17 - 18 SEPTEMBER 2026

IWRAS 26
INTERNATIONAL WORKSHOP

International Workshop on RASopathies

Strengthening Translational Collaboration between Research, Clinical Practice and Patient Communities

RESEARCH | CLINICAL PRACTICE | PATIENT COMMUNITIES

VENUE

Rectorate of the University of Málaga

Avenida de Cervantes, 2 / Málaga, Spain

- **Scientific programme**

WELCOME TO

IWRAS26

"What happens in the lab must translate into better care in the clinic and real support at home."

THE IWRAS26
ORGANISING COMMITTEE

WELCOME MESSAGE

Dear friends, colleagues & families,

A warm welcome to the International Workshop on RASopathies: Strengthening Translational Collaboration between Research, Clinical Practice and Patient Communities (IWRAS26).

RASopathies are complex conditions that affect individuals and their families far beyond the medical symptoms. Finding answers and developing effective treatments is a journey that requires all of us working together. That is why this workshop is so important to us: it brings together under one roof the scientists pushing the boundaries of research, the clinicians caring for patients every day, and the families who live with these conditions and inspire our work.

Over the next few days, we will discuss the latest scientific breakthroughs, from genetic discoveries to new therapeutic approaches. But beyond the data, what we truly want is to connect these different worlds. We want to ensure that what happens in the lab translates directly into better care in the clinic and real support at home.

Thank you so much for being here in Málaga and for sharing your knowledge, your time, and your stories. Whether you are presenting a study, sharing clinical expertise, or bringing the invaluable perspective of a parent, your presence makes this collaboration possible.

We hope you enjoy the workshop and the meaningful conversations ahead.

With our warmest regards,

THE IWRAS26 ORGANISING COMMITTEE

DAY 01

Thursday, September 17

Opening session / Patient & caregiver perspective

09:00 - 09:15	**Opening Session** Welcome remarks by Local and Institutional Authorities.
09:15 - 09:35	**Inaugural Conference** Bioinformatics: Essential support for rare disease diagnosis, clinical assays and treatment. Dr. Juan A. G. Ranea (IBIMA-Rare, UMA, Spain)
09:35 - 11:15	**Session 1: Patient and Caregiver Perspective** *Chair: Dr. Marina Calleja Reina (UMA, Spain)*
09:35 - 09:55	**From Patient Idea to EU Consortium and how we Learned to Value Registries as the Key to Future Therapies** Marcos Mengual Hinojosa (Father and patient advocate EURAS, EU)
09:55 - 10:15	**SYNGAP1 Advocacy: Challenges, progress and impact.** Dr. Jaime Aranda Durán (Asociación SYNGAP1 España)
10:15 - 10:35	**Living with Rare Diseases: Challenges faced by families.** Dr. Jesús M. Bonill Jiménez (UMA, Spain)
10:35 - 10:55	**Overview of computational methods for drug repurposing.** Dr. Florencio Pazos (CNB-CSIC, Spain)
10:55 - 11:15	**Managing Regression: The families' perspective.** Dr. Encarnación Postigo Pinazo (UMA, Spain)
11:15 - 11:45	**Coffee Break & Networking**

DAY 01

Thursday, September 17

Round tables / Research & future prospects

11:45 - 13:30 — **Round Table 1: Needs and Advances in Patient Care**
An open panel discussion with speakers, clinicians, researchers, and families addressing educational hurdles, psychosocial support, and standardizing multidisciplinary care pathways.

13:30 - 15:00 — **Lunch Break**

15:00 - 17:30 — **Session 2: Research & Future Prospects**
Chair: Dr. Fernando Cabrera Bueno (HUVVM, Spain)

15:00 - 15:20 — **Redefining Clinical Endpoints in SYNGAP1-DEE: From the Patient-Centered PATRE Registry to Neurobehavioral Insights**
Lorenz Kiwull, M.D. (PMU, Salzburg, Austria)

15:20 - 15:40 — **SYNGAP1 Haploinsufficiency: Developmental consequences.**
Dr. María Isabel Carreño Muñoz (Montreal, Canada)

15:40 - 16:00 — **UNICAS-IMPaCT: National pediatric network for personalized medicine.**
MsC. Helena Rodríguez González (HSJD, Spain)

16:00 - 16:20 — **RASopathies in neurology: from intracellular signaling to epileptogenesis.**
Dr. Jorge Romero Godoy (Hospital Virgen de la Victoria, Spain)

16:20 - 16:40 — **Costello Syndrome: Modeling and treating with CRISPR iPSCs.**
MsC. Joana Jeremias (IBB, Portugal)

16:40 - 17:30 — **Round Table 2: Future Perspectives on RASopathies Research**
Strategic discussion focused on multi-omics integration, funding frameworks, international registries, and cooperative clinical trial design.

DAY 02

Friday, September 18

Keynote lecture / Translational activities / Closing

09:00 - 09:45	**Keynote Lecture** A gene therapy approach to correcting SYNGAP1-related disorder. Dr. Boaz P. Levi (Allen Institute, Seattle, U.S.A.)
09:45 - 11:05	**Session 3: Translational Activities** *Chair: Dr. Raquel Yahyaoui Macías (IBIMA-Rare, Spain)*
09:45 - 10:05	**Establishing Patient-Reported Outcome Measures (PROMs) for Medication Management in SYNGAP1-DEE: Insights from the PATRE Registry.** Kirsten Eschermann, M.D. (PMU, Salzburg, Austria)
10:05 - 10:25	**AI in Diagnosis: Accurate phenotyping approaches in RASopathies.** Dr. María Elena Rojano Rivera (UMA, Spain)
10:25 - 10:45	**Bioinformatics analysis of rare-disease cohorts: Applications in RASopathies/SYNGAP1.** Dr. James Richard Perkins (UMA, Spain)
10:45 - 11:05	**Artificial Intelligence: Future perspectives and clinical decision support systems.** Julián Isla (Father and patient advocate, Microsoft / Fundación 29)
11:05 - 11:30	**Coffee Break**
11:30 - 13:00	**Round Table 3: Translational Activities for RASopathies** Discussion on present and future of translational activities for RASopathies and their associated difficulties.
13:00 - 13:30	**Conclusions and Workshop Closure** SYNGAP1 in Europe: The Way Forward. Marcos Mengual Hinojosa (Father and patient advocate EURAS, EU)

Workshop interpreter: Dr Laura Parrilla Gómez (UPO, Spain)

PEOPLE BEHIND IWRAS26

Scientific & organising committee

A multidisciplinary team connecting science, care and community.

01 **Dr. Encarnación Postigo Pinazo**

02 **Dr. Francisca Sánchez Jiménez**

03 **Dr. Juan A. G. Ranea**

04 **Dr. María Elena Rojano Rivera**

05 **Dr. Manuel Bernal Muñoz**

06 **Dr. Raquel Yahyaoui Macías**

07 **Dr. James Richard Perkins**

08 **Dr. Pedro Seoane Zonjic**

SPONSORS & COLLABORATORS

Progress is a shared effort.

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EURAS

EURAS Network

European network for RASopathies research and patient collaboration.