CRBM external seminar
BIOLuM
March 20th, 2025 11:00 Salle Marcel Dorée

Identifying therapeutical options in Cornelia de Lange syndrome

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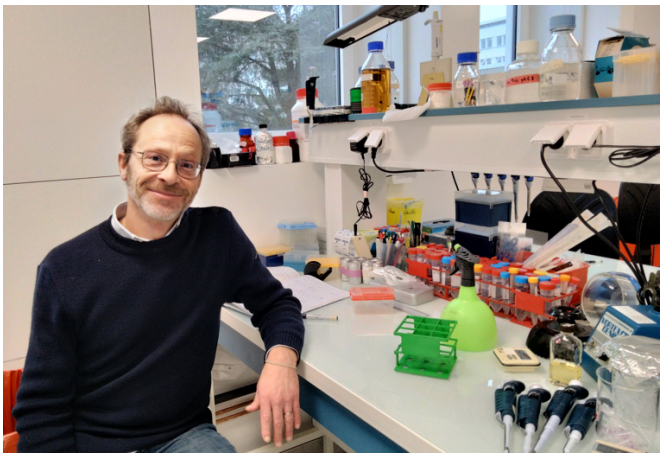

Erwan WATRIN completed his Ph.D. with Vincent LEGAGNEUX at the IGDR in Rennes in 2003. Then he moved to Vienna for a Postdoctoral training in IMP with Jan-Michael PETERS (2004-2008). He was recruited CRCN CNRS in 2008 in the Cell Cycle team of Claude PRIGENT at the IGDR, Rennes.
Erwan WATRIN is an expert in chromosome cohesion and cohesinopathies.
He has been working for many years on Cornelia De Lange syndrome one of the cohesinopathies.

Abstract

Cornelia de Lange syndrome, CdLS, is a rare neurodevelopmental disorder arising mainly from causative mutations in genes encoding members of the sister chromatid cohesion apparatus, i.e., cohesin and friends. CdLS patients present with multiple affected organs and functions- including heart and brain- short stature, typical craniofacial features as well as mild-to-severe cognitive and developmental delays.
Over the last decade, we and our collaborators have investigated CdLS at molecular and cellular levels, aiming at establishing genotype-phenotype correlation, improving diagnosis and prognosis.
Here, I will present some of this work, with a particular emphasis on unpublished results that have identified dysregulated cellular pathways and corresponding pharmacological compound relevant for therapy.

Selected publications

Vitoria Gomes et al. The cohesin ATPase cycle is mediated by specific conformational dynamics and interface plasticity of SMC1A and SMC3 ATPase domains. Cell Rep. 2024

Guénantin et al. Targeting the histone demethylase LSD1 prevents cardiomyopathy in a mouse model of laminopathy. J Clin Invest. 2021

Parenti et al. MAU2 and NIPBL Variants Impair the Heterodimerization of the Cohesin Loader Subunits and Cause Cornelia de Lange Syndrome. Cell Rep. 2020

Parenti et al. Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. Hum Genet. 2017