELITIS FORM TUBERCULOSIS IN EARLY STAGES** / M. Kostik\* - O. Kopchak - A. Maletin - V. Zorin - A. Mushkin
- P2044** **THE FEATURES AND DISTRIBUTION OF CHRONIC NON-BACTERIAL OSTEOMYELITIS IN RUSSIAN FEDERATION** / M. Kostik\* - M. Makhova - V. Zorin - E. Suspitsin - E. Isupova - S. Magomedova - I. Kostik - A. Mushkin
- P2045** **KAWASAKI DISEASE SHOCK SYNDROME: CLINICAL CHARACTERISTICS AND POSSIBLE USE OF IL-6, IL-10 AND IFN-GAMMA AS BIOMARKERS FOR EARLY RECOGNITION** / M. Lu\* - Y. Li - Q. Zheng
- P2046** **OUTCOMES FOLLOWING TONSILLECTOMY IN PATIENTS WITH PERIODIC FEVER, APHTHOUS STOMATITIS, PHARYNGITIS, AND ADENITIS (PFAPA) SYNDROME** / K. Manthiram\* - S. Lapidus - K. Barron - A. Ombrello - D. Kastner - K. Edwards
- P2047** **RELATIONSHIP BETWEEN MEFV GENE AND CLINICAL FINDINGS OF CHRONIC RECURRENT MULTIFOCAL OSTEOMYELITIS** / S. Özdemir Çiçek\* - N. Şahin - Z. F. Karaman - S. N. Taşkın - A. Paç Kisaarslan - Z. Gündüz - M. H. Poyrazoğlu - R. Düşünsel
- P2048** **BEHÇET'S DISEASE WITH PSEUDOTUMOR CEREBRI AND CEREBRAL VENOUS SINUS THROMBOSIS: A CASE REPORT** / S. Özdemir Çiçek\* - N. Şahin - S. N. Taşkın - A. Paç Kisaarslan - Z. Gündüz - M. H. Poyrazoğlu - R. Düşünsel

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### POSTER VIEWING - MONOGENIC AUTOINFLAMMATORY DISEASES (BASIC SCIENCE)

- P2049**      **THE USE OF NEXT GENERATION SEQUENCING PANEL IN UNDIFFERENTIATED AUTOINFLAMMATORY DISEASES IDENTIFY A SEPARATE SUBSET OF COLCHICINE-RESPONDER RECURRENT FEVERS DISTINCT FROM PFAPA SYNDROME** / R. Papa\* - M. Rusmini - S. Volpi - R. Caorsi - P. Picco - A. Grossi - F. Caroli - F. Bovis - V. Musso - L. Obici - C. Castana - A. Ravelli - M. E. Van Gijn - I. Ceccherini - M. Gattorno
- P2050**      **EPIDEMIOLOGY AND CLINICAL FEATURES OF PFAPA IN WESTERN SWEDEN** / K. Rydenman\* - H. Fjeld - A. Karlsson - S. Berg - A. Fasth - P. Wekell
- P2051**      **OBSERVATIONS IN A PATIENT WITH PFAPA-LIKE DISEASE ASSOCIATED WITH ERYSIPELAS-LIKE ERYTHEMA AND R202Q VARIANT IN MEFV** / P. Wekell\* - M. Olsson - P. Söderkvist - A. Karlsson - S. Berg - A. Fasth
- P2052**      **YAO SYNDROME VERSUS FAMILIAL MEDITERRANEAN FEVER** / Q. Yao\* - P. Gorevic
- P2053**      **MAY SOME OF THE MEFV GENE VARIANTS CAUSE PFAPA SYNDROME LIKE SYMPTOMS?** / A. Adrovic - M. Yildiz\* - I. Ulkersoy - N. Gucuyener - O. Koker - S. Sahin - K. Barut - O. Kasapcopur

### POSTER VIEWING - NON-MONOGENIC SAIDS (BASIC SCIENCE)

- P2054**      **NEXT GENERATION SEQUENCING ANALYSIS OF FAMILIAL HAEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS RELATED GENES IN MACROPHAGE ACTIVATION SYNDROME AND SECONDARY HLH** / C. Passarelli - M. Pardeo - I. Caiello - E. Pisaneschi - A. Insalaco - F. Minoia - A. Taddio - F. Licciardi - A. Novelli - F. De Benedetti - C. Bracaglia\*
- P2055**      **EVALUATION OF THE NEW CLASSIFICATION CRITERIA FOR PFAPA SYNDROME** / F. Crimi - V. Hentgen - G. Dingulu - I. Koné-Paut - S. Georgin-Lavialle - P. Pillet - M. Hofer\*
- P2056**      **HUMAN (PRO)CASPASE-1 VARIANTS INFLUENCE CELL DEATH AND MECHANICAL PROPERTIES OF HUMAN MONOCYTES/MACROPHAGES** / F. Schulze - F. Kapplusch - S. Rabe-Matschewsky - S. Russ - M. Herbig - U. Range - A. Rösen-Wolff - S. Winkler - C. M. Hedrich - J. Guck - S. R. Hofmann\*
- P2057**      **PATIENT WITH NOD2, NLRP1 AND C9 VARIANTS WITH RECURRENT FEVER ATTACKS, INFECTIOUS SUSCEPTIBILITY AND OSTEOMYELITIC LESIONS INDICATIVE OF NAID?** / C. Høst\* - M. Christiansen - S. E. Jørgensen - T. Herlin - M. Glerup - B. Mahler - T. Mogensen
- P2058**      **THE CYTOKINES PROFILES IN CHRONIC NON-BACTERIAL OSTEOMYELITIS NON-SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS AND HEALTHY CONTROLS** / M. Kostik\* - D. Kozlova - D. Vasiliev - O. Karlina - M. Makhova - L. Sorokina - V. Zorin - E. Isupova - S. Magomedova - I. Kostik - A. Mushkin
- P2059**      **GENETIC ANALYSIS OF THE PATIENTS WITH EARLY-ONSET CHRONIC NON-BACTERIAL OSTEOMYELITIS FROM NORTH CAUCASIAN REGIONS OF RUSSIA** / M. Kostik\* - M. Makhova - E. Suspitsin - A. Sokolenko - V. Zorin - E. Isupova - S. Magomedova - I. Kostik - H. Takayanagi - A. Mushkin - E. Imyanitov
- P2060**      **PROTEOMIC PROFILE OF PERIPHERAL BLOOD MONONUCLEAR CELLS IN BEHÇET'S** / G. Turan - I. Karacan - S. Ugurlu - S. Dasdemir - H. Ozdogan - A. Tolun - E. Tahir Turanli\*
- P2061**      **IL1RAP AS A CANDIDATE GENE FOR AUTOSOMAL DOMINANT BEHÇET'S DISEASE** / G. Turan - I. Karacan - S. Ugurlu - S. Dasdemir - H. Ozdogan - A. Tolun - E. Tahir Turanli\*



