



Yale SCHOOL OF MEDICINE

GENETICS DEPARTMENT SEMINAR SERIES

Special Seminar

R(e)SPONDIN' to WNT Signaling for Organogenesis

It is believed that the RSPONDIN secreted ligands primarily act via their cognate LGR4/5/6 receptors to enhance WNT signaling. Here I will reveal an allelic series of *RSPO2* mutations in humans causing a spectrum of birth defects ranging from digit anomalies to Tetra-Amelia Syndrome characterized by lung aplasia and a total absence of the four limbs.

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



Dr. Bruno Reversade, PhD

Professor & Research Director
Institute of Medical Biology, A*STAR

Host: Dr. Antonio Giraldez, PhD

Chair and Professor of Genetics
YSM Department of Genetics

Tuesday, April 16, 2019

11:30am - 12:30pm

The Anlyan Center – N107

The Genetics Calendar of Events can be viewed on-line at
<https://medicine.yale.edu/genetics/events/seminars.aspx>