



22 y 23 de FEBRERO de 2026

# I Congreso Internacional Mutación Gen IQSEC2 España

en **MADRID**

 Hospital Universitario Ramón y Cajal



**Hospital Universitario  
Ramón y Cajal**

**feder**  
FEDERACIÓN ESPAÑOLA DE ENFERMEDADES RARAS

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Inscripciones

# I International Congress Gen IQSEC2 Mutation

**Date: February 22 and 23, 2025,  
Week of Rare Diseases Day**

**Location: Joaquin Ortuño Hall. Ramón y Cajal University Hospital. Madrid.**

## **Day 1 – February 22, 2026. Afternoon schedule**

Meeting of IQSEC2 Association families with researchers. Only for associated families and the scientific community.

## **Day 2 – February 23, 2026. I International Medical Congress IQSEC2 Gene Mutation**

### **PROGRAM – *PROVISIONAL***

#### **8:30 – 9:00. Welcome and accreditations**

#### **9:00 – 9:15. Opening**

- Dr. Miguel Ángel Moreno. Head of the Genetics service at Ramón y Cajal Hospital and IRYCIS researcher and Vice-president of the Spanish Association of Human Genetics.
- Ms. Empar Cebrian. President of the Spanish Association IQSEC2 Gene Mutation

#### **9:15 – 11:00. The IQSEC2 Genetic Mutation and current lines of research**

- 9:15: Dr. Andrew Levy. IQSEC2 Researcher. Technion. Israel Institute of Technology. Member of the Scientific Committee of the Spanish IQSEC2 Association
- 10:00: Dr. Alessandra Renieri \/\* Ilaria Meloni. Department of Medical Biotechnology. University of Siena. Member of the Scientific Committee of the Spanish IQSEC2 Association \On line\\*
- 10:45 Dr. Vassia Tegoulia. Senior Director of Pharmaceutical Technical Development at Genentech. Roche Group. Greece.

**Moderator:** Empar Cebrian. Spanish Association IQSEC2 Gene Mutation

#### **11:00 – 11:30. Q&A Session**

### **11:30 – 12:00. Coffee Networking**

### **12:00 – 13:30. Panel discussion. Spanish health entities. Challenges and future**

- 12:00. Dr. Raquel Buenhache. Neuropediatrician specializing in IQSEC2. Ramón y Cajal Hospital. Member of the Scientific Committee of the Spanish IQSEC2 Association
- 12:30: Dr. Eva Bermejo-Sánchez. Director and Scientific Researcher, Institute for Research on Rare Diseases. Carlos III Health Institute
- 12:50: Dra. Inmaculada Pitarch. Health Research Institute. La Fe Hospital. Valencia. Gene therapies of the Valencian Community. Member of the Scientific Committee of the Spanish IQSEC2 Association
- 13:10. Dra. Cristina de Frutos. Nutritionist. Ramón y Cajal Hospital. The ketogenic diet for the control of genetic epilepsy.

**Moderator:** Dr. Rosario Cazorla. Neuropediatrician. Puerta de Hierro Hospital  
(*confirmation pending*)

### **13:30 – 14:15. Families' round table. Our boys and girls with a rare disease**

- 13:30. Video presentation of the Spanish Association IQSEC2 Gene Mutation
- 13:35. Panel discussion with families. Living with the iqsec2 gene mutation. Present and future
- 13:55. Carolina Moreno, Irai's mother, "the girl who was born twice". Irai Association. Results of gene therapy treatment (*video*)
- 14:05. Rodrigo Bartolome. Vice-president of the CNNTB1 Syndrome Association. Showing that there is hope

**Moderator:** (...) Spanish Federation of Rare Diseases.

### **14:15 – 14:30. Building the IQSEC2 Support Network**

- 14:15. (...) The importance of associationism and support networks. Spanish Federation of Rare Diseases. FEDER

**Moderator:** Empar Cebrián. Spanish Association IQSEC2 Gene Mutation

### **14:30 – 14:45. Q&A Session**

### **14:45 – 15:00. Closing and conclusions**

**Organized by:** Spanish Association IQSEC2 Gene Mutation

**Collaborating Organizations:** Ramón y Cajal University Hospital  
FEDER, Spanish Federation of Rare Diseases.  
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