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ÁREA TRANSVERSAL IBIMA-RARE

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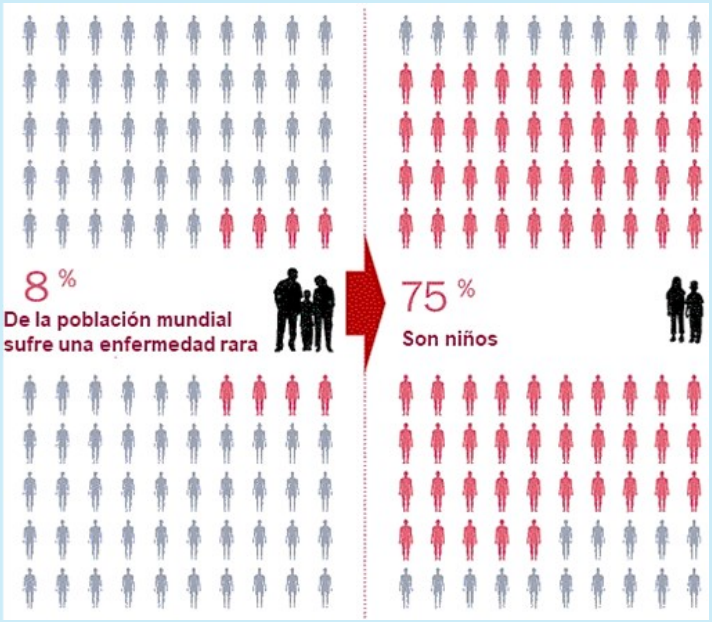

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Instituto de Investigación Biomédica de Málaga (IBIMA)

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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

SALÓN DE GRADOS

FACULTAD DE CIENCIAS

UNIVERSIDAD DE MÁLAGA

PROGRAMME

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 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 rare 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 in Andalusia and 150000 affected in the province of Malaga with more than 50000 in the capital. It mainly affects children. 75% of rare pathologies are initiated in childhood. When parents and close relatives who may be directly or indirectly affected, as they are inherited genetic diseases, these diseases can affect about 25% of the population of any country. Rare diseases are a global problem, requiring the involvement of related areas such as health, education, work and social systems.

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