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### POSTER VIEWING - SYSTEMIC-ONSET JIA AND AOSD

- P2062** **CANAKINUMAB IN SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS: CLINICAL INACTIVE DISEASE RATE AND SAFETY IN ITALIAN PATIENTS** / M. Pardeo\* - C. Bracaglia - A. L. Piscitelli - A. De Matteis - J. Tibaldi - M. Alessio - A. Marino - G. Conti - M. C. Maggio - C. Alizzi - F. Licciardi - A. N. Olivieri - G. Filocamo - F. Orlando - S. Martino - R. Cimaz - A. Ravelli - F. De Benedetti
- P2063** **WHOLE BLOOD PHOSPHORYLATED STAT1 LEVELS IN PATIENTS WITH ACTIVE MACROPHAGE ACTIVATION SYNDROME AND SECONDARY HEMOPHAGOCYtic LYMPHOHISTIOCYTOSIS** / A. Pascarella\* - C. Bracaglia - E. Marasco - I. Caiello - G. Moneta - L. Farina - G. Marucci - A. Arduini - M. Pardeo - F. De Benedetti - G. Prencipe
- P2064** **WHOLE BLOOD CELLS FROM PATIENTS WITH SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS (SJIA) IN CLINICAL INACTIVE DISEASE DISPLAYED A DYSREGULATED RESPONSE TO TLR-4 STIMULATION** / A. Pascarella - M. Pardeo - I. Caiello - C. Bracaglia - M. N. Rossi - G. Marucci - E. Sacco - F. De Benedetti - G. Prencipe\*
- P2065** **ANAKINRA DRUG RETENTION RATE AND PREDICTIVE FACTORS OF DRUG SURVIVAL IN SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS AND ADULT ONSET STILL'S DISEASE** / J. Sota\* - D. Rigante - A. Insalaco - P. Sfriso - S. de Vita - R. Cimaz - G. Lopalco - G. Emmi - F. La Torre - C. Fabiani - A. N. Olivieri - M. Cattalini - D. Cammelli - R. Gallizzi - M. Alessio - R. Manna - O. Viapiana - M. Frassi - A. Maier - C. Salvarani - R. Talarico - R. Priori - M. C. Maggio - M. Pardeo - C. Gaggiano - S. Grosso - F. de Benedetti - A. Vitale - L. Cantarini
- P2066** **PREDICTORS OF EFFECTIVENESS OF ANAKINRA IN SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS** / J. Tibaldi\* - B. Saccomanno - F. Minoia - F. Bagnasco - A. Pistorio - A. Guariento - R. Caorsi - A. Consolaro - M. Gattorno - A. Ravelli
- P2067** **CANAKINUMAB TREATMENT IN ADULT-ONSET STILL'S DISEASE: CASE SERIES** / S. Ugurlu\* - G. Guzelant - B. Yurttas - B. Ergezen - E. Dalkilic - T. Kasifoglu - B. Yagiz - H. Ozdogan

### POSTER VIEWING - THERAPY OF SYSTEMIC AUTO-INFLAMMATORY DISEASES

- P2068** **MICROBIOTA TRANSPLANT TO CONTROL INFLAMMATION IN A NLR4-RELATED DISEASE PATIENT WITH RECURRENT HEMOPHAGOCYtic LYMPHOHISTIOCYTOSIS (HLH)** / C. Bracaglia\* - G. Marucci - F. Del Chierico - A. Russo - M. Pardeo - A. Insalaco - G. Prencipe - I. Caiello - S. Fingerhutova - P. Dolezalova - F. De Benedetti - L. Putignani
- P2069** **EMAPALUMAB, AN ANTI-INTERFERON GAMMA MONOCLONAL ANTIBODY IN TWO PATIENTS WITH NLR4- RELATED DISEASE AND SEVERE HEMOPHAGOCYtic LYMPHOHISTIOCYTOSIS (HLH)** / C. Bracaglia\* - G. Prencipe - A. Insalaco - I. Caiello - G. Marucci - M. Pardeo - R. Pecoraro - P. Dolezalova - S. Fingerhutova - M. Ballabio - C. de Min - F. De Benedetti
- P2070** **IL-1 BLOCKADE IN PAEDIATRIC RECURRENT PERICARDITIS: A MULTICENTRIC RETROSPECTIVE STUDY OF THE ITALIAN COHORT** / R. Caorsi\* - A. Insalaco - C. Longo - G. Martini - M. Cattalini - R. Consolini - G. Filocamo - A. Rimini - S. Federici - C. Celani - M. C. Maggio - M. Romano - B. Teruzzi - A. Taddio - A. Miniaci - S. Martino - F. Latorre - A. De Fanti - G. Cavalli - B. Bigucci - A. Brucato - F. De Benedetti - M. Gattorno on behalf of The Italian study group on anti-IL-1 drugs' efficacy in recurrent pericarditis
- P2071** **EFFICACY AND SAFETY OF ANAKINRA IN THE TREATMENT OF INFLAMMATORY HEART FAILURE IN MYOCARDITIS** / G. De Luca\* - C. Campochiaro - G. Cavalli - S. Sartorelli - L. Dagna
- P2072** **EFFICACY, SAFETY AND COST-EFFECTIVENESS OF A VIAL-SHARING PROGRAMME FOR CANAKINUMAB TREATMENT FOR PAEDIATRIC PATIENTS WITH CRYOPYRIN-ASSOCIATED PERIODIC SYNDROME** / A. A. Elmi\* - K. Wynne - I. L. Cheng - D. Eleftheriou - H. J. Lachmann - P. N. Hawkins - P. Brogan

