ASHBi SEMINAR

Rare begets Common

Lecturer: Prof Bruno Reversade

IMB & IMCB, A*STAR, Singapore
Amsterdam UMC, Netherlands
Koç University, Turkey

Date **Friday, 3 September**

Time **15:00 – 16:00**

Venue **Zoom Online Meeting***

***Register via the right QR code**

Abstract

Over the last 10 years, a revolution in the speed and accessibility of high volume sequencing has introduced a paradigm shift in human genetics and medicine. Rare diseases – the human experiment – are a primary tool for assigning function to the human genome.

By identifying the genes responsible for rare diseases, we are best positioned to pinpoint the subverted biological pathways which not only underlie the pathology of a specific condition but which will, in many cases, reveal biological nodes whose perturbation contributes to more common pathologies – i.e. rare begets common.

I will illustrate this paradigm by highlighting how we have gone from discovering mutations in genes controlling early organogenesis in humans to the development of therapeutic drugs for oncology indications.

Organizer : Prof Takashi Hiiragi

[E-mail] hiiragi@embl.de

Hosted by Institute for the Advanced Study of Human Biology (WPI-ASHBi)