Cadherin dysfunctions hack brain wiring leading to neurodevelopmental disorders

Protocadherins are calcium-dependent cell-adhesion molecules, whose dysfunctions affect brain connectivity and information processing leading to several neuropsychiatric disorders. In particular, the protocadherins PCDH9, PCDH10 and PCDH19 have been implicated in autism spectrum disorder (ASD), epilepsy and cognitive impairment. Recently, we have reported that PCDH19, which is implicated in Early Infantile Epileptic Encephalopathy-9 (EIEE9), interacts with GABA(A) receptors and regulates the inhibitory transmission and neuronal excitability (Bassani et al. 2018; Serratto et al. 2020). Furthermore, we generated a Pcdh19 conditional KO mouse that reproduces key features of EIEE9, and we are currently characterizing it from a molecular, functional, and behavioral point of view.

In addition, to deepen the characterization of the Pcdh19 KO mouse model, we are now expanding our studies to other protocadherins, especially PCDH9 and PCDH10, in order to understand the pathological mechanisms underlying neurodevelopmental disorders.

References:


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