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- P2073** ANTI-IL-1 THERAPIES IN PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER RELATED AA-AMYLOIDOSIS / S. Ugurlu - B. Ergezen\* - O. Selvi - B. H. Egeli - H. Ozdogan
- P2074** ANTI-INTERLEUKIN-1 PRESCRIPTION IN ADULT PATIENTS WITH FAMILIAL MEDITERRANEAN FEVER: A REAL-LIFE STUDY FROM THE FRENCH NATIONAL REFERENCE CENTER / A. Fayand - L. Savey - V. Hentgen - P. Quartier - C. Bachmeyer - J. Galland - S. Amselem - C. Louvrier - I. Giurgea - G. Grateau - S. Georgin-Lavialle\* on behalf of French national reference center for autoinflammatory diseases
- P2075** EFFECTIVENESS OF ANTI-IL6 DRUG (TOCILIZUMAB) IN PULMONARY ALVEOLAR PROTEINOSIS ASSOCIATED TO SEVERE PERSISTENT SYSTEMIC INFLAMMATION: A CASE REPORT / I. Gueli\* - R. Caorsi - O. Sacco - D. Giroi - G. M. Magnano - A. Ravelli - M. Gattorno - P. Picco
- P2076** IS FIBRODYSPLASIA OSSIFICANS PROGRESSIVA AN INTERLEUKIN-1 DRIVEN AUTO-INFLAMMATORY SYNDROME? / R. Haviv\* - V. Moshe - N. Rabinowicz - G. Prencipe - F. De Benedetti - Y. Uziel
- P2077** KAWAKINRA: A PHASE IIA MULTICENTER TRIAL TO ASSESS THE EFFICACY, AND SAFETY OF ANAKINRA IN PATIENTS WITH INTRAVENOUS IMMUNOGLOBULIN-RESISTANT KAWASAKI DISEASE / I. Koné-Paut\* - S. Tellier - K. Brochard - V. Lambert - C. Guitton - A. Belot - P. Dusser - L. Rossi-Semerano - I. Marie - G. Allain - H. Agostini - C. Piedvache
- P2078** CONTINUOUS EFFECTIVENESS OF CANAKINUMAB TREATMENT IN SCHNITZLER'S SYNDROME: A 4-YEAR OPEN-LABEL MULTI-CENTER STUDY / K. Krause\* - H. Bonnekoh - A. Ellrich - A. Tsiannakas - N. Wagner - J. Fischer - M. Maurer
- P2079** LONG-TERM EFFECTIVENESS OF CANAKINUMAB IN PATIENTS WITH MONOGENIC PERIODIC FEVER SYNDROMES – FIRST INTERIM ANALYSIS OF THE CAPS SUBGROUP / J. B. Kuemmerle-Deschner\* - N. Blank - M. Borte - I. Foeldvari - G. Horneff - P. Oommen - C. Schuetz - F. Weller-Heinemann - J. Weber-Arden - T. Kallinich
- P2080** THE EFFECTIVENESS OF ANTI-TNF THERAPY IN ASSOCIATION OF FAMILIAL MEDITERRANEAN FEVER AND ANKYLOSING SPONDYLITIS / S. Atar - B. Colak - O. Kuru\*
- P2081** DESENSITIZATION TO ANAKINRA IN REFRACTORY RECURRENT PERICARDITIS / C. Longo\* - S. Signa - M. D'Alessandro - M. Bustaffa - R. Consolini - M. Tosca - L. Mendonça - A. Ravelli - R. Caorsi - M. Gattorno
- P2082** ONE DECADE REAL LIFE EXPERIENCE WITH ANTI-IL1 THERAPY IN MONOGENIC AND MULTIFACTORIAL AUTOINFLAMMATORY DISEASES / L. O. Mendonca\* - A. Guariento - R. Caorsi - S. Signa - I. Gueli - A. Ravelli - M. Gattorno
- P2083** A PROSPECTIVE OUTCOME OF ALL 17 CONSECUTIVE PEDIATRIC PATIENTS WITH CHRONIC NONBACTERIAL OSTEOMYELITIS TREATED WITH INTRAVENOUS PAMIDRONATE AND FOLLOWED AT A SINGLE CENTER OVER 15 YEARS / P. Miettunen\* - C. M. Stephenson - S. L. Stephenson - X.-C. Wei
- P2084** EFFICACY OF ANAKINRA IN THE TREATMENT OF RECURRENT IDIOPATHIC PERICARDITIS IN PEDIATRIC POPULATION: EXPERIENCE IN A TERTIARY HOSPITAL / S. Murias\* - L. García-Guereta - R. Alcobendas - D. Salas - C. Udaondo - P. Fernandez - C. Ferverza - A. Remesal
- P2085** CLINICAL MANIFESTATIONS AND TREATMENT OF IDIOPATHIC RECURRENT ASEPTIC MENINGITIS (IRAM) / M. Nehrebecky\* - D. Beck - A. Ombrello - A. Soldatos - B. Smith - P. M. Hoffman - L. Reoma - A. Nath - K. S. Barron - A. Jones - I. Aksentjevich - W. A. Gahl - C. Toro - D. L. Kastner
- P2086** AUTOINFLAMMATORY SYNDROME CAUSED BY MUTATIONS IN TRNT1: SUCCESSFUL TREATMENT OF TWO CASES WITH ETANERCEPT / F. Orlando\* - R. Naddei - C. M. Gallinoro - D. Melis - M. Alessio

