ORGANIZER:
ÁREA TRANSVERSAL IBIMA-RARE

ORGANIZING COMMITTEE:

MIGUEL ANGEL MEDINA TORRES. Coordinador IBIMA-RARE. Dept. de Biología Molecular y Bioquímica. Universidad de Málaga.

YOLANDA DE DIEGO OTERO. Coordinadora adjunta IBIMA-RARE. UGC Salud Mental. Hosp. Regional Universitario de Málaga.

PEDRO VALDIVIELSO FELICES. IBIMA-RARE. UGC de Medicina Interna. Hospital Clínico Virgen de la Victoria. Departamento de Medicina y Dermatología. Universidad de Málaga.

ISABEL M. MUÑOZ CASTILLO. IBIMA-RARE. UGC Farmacia. Hospital Regional Universitario de Málaga.

JUAN ANTONIO GARCÍA RANEA. IBIMA-Rare. Unidad 741 CIBERER. Departamento de Biología Molecular y Bioquímica. Facultad de Ciencias. Universidad de Málaga.

RAQUEL YAHYAOUI MACÍAS. IBIMA-RARE. UGC Laboratorio. Unidad de Metabolopatías. Hospital Regional Universitario de Málaga.

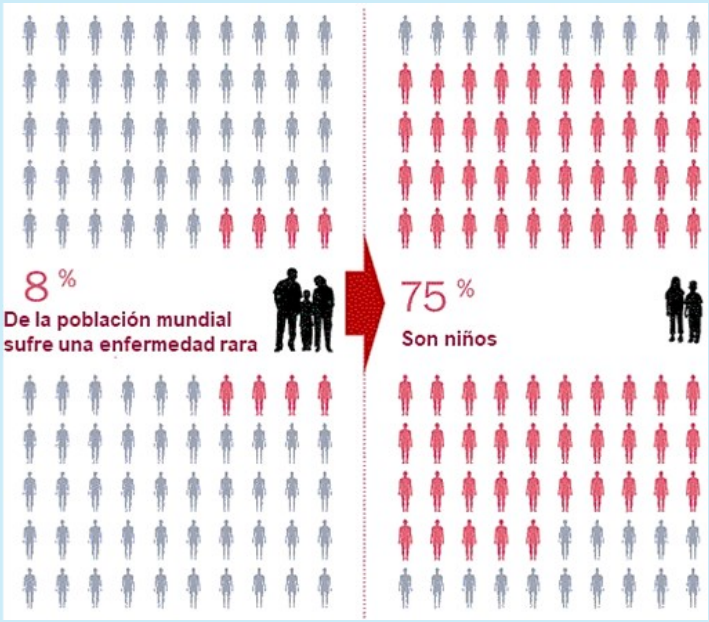
ROCÍO CALVO MEDINA. IBIMA-RARE. UGC Pediatría. Hospital Regional Universitario de Málaga.

ROCIO ASENSI DÍEZ. IBIMA-RARE. UGC Farmacia. Hospital Regional Universitario de Málaga.

ROSA BERMÚDEZ DE ALVEAR. IBIMA-RARE. Departamento Radiología y Medicina Física. Oftalmología y Otorrinolaringología. Universidad de Málaga.

GINÉS MARTÍNEZ ARQUERO. IBIMA-RARE. UGC Otorrinolaringología. Hospital Regional Universitario de Málaga.

FRANCISCA SÁNCHEZ JIMÉNEZ. IBIMA-RARE.



Venue (AULA MAGNA. Facultad de Medicina):
Bulevar Louis Pasteur, 32, 29010 Málaga

https://goo.gl/maps/AgQKFRGbig72EfNt7

PG8G+2H Málaga

ONLINE FREE INSCRIPTION :

https://forms.gle/qhpiiZo9Ez7MA2nR7

ORGANIZER:
ÁREA TRANSVERSAL IBIMA-RARE
Instituto de Investigación Biomédica de Málaga (IBIMA)

ibima
Instituto de Investigación Biomédica de Málaga

1st IBIMA-Rare Winter Course 2019
(1st IRWI-Course)

ADVANCES IN MEDICAL GENOMICS
Diagnosis, prevention, novel orphan treatments and challenges to achieve precision medicine in Rare Disorders

AVANCES EN MEDICINA GENOMICA
Diagnóstico, prevención, nuevos tratamiento huérfanos y retos para alcanzar medicina de precisión en Enfermedades Raras

19-20, December 2019

AULA MAGNA

FACULTAD DE MEDICINA

UNIVERSIDAD DE MÁLAGA

PROGRAM

Thursday, December 19th 2019.

9:00-9:15h Welcoming remarks.

9:15-9:45h Stem cell as novel treatments on Rare disorders. Experimental approach for hereditary congenital hydrocephalus. **Prof. Dr. Antonio J. Jiménez Lara.** Dept of Cellular Biology, genetics and Physiology. University of Malaga.

9:45-10:15h Pseudoxanthoma elasticum (PXE): Molecular diagnosis and experimental treatment for a rare disorder. **Prof. Dra. María García Fernández.** Department of Physiology. University of Malaga.

10:15-10:45h Experimental CRISPR/Cas9 based technology for human genome edition: a potential novel treatment for Rare Genetic Disorders. **Dr. Enrique Viguera Mínguez.** Department of Cellular Biology, Genetics and Physiology. University of Malaga.

