

A close-up photograph of a woman with dark hair hugging a young child with light brown hair. The woman is wearing a black top, and the child is wearing a plaid shirt. They are outdoors, with green foliage visible in the background.

""Before they  
cure me, I  
want them to  
take care of  
me""

(Albert Jovell)

# XI INTERNATIONAL CONGRESS ON RARE DISEASES



UNIVERSIDAD CATÓLICA  
DE MURCIA

• NOVEMBER 16 AND 17, 2018

• Catholic University of San  
• Antonio, UCAM - MURCIA

• Campus of the Jerónimos Nº 135, Guadalupe  
• 30107 (Murcia) - Spain



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# XI CONGRESO INTERNACIONAL DE ENFERMEDADES RARAS

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Su Majestad la Reina Letizia

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# XI INTERNATIONAL CONGRESS ON RARE DISEASES

## FRIDAY NOVEMBER 16

08.00 – 09.00

Accreditations

09.30 – 11.00

Opening session

Testimonials

Moderator: Miguel Ángel Ruíz Carabias. Director of D´Genes.

- Testimony 1: ANA PUGA GARCIA. The difficult path of re-entering the labor market

- Testimony 2: José Javier Martínez Aguilar. Access to equity treatments

- Testimony 3: Leire Gonzalez Diaz: Suddenly I have Ehler Danlos

Officers table

- Moderator: President UCAM

- Naca Eulalia Perez from Tudela. President of AELIP.

- Juan Carrión Tudela. President of D´Genes, FEDER and ALIBER.

- MSSSI Representative

- Diego Conesa Alcaraz. Government delegate of the Region of Murcia

- President UCAM. José Luis Mendoza.

-President CARM. Fernando López Miras.

11.00 – 11.30

Coffee Break

11.30 – 13.00

Good practices from the Administration in rare diseases

- Moderator: M<sup>a</sup> Teresa Martínez Ros. General Director of Planning, Pharmacy, Research and Healthcare of the Region of Murcia.

- Good practices in rare diseases in Madrid. Health Advisor Community of Madrid

- Good practices in rare diseases in Andalusia. Councilor of Health of Junta de Andalucía

- Good practices of the Murcia Regional Plan. Regional Minister of Health of the Region of Murcia

- European programs in rare diseases. Alba Ancochea. Director of FEDER and member of the board of directors of EURORDIS.



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# XI INTERNATIONAL CONGRESS ON RARE DISEASES

## FRIDAY NOVEMBER 16

13.00 – 14.30

**Invest in treatments a matter of the future**

- Moderator: Casimiro Jiménez. Subdirector General of Pharmacy of the Region of Murcia.
- Map of orphan drugs in Spain. David Beas. Vice President of Aelmhu.
- A good practice of starting a treatment: the example of Spinraza. Eduardo Martínez Salcedo. Neuropediatric University Hospital Virgen de la Arrixaca
- Home therapy: a practical experience. Javier Salguero. Clinical Business Unit Director Ashfield Healthcare
- Testimony. Documentary Family Mediterranean fever. Monica Tortosa. FMF President

14.30 – 16.00

**Lunch**

15.00 – 16.00

**Poster session**

16.00 – 17.30

**Main current research lines**

Moderator: Mari Ballesta. Optional specialist in Genetics University Hospital Virgen de la Arrixaca

- Systemic autoinflammatory amañifes. Advances in the diagnosis and treatment. Pablo Pelegrin. Scientific deputy director of IMIB Murcia.
- Animal models in the rare disease: Congenital dyskeratosis. "Dr. Maria Luisa Cayuela, Professor of Molecular Biology of the Degree in Medicine (UCAM) and Researcher of the IMIB-Arrixaca.
- Applications of animal models of Computational Chemistry in rare diseases. Dr. Horacio Pérez Sánchez .Structural Bioinformatics and High Performance Computing Research Group (BIO-HPC) (UCAM).
- Specialized administration system for the lysosomal release of enzymes used in the enzymatic replacement treatment of lysosomal storage diseases. M<sup>ª</sup> Luz Couce Pico. Diagnosis and Treatment of Metabolic Diseases Unit of the University Hospital Complex of Santiago
- New advances in research in gene therapy: the example of Wilson disease. Gloria Gonzalez-Aseguinolaza. TOP. Applied Medical Research Center of the University of Navarra

17.30 – 18.00

**Break**



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# XI INTERNATIONAL CONGRESS ON RARE DISEASES

## FRIDAY NOVEMBER 16

18.00 – 19.00

Conference

Moderator: Juan Carrión Tudela. President of Dgenes, FEDER and ALIBER

-Awareness and education, the path to transformation in research into rare diseases. Christian Gálvez

19:00-20:30

Advances in rare diseases

Moderator: Rosario Domingo. Optional specialist in Neuropediatrics. Virgen de la Arrixaca University Hospital.

- Neurodegenerative diseases with iron brain accumulation: clinical and genetic characterization through a Spanish multicentric research network. Carmen Espinós, Chief Investigator of the Unit of Genetics and Genomics of Neuromuscular and Neurodegenerative Diseases, Prince Felipe Research Center (CIPF), Valencia.