## TUESDAY 02 APRIL 2019

- P2087** **DOES COLCHICINE REALLY CAUSE LEUKOPENIA?** / E. Sag - Y. Bayindir - A. Adiguzel - S. Demir - Y. Bilginer - S. Aytac - S. Ozen\*
- P2088** **MYCOPHENOLATE MOFETIL (MMF) IN DEFINED AND UNDEFINED INTERFERONOPATHIES** / C. G. L. Raffaele\* - G. Moneta - S. Federici - M. Pardeo - C. Bracaglia - F. De Benedetti - A. Insalaco
- P2089** **SUCCESSFUL IL-17 TARGETED THERAPY IN A PATIENT WITH SEVERE BLAU SYNDROME AND NOVEL NOD2 MUTATION** / N. Rieber\* - M. Steinhauser - M. Klopfer - S. Pietzsch - P. Strotmann - H. Engelhardt - U. Behrends - R. Berendes - S. Burdach
- P2090** **RECURRENT FEVERS ASSOCIATED WITH NEURODEVELOPMENTAL DISORDERS AND THEIR TREATMENT WITH ANAKINRA** / T. M. Romeo\* - A. K. Ombrello - K. Barron - A. Jones - S. Berger - A. C. Smith - D. L. Kastner - D. L. Stone
- P2091** **HEMATOPOIETIC STEM CELL TRANSPLANTATION (HSCT) SUCCESSFULLY CURED TWO PATIENTS WITH PSTPIP1-ASSOCIATED AUTOINFLAMMATORY DISEASES (PAMI)** / A. Laberko - D. Yukhacheva - I. Shipitsina - M. Dunaykina - A. Kozlova - V. Burlakov - I. Mersiyanova - Y. Rodina - E. Raykina - D. Balashov - L. Shelikhova - A. Shcherbina\*
- P2092** **RESPONSE TO JAK INHIBITION IN TWO CHILDREN WITH HAPLOINSUFFICIENCY OF A20 (HA20) CAUSED BY TRUNCATING MUTATIONS IN THE ZNF4 DOMAIN** / D. L. Stone\* - A. K. Ombrello - K. Barron - T. Romeo - N. Sampaio Moura - I. Aksentijevich - D. L. Kastner
- P2093** **SQUALENE SYNTHASE INHIBITORS TO REGULATE THE INFLAMMATION IN MEVALONATE KINASE DEFICIENCY** / A. Marcuzzi - E. Piscianz - E. Valencic - A. Tesser\* - A. Taddio - A. Tommasini
- P2094** **IL1B BLOCKADE BY CANAKINUMAB LEADS INTO REMISSION RECURRENT PERICARDITIS ASSOCIATED WITH THE MEFV VARIANT P.GLU148GLN (E148 Q)** / M. Tsinti\* - M. Zamanakou - V. Dermentzoglou - E. Tsitsami
- P2095** **ANAKINRA TREATMENT IN RECURRENT PERICARDITIS: SINGLE CENTER EXPERIENCE** / Z. Toker Dincer - O. Corbali - S. Ugurlu\* - H. Ozdogan

### POSTER VIEWING - IMMUNEDYSREGULATION

- P2096** **INAUGURAL HEMOPHAGOCYTIC SYNDROME IN AUTOIMMUNE DISEASES: A CASE SERIES** / L. Damian\* - M. Sfichi - M. Lupse - S. Cocu - L. Urian - C. Pamfil - M. Zdrengea - I. Kacso - S. Rednic - A. Bojan
- P2097** **ASSESSING THE MUTATION DETECTION RATE AND GENOTYPE-PHENOTYPE CORRELATION WITH A LARGE HAEMATO-IMMUNOLOGICAL NGS PANEL: CROSSING THE BORDERS AMONG DIFFERENT SUBSPECIALTIES** / A. Grossi\* - M. Miano - S. Volpi - R. Caorsi - F. Fioredda - M. Lanciotti - E. Capelli - C. Dufour - M. Gattorno - I. Ceccherini
- P2098** **VITAMIN D STATUS & ITS THERAPEUTIC ROLE IN PEDIATRIC PATIENTS WITH IDIOPATHIC NEPHROTIC SYNDROME** / D. E. Sallam\*
- P2099** **IMMUNE RECONSTITUTION IN AUTOINFLAMMATORY CANDLE LIKE SYNDROME DUE TO SAMD9L MUTATION** / A. G. Seminario\* - M. S. Caldirola - I. Moreira - V. Regairaz - M. Gattorno - R. Goldbach-Mansky - L. Bezrodnik
- P2100** **FASL GENE MUTATION IN A CHILD WITH AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME** / M. Sharma\* - A. Rawat - D. Suri - V. Pandiarajan - K. W. Chan - Y. L. Lau - S. Singh
- P2101** **CHARACTERISTICS OF THE IMMUNOLOGICAL STATUS IN PATIENTS WITH SYSTEMIC AND ARTICULAR FORM OF JUVENILE IDIOPATHIC ARTHRITIS** / A. Y. Spivakovskaya\* - Y. Spivakovskiy - Y. Chernenkov

## TUESDAY 02 APRIL 2019

## POSTER VIEWING - UNUSUAL OR UNSOLVED CASE REPORTS

- P2102** **EARLY ONSET BEHÇETS DISEASE IN AN INDIAN FAMILY: IS IT RELATED TO A20 HAPLOINSUFFICIENCY?** / V. Pandiarajan\* - D. Suri - A. Rawat - A. Gupta - S. Singh
- P2103** **TWO TYPES OF SYSTEMIC AMYLOIDOSIS IN A SINGLE PATIENT** / R. Papa\* - J. A. Gilbertson - N. Rendell - A. D. Wechalekar - J. D. Gillmore - P. N. Hawkins - H. J. Lachmann
- P2104** **MAFB NOT SEEMS TO REGULATE SERUM C1Q CONCENTRATION IN HUMANS** / R. Papa\* - A. Madeo - S. Volpi - R. Caorsi - G. Barbano - M. Botto - A. Superti-Furga - M. Gattorno - M. Di Rocco - P. Picco
- P2105** **SIBSHIP WITH UVEITIS AND SENSORINEURAL HEARING LOSS** / P. Pimpale Chavan\* - M. Morekar - R. P. Khubchandani
- P2106** **AN UNSOLVED CASE INCLUDING GRANULOMATOUS PANUVEITIS FROM INDIA** / P. Pimpale Chavan\* - M. Kothari - R. P. Khubchandani
- P2107** **AN UNSOLVED CASE: IS THIS A CANDLE-LIKE SYNDROME?** / A. Pin\* - A. Tesser - F. Faletra - A. Tommasini - S. Pastore - A. Taddio
- P2108** **A CASE OF STING-ASSOCIATED VASCULOPATHY WITH ONSET IN INFANCY (SAVI) WITHOUT SKIN INVOLVEMENT** / C. G. L. Raffaele\* - V. Messia - M. Pardeo - C. Celani - S. Federici - G. Moneta - I. Caiello - C. Bracaglia - F. De Benedetti - A. Insalaco
- P2109** **A BOY WITH SEVERE PHENOTYPE OF IMMUNE DEFICIENCY AND RELAPSING HYPERINFLAMMATION CURED WITH A HEMATOPOIETIC STEMCELL TRANSPLANTATION** / K. Ramme\* - Y. Bryceson - M. Sundin - S. Vonlanthen - A. Horne
- P2110** **TWO GENES AND ONE PHENOTYPE: SAME THERAPY?** / S. Ricci\* - I. Pagnini - E. Sieni - C. Azzari - M. Cortimiglia - G. Mangone - L. Lodi - G. Mangone - M. Cortimiglia - E. Calistri
- P2111** **AUTOINFLAMMATORY PHENOTYPE AS A MASK OF ONCOLOGICAL PATHOLOGY** / E. Fedorov - S. Salugina\*
- P2112** **COEXISTENCE OF FAMILIAL MEDITERRANEAN FEVER WITH CELIAC DISEASE IN ARMENIAN PATIENT (CASE REPORT)** / H. Sargsyan\*
- P2113** **INFLAMMATION ARISING ON COMBINED GENETIC BACKGROUND CHARACTERIZED BY SIMULTANEOUS OCCURRENCE OF NOD2 AND IFNGR1 MUTATIONS** / A. Sediva\* - Z. Parackova - M. Bloomfield - A. Klocperk - I. Zentsova - M. Svaton - E. Froňková
- P2114** **RELAPSING-REMITTING SYSTEMIC AND NEUROLOGICAL SYMPTOMS IN GRISCELLI DISEASE** / A. Tozzo\* - N. Milani - G. Marucci - L. Chiapparini - A. Erbetta - E. Sieni - N. Nardocci
- P2115** **DEFICIENCY OF ADENOSINE DEAMINASE 2: THE IMPORTANCE OF A DIAGNOSIS BEFORE IRREVERSIBLE DAMAGE** / E. Tronconi\* - F. Conti - D. M. Cordelli - S. Volpi - M. Gattorno - A. Pession - A. Miniaci
- P2116** **THE CASE STUDY OF UNDIFFERENTIATED AUTOINFLAMMATORY DISEASE TREATED WITH ANAKINRA** / M. Visnevska\* - A. Meiere - Z. Davidsova - R. Santere - V. Stanevica
- P2117** **ETANERCEPT IN THE TREATMENT OF FAMILIAL MEDITERRANEAN FEVER: A CASE REPORT** / Y. Zahiroglu\*

