Sébastien Jacquemont is a medical geneticist and associate professor at the of St. Justine University Hospital. He holds a Canadian research chair, a JJL Levesque chair in genetics of neuropsychiatric disorders as well as a Swiss national foundation assistant professorship. SJ was trained as a clinical geneticist and subsequently completed a research fellowship in developmental pediatrics at the MIND Institute, UC Davis.

His group is interested in mapping the effects of rare variants genome-wide onto cognitive and behavioral dimensions. They have shown that genomic dosage due to CNVs could modulate brain structure, cognition and behavior. This work was initially performed on specific variants including 16p11.2 CNVs by pooling data from European and North American consortiums. These investigations are now extended to genome-wide rare recurrent and non-recurrent CNVs and SNVs using new strategies, which are neither univariate nor burden analyses. Instead, we are identifying genetic scores and functional annotations that best explain the variance of cognitive and behavioral traits. This approach represents a new framework to investigate genomic variants too rare to be studied in individual association studies.

His group is also actively working on drug development programs and clinical trials in fragile X syndrome (a “monogenic/syndromic” form of autism and intellectual disability).