Symposium on Genome Research: Rare Diseases

DATE: October 29th, 2015 **SITE : Aula Magna** (Room)– Fundación Jiménez Díaz **TIME:** 8:45 h – 18:00 h

Most of the rare diseases are genetic and typically caused by mutations in genome and exome .

The overall objective of the symposium is to show the latest developments occurred in the molecular characterization of rare diseases using new genomic tools.

TIME	TITLE	SPEAKER
8:45-9:00	Registration	
9:00-9:30	Opening and presentation.	Dr. Jesús Fernandez Crespo Instituto de Salud Carlos III (ISCIII), Madrid Dr. Ivo Gut Centro Nacional de Análisis Genómico (CNAG-CRG) ,Barcelona Dra. Carmen Ayuso Instituto de Investigación Sanitaria (IIS-FJD) ,Madrid
9:30-10:10	Gene discovery in Glaucoma.	Dr. Julio Escribano Universidad de Castilla La Mancha (UCLM), Albacete
10:10-10:50	Sensorial disorders (BBS-like): Approach from WES	Dra. Diana Valverde Universidad de Vigo, Vigo
10:50-11:20	COFFEE BREAK	DESCANSO
11:20:12:00	Non-syndromic aortic disease and NGS	Dra. Marina Gago Instituto de Investigación Sanitaria, Santiago de Compostela
12:00-12:40	Motor neuropathies; new genes and mechanisms	Dra. Carmen Espinos Centro de Investigación en Red de Enfermedades Raras (CIBERER) ,Valencia
12:40-13:20	Several rare disorders	Dr. Milan Macek Charles University. Praga
13:20-14:30	LUNCH	DESCANSO
14:30-15:10	*Bloque III: Moderadora*	Dra. Montserrat Baiget Hospital de la Santa Creu i Sant Pau, Barcelona.
15:10-15:50	Complement Dysfunction and Disease	Dra. Margarita Lopez Trascasa Hospital Universitario La Paz, Madrid
15:50-16:30	Hereditary breast cáncer families with a recessive pattern of inheritance	Dra. Ana Osorio Cabrero Centro Nacional de Investigaciones Oncológicas, Madrid
16:30-17:00	Integration of resources and data sharing for rare disease research and diagnostics	Dr. Sergi Beltrán Centro Nacional de Análisis Genómico (CNAG-CRG), Barcelona
17:00-17:40	BREAK	DESCANSO
	Closure	