## TUESDAY 02 APRIL 2019

### POSTER VIEWING - NEW MONOGENIC DISEASES

- P2118**      **A CASE OF NEONATAL-ONSET PROTEASOME-ASSOCIATED AUTOINFLAMMATION AND IMMUNODEFICIENCY DISEASE RESEMBLING BUT DISTINCT FROM NAKAJO-NISHIMURA SYNDROME** / N. Kinjo\* - S. Hamada - K. Nakanishi - H. Mishima - A. Kinoshita - K.-I. Yoshiura - T. Mizushima - J. Hamazaki - S. Murata - H. Hemmi - T. Kaisho - N. Kanazawa - D. Dima - A. Albu
- P2119**      **PROTEASOME-ASSOCIATED AUTOINFLAMMATORY SYNDROME 2 (PRAAS2) IN A FIVE MONTHS OLD BOY** / A. Kozlova\* - E. Viktorova - V. Zaharova - D. Konovalov - A. Terentjeva - V. Burlakov - E. Raikina - N. Kuzmenko - J. Rodina - A. Maschan - A. Shcherbina
- P2120**      **A CASE OF NOVEL IDENTIFIED PROTEASOME-RELATED AUTOINFLAMMATION AND IMMUNODEFICIENCY SYNDROME CAUSED BY PSMB9 MUTATION** / H. Ohnishi\* - S. Kataoka - H. Muramatsu - E. Kadoi - N. Kanazawa - S. Okada - Y. Honda - K. Izawa - R. Nishikomori - T. Taketani - J. Hamazaki - S. Murata - Y. Takahashi - T. Fukao
- P2121**      **'NON-SELF' MUTATION: DOUBLE-STRANDED RNA ELICITS ANTIVIRAL PATHOGENIC RESPONSE IN A DROSOPHILA MODEL OF EXPANDED CAG REPEAT NEURODEGENERATIVE DISEASES** / R. Richards\* - S. Samaraweera - A. Scott - D. Webber - D. Harvey - O. Mecinger - L. O'Keefe - J. Cropley - P. Young - J. Ho - C. Suter

### POSTER VIEWING - I HAVE A POSSIBLE GENE OR A STRANGE PHENOTYPE. LOOKING FOR A SECOND CASE

- P2122**      **A FAMILY WITH SAVI-LIKE VASCULOPATHY, ARTHRITIS AND COLD-INDUCED INFLAMMATORY URTICARIAL RASH** / E. Conversano\* - E. Piscianz - E. Valencic - A. Tesser - A. Tommasini - S. Pastore - V. Moressa - A. Omenetti - B. Lattanzi - G. Gortani - A. Taddio
- P2123**      **NEW ATM VARIANTS AND A RARE ASSOCIATION OF JUVENILE IDIOPATHIC ARTHRITIS AND ATAXIA-TELANGIECTASIA: COINCIDENCE OR RELATIONSHIP?** / N. Kifer\* - T. Arsov - M. Sestan - M. Frkovic - C. G. Vinuesa - M. Jelusic
- P2124**      **A CASE OF DRUG-INDUCED UNDIFFERENTIATED CONNECTIVE TISSUE DISEASE IN A 2 YEARS OLD GIRL AS A MANIFESTATION OF MUNCHAUSEN SYNDROME BY PROXY** / T. Kovalchuk\*
- P2125**      **SEVERE LARGE VESSELS VASCULITIS WITH TOE GANGRENE IN A YOUNG CHILD** / C. Matucci Cerinic\* - R. Caorsi - S. Volpi - S. Rosina - R. Papa - P. P. Picco - A. Ravelli - M. Gattorno
- P2126**      **IS THIS DISEASE RELATED TO NLRP3 OR MEFV OR BOTH?** / A. Paç Kisaarslan\* - N. Şahin - S. Özdemir Çiçek - A. Berdeli - Z. Gündüz - H. Poyrazoğlu - R. Düşünsel
- P2127**      **M1184V MUTATION IN NLRP1: IS THAT POLYMORPHISM OR PATHOGENIC MUTATION?** / N. Sahin\* - S. O. Cicek - A. Berdeli - A. P. Kisaarslan - Z. Gündüz - M. H. Poyrazoglu - R. Düşünsel
- P2128**      **PERIODIC FEVER SYNDROME WITH NOVEL TRNT1 VARIANT - POSSIBLE CAUSE OF TRNT1 DEFICIENCY OR JUST AN INCIDENTAL FINDING?** / M. Sestan\* - T. Arsov - N. Kifer - M. Frkovic - C. G. Vinuesa - M. Jelusic
- P2129**      **A NOVEL AUTOINFLAMMATORY AND LYMPHOPROLIFERATIVE SYNDROME ASSOCIATED WITH PIM1 MUTATIONS** / G. Ferrara - S. Polizzi - E. Valencic - A. Chiocchetti - J. Vuch - A. Pin - E. Piscianz - D. Vozzi - S. Pastore - P. Tomietto - A. Taddio\* - F. Faletra - U. Dianzani - A. Tommasini

