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 Dr. Ivo Gut Dr. Carmen Ayuso
9:30-10:10	Gene discovery in Glaucoma.	Dr. Julio Escribano
10:10-10:50	Sensorial disorders (BBS-like): Approach from WES	Dr. Diana Valverde
10:50-11:20	COFFEE BREAK	COFFEE BREAK
11:20:12:00	Brugada syndrome and NGS	Dr. Marina Gago
12:00-12:40	Motor neuropathies; new genes and mechanisms	Dr. Carmen Espinos
12:40-13:20	Several rare disorders	Dr. Milan Macek
13:20-14:30	LUNCH	LUNCH
14:30-15:10	Complement Dysfunction and Disease	Dr. Margarita Lopez Trascasa
15:10-15:50	Hereditary breast cáncer families with a recessive pattern of inheritance	Dr. Ana Osorio Cabrero
15:50-16:30	Integration of resources and data sharing for rare disease research and diagnostics	Dr. Sergi Beltrán
16:30-17:00	BREAK	BREAK
17:00-17:40	Closure	