2017 IGBMC Summer Internship

Molecular basis of transcriptional dysregulations in the cone-rod dystrophy of SpinoCerebellar Ataxia 7

Laboratory of Dr Yvon Trottier

SpinoCerebellar Ataxia 7 (SCA7) is a fatal neurodegenerative disease associated with extensive down-regulation of genes controlling neuronal function and identity, particularly affecting photoreceptors, one major targeted neurons of the disease. SCA7 is due to a polyglutamine expansion in Ataxin-7 (ATXN7), which is a core component of SAGA (Spt-Ada-Gcn5 Acetyltransferase), a multiprotein complex that regulates transcription through histone H3 acetylation and H2B deubiquitination activities. Whether dysfunction of SAGA and subsequent epigenetic alterations underlie gene dysregulation in SCA7 remains unclear.

The student will participate to the characterization of epigenetic marks underlying the photoreceptor degeneration using functional genomic approaches (RNA-seq and ChIP-QPCR). This will improve our understanding of the molecular basis of neuron-specific gene repression in SCA7 mouse tissues. Our long term objective is to search for common and unique molecular signatures characteristic for SCA7 pathophysiology in neuronal cell types.
The student will learn about functional genomic approaches and the physiology and pathophysiology of the retina