- Molecular bases of bone sclerosing disorders. Doctor Edgar Soriano Navarro. Deputy Medical Hospital Santa Lucía de Murcia and Tutor Tutor Practice of the Degree in Medicine UCAM.

- Research in ELA in Spain: collaborative projects, clinical trials and basic research. Eva Fages Caravaca, coordinator of UMDTELA, Unit of Diagnosis and Treatment of Amyotrophic Lateral Sclerosis of the Santa Lucia Hospital of Cartagena.

## SATURDAY NOVEMBER 17

9:30-11:00

Corporate social responsibility: humanization of care

Moderator: Isabel Fernández Aldeguer. D'Genes social worker.

- Social Responsibility as a therapeutic tool in Disability. The example of the Hospital de Molina. Pedro Hernández Managing Director Hospital de Molina de Segura

- Pain in children with communication difficulties: a proposal for humanization. M<sup>a</sup> Dolores Cárceles Barón. Chief of the child anesthesia section of the Hospital Virgen de la Arrixaca.

- Project HURGE: 12 months to dream. José Manuel Salas. Emergency physician.

- The ambulance the last wish. Manuel Pardo Rios. Vice-dean Podiatry UCAM

11:00-11:30

Coffee Break



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# XI INTERNATIONAL CONGRESS ON RARE DISEASES

## SATURDAY NOVEMBER 17

11:30-13:00

### Innovative elements in rare diseases

Moderator: Enrique Pastor. Vice President of the official social work college.

- Iris bond applied to children with Rett syndrome and to adults with ALS. Veronica Simón. Specialist in technology applied in augmentative and alternative communication and manager of the TACHIN TACHAN clinic
- Magisterial formulation in rare diseases. Rafael Puerto. Pharmacist and expert in Master Formulation.
- Presentation APP Combinations. Manuel Armayones. Director of Development of the eHealth Center.
- Support technology for communication and autonomy in people with multiple and complex needs. Soraya Muñoz Mendieta, Hearing and Language Teacher and Adviser in support technology of BJ Adaptaciones.

13:00-14:30

### Diagnostic updates

Moderator: Isabel López Expósito. Director of the Center for Biochemistry and Clinical Genetics of the Region of Murcia.

- New diagnostic techniques in obstructive pathology of pediatric central area. Dr. Juan Luis Antón-Pacheco. Pediatric surgeon and coordinator of the Airway Unit of the Hospital 12 de Octubre, Madrid
- Advances in acute intermittent porphyria. Maria Barreda Professor of the UCAM.
- Contributions of the genome to the diagnosis of rare diseases. Jorge Montoya, Physician of Genomic Systems.
- Raregenomics: an approach to the collaborative study of rare diseases. Carmen Ayuso García. Chief Medical Officer of the Genetics Service. Scientific Director of the IIS-FJD.

14:30-16:00

Lunch

15:00-16:00

Poster Session





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# XI INTERNATIONAL CONGRESS ON RARE DISEASES

## SATURDAY NOVEMBER 17

16:00-18:00

Practical workshops

1- Flamenco as therapy for Special Education. Dr. Victoria Cava. Cantora, teacher of Music Education, graduate in Journalism and doctor by the University of Murcia.

Moderator: Pascual Donate Ondoño. Director of D'Genes.

2- Feeding with pleasure: example in patients with ALS.

Inmaculada Jiménez. Member of the Nutrition Service of H.U. of Bellvitge.

Moderator: Encarna Bañon. Psychologist of D'Genes.

3- Medical ozone therapy: Interest in oxidative stress diseases. Javier Hidalgo. Bachelor of Medicine and Dentistry. Doctor in medicine

Moderator: María José Barba. Logopeda of D'Genes.

4- Practical aspects of the treatment of epilepsy. Helena Alarcón Martínez (Deputy Medical Doctor at the HCUVA Children's Neurology Service) and Inmaculada Martínez Albadalejo (Deputy Medical Neuropediatric Service HCUVA).

Moderator: Ángeles Carrión. Social worker of D'Genes

18:00-18:30

Break

18:30-19:00

Reading of conclusions

Moderator: Juan Carrión Tudela. President of Dgenes, FEDER and ALIBER

Videorecording Overcoming the difficulties of life. Roberto Cannessa.

19:00-19:30

Delivery of communications and poster awards

Moderator. Rosario Domingo. Optional specialist in Neuropediatrics. Virgen de la Arrixaca University Hospital.

19:30-20:00

Closing table

Moderator: Estrella Nuñez. Vice-Rector for Research UCAM

- Naca Eulalia Perez from Tudela. President of Aelip

- Juan Carrión Tudela. President of Dgenes, FEDER and ALIBER

- Estrella Núñez. Vice-Rector of Research of the UCAM

- Rosario Domingo: President of the Scientific Committee

## SUNDAY NOVEMBER 18

12:00

LUNCH AND VISIT GUIDED BY CENTER "PILAR BERNAL GIMÉNEZ"