## TUESDAY 02 APRIL 2019

- |                      |  |                                    |
|----------------------|--|------------------------------------|
| <b>16:00 - 17:15</b> | <b>COMPLEMENT AND AUTOINFLAMMATION</b><br><i>Chairpersons: Paul Brogan (UK) &amp; Anna Simon (NL)</i>  | <b>MAESTRALE ROOM</b>              |
| <b>16:00</b>         | <b>Autoinflammation and complement, an overview</b><br>Paul Brogan (UK)  |                                    |
| <b>16:20</b>         | <b>Complement deficiency and inflammation</b><br>Marina Botto (UK)   |                                    |
| <b>16:40</b>         | <b>Complement pathway as a therapeutic target for autoinflammation</b><br>Aoife Waters (UK)  |                                    |
| <b>17:00</b>         | <b>General discussion</b>  |                                    |
| <b>16:00 - 17:15</b> | <b>GENETIC DIAGNOSIS OF SAIDS:<br/>TOWARDS CONSENSUAL LARGE-SCALE APPROACHES<br/>AND GUIDELINES</b><br><i>Chairpersons: Raffaella Manna (IT) &amp; Isabelle Touitou (FR)</i> | <b>SCIROCCO-<br/>LIBECCIO ROOM</b> |
| <b>16:00</b>         | <b>Classification of SAID genetic variants pathogenicity and updating guidelines for genetic diagnostic</b><br>Marielle van Gijn (NL)  |                                    |
| <b>16:20</b>         | <b>Mosaicism in diagnostics, NLRP3 and beyond</b><br>Juan Arostegui (ES)   |                                    |
| <b>16:40</b>         | <b>WES versus panel and sanger in diagnostics: is it time to switch?</b><br>Isabella Ceccherini (IT)   |                                    |
| <b>17:00</b>         | <b>General discussion</b>  |                                    |
| <b>17:15 - 18:30</b> | <b>THIEF MARKET</b><br>Eldad Ben Chetrit (IL) & Joost Frenkel (NL)   | <b>MAESTRALE ROOM</b>              |

## WEDNESDAY 03 APRIL 2019

- 07:30 - 08:30**    **WAKE UP EARLY AND FIND A NEW DISEASE!**    **MAESTRALE ROOM**  
*Discussion led by Marco Gattorno (IT) and Ivona Aksentijevich (US)*
- 08:30 - 10:00**    **NEW FRONTIERS IN THE TREATMENT OF SAID**    **MAESTRALE ROOM**  
*Chairpersons: Marco Gattorno (IT) & Ahmed Gül (TR)*
- 08:30**    **An update on treatment in SAID**  
 Helen Lachmann (UK)
- 08:55**    **BMT in SAID and immune dysregulation**  
 Andrew Cant (UK)
- 09:20**    **Gene therapy**  
 Alessandro Aiuti (IT)
- 09:45**    **O29 - Preclinical studies of gene therapy for deficiency of adenosine deaminase type 2 (DADA2)**  
 Ying Hong (UK)
- 09:52**    **O30 - Experience with and management of HLH-like toxicities following chimeric antigen receptor T-cell therapy for treatment of relapsed/refractory pre-B ALL**  
 Amanda Kilker Ombrello (US)
- 10:00 - 10:30**    **COFFEE BREAK & VISIT OF THE EXHIBITION**    **EXHIBITION AREA**
- 10:30 - 12:00**    **NOVEL TARGETS AND THERAPIES IN AUTOINFLAMMATION**    **MAESTRALE ROOM**  
*Chairpersons: Dirk Foell (DE) & Nicola Ruperto (IT)*
- 10:30**    **New targets for autoinflammatory diseases**  
 Ahmed Gül (TR)
- 10:55**    **Novel therapeutic approaches to Type-1 interferons**  
 Raphaela Goldabch-Mansky (US)
- 11:20**     **$\gamma$ -interferon and IL-18**  
 Fabrizio de Benedetti (IT)
- 11:45**    **O31 - Novel NLRP3 targeted therapy in CAPS**  
 Hal M. Hoffman (US)
- 11:50**    **O32 - Preclinical efficacy of NLRP3 small molecule inflammasome inhibitors: implications for future treatment of autoinflammatory syndromes**  
 Angela Abad-Perez (DE)
- 12:00 - 13:00**    **ORAL COMMUNICATIONS – NEW DISEASES**    **MAESTRALE ROOM**  
*Chairpersons: Ivona Aksentijevich (US) & Daniel Kastner (US)*
- 12:00**    **O33 - Biallelic loss of function mutations in sharpin cause autoinflammation**  
 Hirotsugu Oda (US)
- 12:07**    **O34 - A loss-of-function mutation in USP43, a deubiquitinase gene, is linked to an interferon-mediated autoinflammatory disorder with proteasome defects**  
 Hongying Wang (US)



## WEDNESDAY 03 APRIL 2019

- 12:14 **O35 - A novel autoinflammatory disease characterized by neonatal-onset cytopenia with autoinflammation, rash, and hemophagocytosis (NOCARH) due to aberrant CDC42 function**  
Antonella Insalaco (IT)
- 12:21 **O36 - PSMB10, the last immunoproteasome gene missing for PRAAS (proteasome-associated autoinflammatory syndrome)**  
Guillaume Sarrabay (FR)
- 12:28 **O37 - WNT6 mutation causes an early onset granulomatous intestinal disease with recurrent hemophagocytic lymphohistiocytosis (HLH)**  
Claudia Bracaglia (IT)
- 12:35 **O38 - NFIL3 mutations alter immune homeostasis and sensitize for arthritis pathology**  
Stephanie Humblet-Baron (BE)
- 12:42 **O39 - EROS/CYBC1 mutations: a novel cause of chronic granulomatous disease and more**  
David C. Thomas (UK)
- 12:49 **O40 - Cold-induced urticarial autoinflammatory syndrome related to factor XII activation**  
Karoline Krause (DE)
- 13:00 - 13:30 **CLOSING AND YOUNG INVESTIGATOR AWARD PRESENTATION** **MAESTRALE ROOM**

# PATIENT ORGANISATIONS MEETING

This event is supported by Novartis and ISSAID.