10:45-11:00h *Questions and colloquium.*

11:00-11:30 *Cofee break.*

11:30-12:00 Familial counseling and possible preventive options of hereditary rare disorders. **Dr. José Ignacio Lao Villadóniga.** Medical Director at Genomic Genetics International and Clinical Genetics and Genetic Counselling Unit at Clínica Diagonal. Barcelona.

12:00-12:30h Molecular diagnosis and novel treatments for inherited metabolic disorders**. Dr. Belén Pérez González.** Universidad Autónoma de Madrid.

12:30-13-00h Clinic and basic research at the IBIMA-Rare to develop orphan drugs for rare disorders: New treatment for the Fragile X syndrome. **Dr. Yolanda de Diego Otero.** IBIMA-Rare (IBIMA C03-group). Mental Health Clinical Unit. Regional University Hospital of Malaga.

13:30h-13:50h *Questions and colloquium.*

13:50- 14:00h *Quality Questionnaire and assistance controls.*

14:00h-15:30h Free time for *Lunch.*

15:30-16:00h Preventing rare genetic disorders: prenatal and preimplantational diagnosis. **Dr. Antonio Cejudo Román.** Research Coordinator at IVI Málaga.

16:00-16:30h Newborn screening of inherited disorders: a model of precision medicine. **Dr. Raquel Yahyaoui Macias.** IBIMA-Rare (IBIMA). Laboratory of Metabolic Disorders. UGC Laboratory. Regional University Hospital of Malaga.

16:30-17:00h Relevance of BIG DATA analysis to develop precision medicine of rare disorders. **Dr. Michaela Spiteri.** Former Data Science and Predictive Analysis Lecturer, Malta College of Arts, Science and Technology. University of Malta.

Thursday, December 19th 2019.

17:00-17:30h Future on treatments of neurodegenerative rare diseases. Orphan drug for the treatment of the Spinal Muscular Atrophy (SMA). **Dr. Rocío Calvo Medina.** IBIMA-Rare (IBIMA C03-group). Neuropediatric Unit. Regional University Hospital of Malaga.

17:30-17:50h *Questions and colloquium.*

17:50-18:15h *Cofee Break.*

18:15-18:55h Counselling on hereditary neurodevelopmental rare disorders. **Dr. Pietro Chiurazzi.** Institute of Genomic Medicine, Catholic University, Rome, Italy.

18:55-19:35h Strategies for Molecular diagnosis of hereditary disorders. **Dr. Aida Bertoli Avella.** Research Director. Centogene. Rostock. Germany.

19:35- 19:50h *Questions and colloquium.*

19:50 -20:00h *Quality Questionnaire.*

Friday, December 20th, 2019.

9:00-09:30h Bioinformatics and systems biology approaches for the study of rare disease disorders. **Dr. James Perkins.** IBIMA-Rare (IBIMA C03-group). University of Malaga.

09:30-10:00h Research on neurodevelopmental rare disorders. **Dr. Pietro Chiurazzi.** Institute of Genomic Medicine, Catholic University, Rome, Italy.

10:00-10:30h Research and new strategies for molecular diagnosis of hereditary rare disorders. **Dr. Aida Bertoli Avella.** Research Director. Centogene. Rostock. Germany.

10:30-11:00h Precision medicine to understand rare disorders leading to epilepsy. **Dr. Pedro Serrano Castro.** UGC Neurology. Regional University Hospital of Malaga.

11:00-11:15h *Questions and colloquium*

11:20-11:40 *Cofee break*

11:40-12:10h Deciphering epigenetic mechanisms and regulatory circuits in pulmonary arterial hypertension. **Dr. Armando Reyes Palomares**. Dpt. Biochemistry and Molecular Biology. Complutense University of Madrid.

12:10-12:40h Mendelian neurodegenerative diseases: spotlight on neurodegeneration with brain iron accumulation (NBIA). **Dr. Carmen Espinós Armero.** Group Leader, Unit of Genetics and Genomics of Neuromuscular and Neurodegenerative Disorders, Research Center Principe Felipe (CIPF), Valencia, Spain.

12:40-13.10h From bench to bed: basic research to develop diagnoses and treatments for patients with rare disorders. **Prof. Dra. Francisca Sánchez Jiménez**. IBIMA-Rare.

13:10h-13:30h *Questions and colloquium.*

13:30- 13:45h *Quality Questionnaire and assistance controls.*

13:45h-14:00h Final remarks and departure.

Rare Disorders

A disease is considered a rare disorder when it affects few people, less than 1 in 2000. Despite the low frequency of each rare disease, all as a whole affect 8% of the general population, which means about 600000 affected people in Andalusia and 150000 affected people in the province of Malaga with more than 50000 in the capital. It mainly affects children, since 75% of rare pathologies are initiated in childhood. When parents and close relatives are taken into acount, who may be directly or indirectly affected by these inherited genetic diseases, it may affect about 25% of the population of any country. Rare diseases are a global problem requiring global solutions, that need the involvement of related areas such as health, education and social systems.

ORGANIZER: IBIMA-Rare. IBIMA

ibima
Instituto de Investigación Biomédica de Málaga

ONLINE FREE INSCRIPCION :

https://forms.gle/qhpiiZo9Ez7MA2nR7