



Hospital Universitario La Paz  
Hospital de Cantoblanco  
Hospital Carlos III

IdiPAZ  
Instituto de Investigación  
Hospital Universitario La Paz

ciberer

ibm

## International Workshop on Bone Dysplasias

Aula Jaso Hospital Infantil  
Hospital Universitario La Paz  
4 de mayo de 2017

ENTRADA LIBRE PREVIA INSCRIPCIÓN en [genmed.hulp@salud.madrid.org](mailto:genmed.hulp@salud.madrid.org)

### PROGRAMA:

- 10:00-10:15** Dr. Pablo Lapunzina, Inauguración y Bienvenida  
Hospital La Paz-INGEMM-CIBERER
- 10:15-10:45** Dr. Karen Heath, "Genetic analysis of Skeletal Dysplasias using Next Generation Sequencing (2012-2017)"  
Hospital La Paz-INGEMM-CIBERER
- 10:45-11:15** Prof. Mona Aglan, "Consanguinity and Skeletal Dysplasias"  
National Research Centre, Cairo, Egypt
- 11:15-11:45** Dr. Víctor L Ruiz-Pérez, "Phenotypes and Genotypes in Ellis-van Creveld syndrome"  
Instituto de Investigaciones Biomédicas- INGEMM-CIBERER
- 11:45-12:15** Pausa-Café (Cerramiento-Hospital General)
- 12:15-13:00** Dr. Ghada Otaify, "Metabolic Bone Disorders: Diagnosis and Management"  
National Research Centre, Cairo, Egypt
- 13:00-13:30** Dr. Fernando Santos, "COL2A1 associated Phenotypes"  
Hospital La Paz-INGEMM-CIBERER
- 13:30-13:45** Prof. Hulya Kayserli "From tetra-amelia to longitudinal limb reductions: the newly defined RSPO2 clinical spectrum"  
Koç University School of Medicine, İstanbul, Turkey
- 13:45-14:00** Dres. Víctor L Ruiz Pérez y Pablo Lapunzina, Cierre del Workshop