## PROGRAMME - SUNDAY 31 MARCH 2019

- 08:30 - 09:00**    **WELCOME FROM ORGANISING COMMITTEE, AND KEYNOTE SPEECH FROM ISSAID**    **BORA ROOM**
- Welcome**  
Marco Gattorno (IT)
- Keynote Speech**  
Daniel Kastner (US)
- Topics:**  
How far that we have come in the past 20-30 years with autoinflammatory diseases.  
What are the goals of ISSAID, and how patient orgs can help?  
Where are the aspirations of ISSAID for the next 5-10 years?
- 09:00 - 10:00**    **A. IMPACT OF DISEASE ON PATIENTS THROUGHOUT THE LIFESPAN, PRESENTED BY ISSAID DOCTORS**    **BORA ROOM**
- 09:00**    **Psychosocial, work, education, peer/social life, relationships**  
Jasmin Kümmerle-Deschner (DE)
- 09:20**    **Reproductive years/ family planning**  
Helen Lachmann (UK)
- 09:40**    **Group discussion:**  
-How to address unmet needs and improve access to help with these patient challenges?
- 10:00 - 10:45**    **B. HOW TO HELP PATIENTS COMMUNICATE WITH DOCTORS, PEERS, TEACHERS, RELATIVES, ETC. ABOUT THEIR DISEASE, FROM CHILDHOOD TO ADULTHOOD**    **BORA ROOM**
- 10:00**    **Empowered patients: How to explain your disease in terms for peers to understand**  
Sivia Lapidus (US)
- 10:15**    **The “dos and don’ts” of communication with doctors: Planning for doctor visits, organising medical info, thoughts and questions to ask, etc.**  
Sivia Lapidus (US) & Karen Durrant (US)
- 10:30**    **Group discussion:**  
-What materials do patient organizations have  
-What works and what do patients still need?  
-Patient support group outreach, online information & social media best practices  
Marta Campabadal from Rare Connect (EURORDIS initiative)
- 10:45 - 11:00**    **COFFEE BREAK**    **BAR AREA**
- 11:00 - 11:30**    **C. COPING SKILLS, EDUCATION AND WORK/CAREER SUPPORT FOR PATIENTS**    **BORA ROOM**
- 11:00**    **Resilience strategies & how to seek help for psychosocial needs**  
Fatma Dedeoglu (US)
- 11:15**    **Transition to adult care. Work & school accommodations**  
Anna Simon (NL)

## PROGRAMME - SUNDAY 31 MARCH 2019

11:30	<b>Group discussion:</b> -Helpful resources, and what more is still needed for patients?	
11:45-12:30	<b>NETWORKING LUNCH</b>	<b>BAR AREA</b>
12:30 - 13:30	<b>A CALL TO ACTION: OPEN FORUM DISCUSSION FOR ALL ATTENDEES, LED BY THE ISSAID STEERING COMMITTEE</b> Marco Gattorno (IT), Helen Lachmann (UK), Paul Brogan (UK), Erkan Demirkaya (CA) & Daniel Kastner (US)	<b>BORA ROOM</b>
12:30	<b>Next Steps: How to begin to address the unmet needs discussed at the end of each morning session (A, B, C), and on this effort.</b> -Selection of the top 3 action items to address collaboratively before the next ISSAID Congress	
12:45	<b>Commitment/ Plan for when/how to tackle the 3 action items</b>	
13:00	<b>Open discussion: How patient organisations can support ISSAID &amp; autoinflammatory needs in general</b>  Discussion on how to have patient organisation representation with ISSAID: -Guidelines for the involvement of patient organisations with ISSAID -Qualifications for patient organisation representatives for ISSAID -Democratic process for determining ISSAID patient organisations' representatives  Research (fundraising, directing patients to research, etc) Outreach and awareness efforts that organisations can partner on with ISSAID globally	
13:30 - 13:35	<b>CLOSING THANKS AND REMARKS-FROM THE ISSAID ORGANISING COMMITTEE</b>	<b>BORA ROOM</b>
13:35 - 14:00	<b>SOCIALISING AND NETWORKING-DISTRIBUTION OF THANK YOU GIFTS TO ATTENDEES</b>	<b>BORA ROOM</b>

**MONDAY 01 APRIL 2019**

Organised by:



<b>12:30 - 13:30</b>	<b>NEW INSIGHTS FOR THE MANAGEMENT OF STILL'S DISEASE</b>	<b>MAESTRALE ROOM</b>
<b>12:30</b>	<b>Introduction</b> Marco Gattorno (IT)	
<b>12:35</b>	<b>Management of SJIA: Challenges and opportunities</b> Jordi Antón (ES)	
<b>12:45</b>	<b>Management of AOSD: Challenges and opportunities</b> Sinisa Savic (UK)	
<b>12:55</b>	<b>Case reports on Still's patients</b> Sophie Georgan-Lavialle (FR)	
<b>13:05</b>	<b>Debate/Q&amp;A session</b> All	
<b>13:25</b>	<b>Closing remarks</b> Marco Gattorno (IT)	

**TUESDAY 02 APRIL 2019**

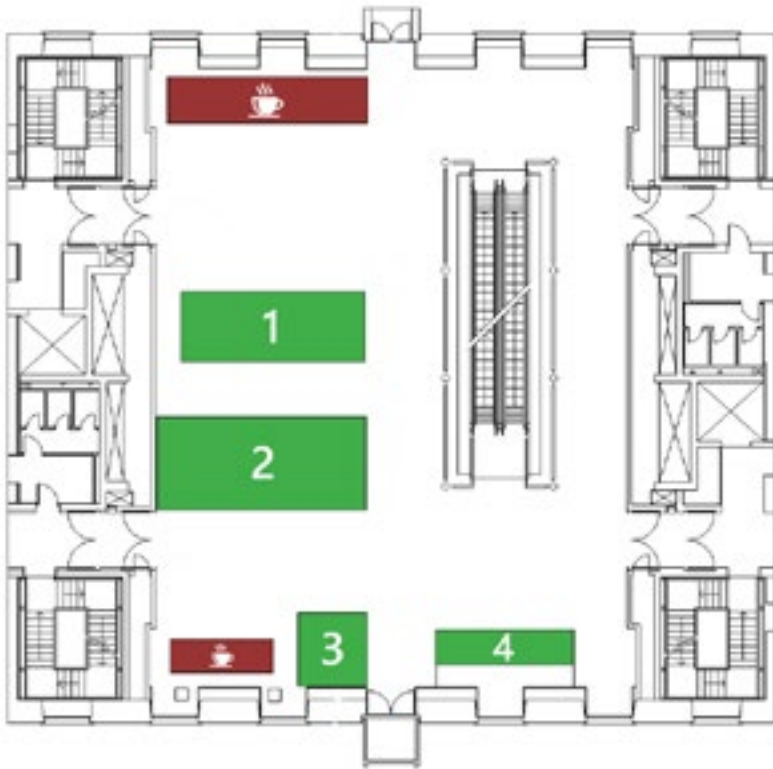
Organised by:



