



# CDB SEMINAR

## Bruno Reversade

IMB & IMCB, A\*STAR, Singapore,  
AMC/VUmc, Netherlands,  
Koç University, Turkey

Thursday, April 20, 2017

13:30~15:00 Auditorium C1F

### R(e)spondin-2 WNT signaling for Limb & Lung Development

#### Summary

It is believed that the four R-SPONDIN secreted ligands primarily act via their cognate LGR4/5/6 receptors to enhance WNT signaling. Here I will document an allelic series of recessive R-SPONDIN2 (RSPO2) mutations causing a spectrum of congenital abnormalities in humans. Six fetuses with RSPO2-null alleles presented with Tetra-Amelia Syndrome characterized by lung aplasia, cleft lip/palate and a total absence of the four limbs.

Functional work revealed impaired LGR5-binding and reduced WNT potentiation which correlated with allele severity. Surprisingly however a triple ubiquitous knockout of Lgr4, Lgr5 and Lgr6 in mice suggested a very different interpretation.

#### Biography

Bruno was trained as a developmental biologist in the HHMI lab of Prof. De Robertis at UCLA. After his PhD in 2008, he was awarded the inaugural A\*STAR investigatorship and set up his team at the Institute of Medical Biology in Singapore. There, he switched to human genetics, placing emphasis on monogenic, fully penetrant and unique genetic traits as a means to understand complex and common diseases.

Combining the power of deep sequencing, patient-derived iPSCs and animal modeling in zebrafish, Xenopus and mice, his team has resolved numerous human disorders affecting embryogenesis, metabolism, ageing, cognition and familial cancers.

Bruno is a Professor and Research Director at A\*STAR in Singapore, an adjunct Professor of Paediatrics at NUS, a fellow of the Branco Weiss Foundation in Switzerland, the first EMBO Young Investigator based outside Europe and a distinguished Professor of Genetics at AMC/VUmc in Amsterdam, Netherlands and Koç University in Istanbul, Turkey.

#### Host:

**Hiroshi Hamada**

Organismal  
Patterning, CDB  
[hiroshi.hamada@riken.jp](mailto:hiroshi.hamada@riken.jp)  
Tel:078-306-3002  
(ext:3325)

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