7th international conference on ectodermal dysplasia (ED)

1st International course on ED
International

ED Families Meeting

April 12-14, 2018
Murcia, Spain

First European Year of Cultural Heritage 2018

PROVISIONAL PROGRAMME

Wednesday, 11 April 2017

19.30 Regional Government Oficial Reception
Palacio de San Esteban (Murcia)

Thursday, 12 April 2018

08:30 Scientific Conference Registration

09:00 Opening
Authorities
Organizing Committee President
Scientific Committee President

09:30 Presentation of the conference
José Manuel Montoya, EDIN

9:45 Opening lecture
Ectodermal dysplasias: an overview and international classification system review

John T. Wright, Director of Strategic Initiatives. School of Dentistry. University of North Carolina. USA
10.45 Session 1: Dermatological aspects in ED

**Update in Sweat gland dysfunction**
*Smail Hadj Rabia.* INSERM U781 et Département de Génétique, Hôpital Necker-Enfants Malades, Paris, France

**Dermatological Care in ED**
*Victoria Martínez, Teresa Martínez-Menchón, Paloma Sánchez-Pedreño,* Dermatology Service. Hospital Clínico Universitario Virgen de la Arrixaca. IMIB-Arrixaca. Universidad de Murcia. Spain

11:30 Coffee break and poster viewing

12:00 Honor Lecture

**Orodental manifestations in ED and principles for dental treatment**
*Birgitta Bergendal.* National Oral Disability Centre for Rare Disorders. The Institute for Postgraduate Dental Education, Jönköping, Sweden.

12. 45 Session 2: Dental and Eye Management in ED clinical cases

*Julio Anibal Cortés.* Oral and Maxillofacial Surgery Service. Hospital Clínico Universitario Virgen de la Arrixaca. Murcia, Spain

**Clinical experience in osseus craniomaxillofacial reconstruction in ED**

**Eye management in ED.**
*Mª Dolores Romero Caballero.* Ophthalmology Service. Hospital Reina Sofía. Universidad de Murcia, Spain

13:30 Lunch and poster viewing

15:00 Session 3: Original ED Clinical Cases

*Gianluca Tadini,* Dermatology Service. Ospedale Universitario L. Sacco Coordinador del Centro de enfermedades cutáneas hereditarias. Milan University. Italy

*Christine Bodemer,* Head of the Department of Dermatology, Coordinator of the French National Reference Centre for Genodermatoses, Coordinator of the French National Network for Rare Skin Diseases. Coordinator of the European Network for rare and complex skin disorders ERN-Skin. Necker Enfants Malades Hospital, University Sorbonne Paris, France
17.00 Murcia Cultural Heritage visit (Cathedral and Salzillo Museum)

Friday, 13 April 2018

09:00 Session 4: Genetics in ED

Mutational spectrum in ED Spanish patients: implication for genetic counselling
MC Martínez / E Guillén-Navarro. Genética. Hospital Clínico Universitario Virgen de la Arrixaca. IMIB-Arrixaca. CIBERER. Murcia, Spain

Genotype-phenotype correlation in hypohidrotic ectodermal dysplasia
Holm Schneider, Professor of Pediatrics, Head of the Center for Ectodermal Dysplasias & Head of the Division of Molecular Pediatrics, University Hospital Erlangen, Germany

10:00 Session 5: Diagnosis and reproduction in ED

Noninvasive Prenatal Diagnosis of Hypohidrotic Ectodermal Dysplasia by Tooth Germ Sonography.
Johanna Hammersen, German Competence Centre for Children with Ectodermal Dysplasias, Department of Pediatrics, University of Erlangen-Nuremberg, Erlangen, Germany

NIPD by DNA analysis in maternal blood

11:30 Coffee break and poster viewing

12:00 Session 6: Social and ethical aspects in ED

Genetic data protection and use
Magnolia Pardo López
Professor Constitutional Law. Universidad de Murcia
Director Spanish Unit UMU-UNESCO Chair in Bioethics and Health Law (University of Haifa, Israel)

Informed Consents in ED Clinical Trials
Angus Clarke.
Medical Genetics Cancer & Genetics, School of Medicine, Cardiff University, Wales UK

Rare diseases and Social Security benefit: protection needs and legal responses
Belén García Romero
Professor Labour Law and Social Security Universidad de Murcia
Executive Comittee Spanish Unit UMU-UNESCO Chair in Bioethics and Health Law (University of Haifa, Israel)

13:15 Lunch
14:30 Session 7 : Update in diagnosis and treatment research

**Automatic recognition of the XLHED phenotype from facial images**
Smail Hadj Rabia INSERM U781 et Département de Génétique, Hôpital Necker-Enfants Malades, Paris, France

**Pharmacological stimulation of EDAR signaling and prenatal therapy of XLHED – update on ER004**
Holm Schneider, Professor of Pediatrics, Head of the Center for Ectodermal Dysplasias & Head of the Division of Molecular Pediatrics, University Hospital Erlangen, Germany

15:45 ED testimony

16:00 Coffee break and poster viewing

16:30 Session 8: Patient Advocacy Groups and European reference network (*Sponsored by Europe Direct Region of Murcia*)

**FEDER**
- Juan Carrión
- President FEDER

**EURORDIS**
- Denis Costello
- Spanish Eurordis Office

**AADE**
- Gema Chicano
- President AADE

**EDIN**
- Jose Manuel Montoya
- EDIN

**ERN**
- Prof. Christine Bodemer
- INSERM U781 et Département de Génétique,
- Hôpital Necker-Enfants Malades, Paris, France

17:45 Oral communications selected from the abstracts

6 selected communications (8 minutes + 2 min questions)

18.45 Presentation of the 8th International Conference on Ectodermal Dysplasia

_Thomas Counioux (AFDE) & Smail Hadj Rabia_ (Hôpital Necker-Enfants Malades, Paris, France)

19.00 Closing: Remarks of the conference, future goals in ED and farewell.

21:00 Conference dinner (Casino Murcia)
Saturday, 14 April 2018

09:00 International meeting for de ED association leaders (EDIN)

09:30 ED Families meeting

Round Table: Summary of the 7th International Conference for families and future

Dermatology: Dr. MT Martinez Menchón
Ophtalmology: Dr. M. Dolores Romero
Maxillofacial: Dr. Julio Anibal
Genetics/Research: Dr. Encarna Guillén-Navarro
Social: Gema Chicano

Questions from patients and families (1 hour)

12.30 Terra natura visit and Lunch