


Título del Proyecto	Identifying new mechanisms by which DYRK1A regulates the expansion of the cerebral cortex
Nº de expediente asignado	JEROME LEJEUNE FOUNDATION - Grant 2017
Abstract	<p>Intellectual disability (ID) syndromes are neurodevelopmental disorders characterized by deficits in cognitive and adaptive functioning that occurs before 18 years of age. ID affects approximately 1% of the population and it represents an important public health issue due to the requirement of extensive clinical and social support services (1, 2). There is no cure for ID, thus the identification of its physiopathology is crucial to design better intervention strategies. In this project we will focus in a single gene, DYRK1A, that has been linked to two ID syndromes:</p> <p>Down syndrome (DS; OMIM #190685) and a recently defined syndrome named “DYRK1A-related intellectual disability syndrome” (OMIM #614104). DYRK1A is a pleiotropic protein kinase that regulates important developmental processes in the central nervous system (CNS). Indeed, previous studies (in which the applicant has participated) have demonstrated that DYRK1A regulates brain growth in a dosage-dependent manner and that small variations in DYRK1A protein levels are sufficient to modify the neurogenic potential of embryonic cortical progenitors (3, 4). In this project we will continue studying the effect of DYRK1A protein on the behaviour of these progenitors with the ultimate goal of understanding the cellular mechanisms by which pathogenic dosages of DYRK1A alter the expansion of the cerebral cortex, the brain region responsible for higher cognitive functions.</p> <p>To this end, we will use available Dyrk1a mutant mice that model ID syndromes and have an imbalanced dosage of Dyrk1a gene.</p>
Entidad Financiadora	JEROME LEJEUNE FOUNDATION

Convocatoria:	JEROME LEJEUNE FOUNDATION - Grant 2017
Importe de la ayuda	40.000€
Fechas de ejecución del proyecto	julio 2017-julio 2019
	 The logo for the Fondation Jérôme Lejeune features a red square on the left containing a white profile of a man's face. To the right of the square, the text "FONDATION" is in small grey letters, "Jérôme Lejeune" is in large red letters, and "chercher, soigner, défendre" is in smaller grey letters below it. A red horizontal line is positioned under the text.
Enlaces:	https://www.ciberisciii.es/areas-tematicas/grupo-de-investigacion?id=17098