<b>12:30 - 13:30</b>	<b>TARGETING REMISSION IN AUTO-INFLAMMATORY DISEASES WITH ANTI-IL-1<math>\beta</math></b>	<b>MAESTRALE ROOM</b>
<b>12:30</b>	<b>Welcome</b> Marco Gattorno (IT)	
<b>12:35</b>	<b>Timing the therapeutic window of opportunity in SJIA: Can we predict a response?</b> Dirk Föll (DE)	
<b>12:50</b>	<b>Drug-free remission in SJIA: A potential treat-to-target strategy?</b> Ekaterina Alexeeva (RU)	
<b>13:05</b>	<b>Long-term disease control in FMF: A realistic goal?</b> Fabrizio de Benedetti (IT)	
<b>13:20</b>	<b>Putting the data into perspective: What does remission mean for patients?</b> Panel Discussion and Audience Q&A	

# EXHIBITION LAYOUT

## MODULO N°8



COMPANY NAME	BOOTH N°
Sobi	1
Novartis	2
Novartis	3
Octapharma	4
JIR Cohort	5
PRINTO	6
Coming Together for Autoinflammatory Diseases	7

## MODULO N°9



## NOVARTIS



Novartis is reimagining medicine to improve and extend people's lives. As a leading global medicines company, we use innovative science and digital technologies to create transformative treatments in areas of great medical need. In our quest to find new medicines, we consistently rank among the world's top companies investing in research and development.

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Find out more at [www.novartis.com](http://www.novartis.com)

## SOBI



Sobi™ is an international speciality healthcare company dedicated to rare diseases. Our vision is to be recognised as a global leader in providing access to innovative treatments that transform lives for individuals with rare diseases.

The product portfolio is primarily focused on treatments in Haemophilia and Specialty Care. Partnering in the development and commercialisation of products in specialty care is a key element of our strategy. Sobi has pioneered in biotechnology with world-class capabilities in protein biochemistry and biologics manufacturing.

## OCTAPHARMA



Octapharma is one of the largest human protein manufacturers in the world, developing and producing human proteins from human plasma and human cell lines.

As a family-owned company, Octapharma believes in investing to make a difference in people's lives and has been doing so since 1983.

Octapharma employs around 8 000 people worldwide to support the treatment of patients in 113 countries with products across three therapeutic areas as haematology, immunotherapy and critical care. Octapharma has seven R&D sites and six state-of-the-art manufacturing facilities in Austria, France, Germany, Mexico and Sweden.





# Join us for the 11<sup>th</sup> ISSAID Congress in Toronto, in April 2021

Stay tuned for more information on: [www.issaid.org](http://www.issaid.org)





# ACKNOWLEDGEMENTS

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# CHANGE

YOUR PATIENT'S LIFE WITH ILARIS®<sup>1-3</sup>

**ILARIS® : is indicated for the treatment of FMF, HIDS/MKD and TRAPS<sup>4</sup>**

**ILARIS®: biological therapy indicated for the treatment of active Still's disease<sup>4</sup>**

**Periodic fever syndromes.** ILARIS® is indicated for the treatment of the following autoinflammatory periodic fever syndromes in adults, adolescents and children aged 2 years and older: **Cryopyrin-associated periodic syndromes.** ILARIS® is indicated for the treatment of **CAPS** including: **Muckle-Wells syndrome (MWS), Neonatal-onset multisystem inflammatory disease (NOMID)/chronic infantile neurological, cutaneous, articular syndrome (CINCA), Severe forms of familial cold autoinflammatory syndrome (FCAS)/familial cold urticaria (FCU)** presenting with signs and symptoms beyond cold-induced urticarial skin rash. **Tumour necrosis factor receptor associated periodic syndrome (TRAPS)** ILARIS® is indicated for the treatment of tumour necrosis factor (TNF) receptor associated periodic syndrome (TRAPS). **Hyperimmunoglobulin D syndrome (HIDS)/mevalonate kinase deficiency (MKD)** ILARIS® is indicated for the treatment of **hyperimmunoglobulin D syndrome (HIDS)/mevalonate kinase deficiency (MKD) Familial Mediterranean fever (FMF)**. ILARIS® is indicated for the treatment of Familial Mediterranean Fever (FMF). ILARIS® should be given in combination with colchicine, if appropriate. ILARIS® is also indicated for the treatment of **Still's disease. ILARIS® is indicated for the treatment of active Still's disease including adult-onset Still's disease (AOSD) and systemic juvenile idiopathic arthritis (SJIA)** in patients aged 2 years and older who have responded inadequately to previous therapy with non-steroidal anti-inflammatory drugs (NSAIDs) and systemic corticosteroids. ILARIS® can be given as monotherapy or in combination with methotrexate. **Gouty arthritis.** ILARIS® is indicated for the symptomatic treatment of adult patients with frequent gouty arthritis attacks (at least 3 attacks in the previous 12 months) in whom non-steroidal anti-inflammatory drugs (NSAIDs) and colchicine are contraindicated, are not tolerated, or do not provide an adequate response, and in whom repeated courses of corticosteroids are not appropriate.

**References:** 1. Ruperto N, et al. NEJM 2012; 367(25):2396-2406. 2. Junge G, et al. Semin Arthritis Rheum 2017; 47(2):295-302. 3. De Benedetti F, et al. N Engl J Med 2018; 378:1908-1919. 4. Ilaris® (canakinumab). SmPC.

Gouty arthritis is not reimbursed in Italy.

**TRAPS | FMF | CAPS | HIDS/MKD**

**PERIODIC FEVER SYNDROMES**

**SJIA | AOSD**

**STILL'S DISEASE**

**GOUTY ARTHRITIS**

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