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The exon junction complex in neural development and neurodevelopmental disease

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Abstract

Post-transcriptional mRNA metabolism has emerged as a critical regulatory nexus in proper development and function of the nervous system. In particular, recent studies highlight roles for the exon junction complex (EJC) in neurodevelopment. The EJC is an RNA binding complex composed of 3 core proteins, EIF4A3 (DDX48), RBM8A (Y14), and MAGOH, and is a major hub of post-transcriptional regulation. Following deposition onto mRNA, the EJC serves as a platform for the binding of peripheral factors which together regulate splicing, nonsense mediated decay, translation, and RNA localization. While fundamental molecular roles of the EJC have been well established, the *in vivo* relevance, particularly in mammals, has only recently been examined. New genetic models and cellular assays have revealed core and peripheral EJC components play critical roles in brain development, stem cell function, neuronal outgrowth, and neuronal activity. Moreover, human genetics studies increasingly implicate EJC components in the etiology of neurodevelopmental disorders. Collectively, these findings indicate that proper dosage of EJC components is necessary for diverse aspects of neuronal development and function. Going forward, genetic models of EJC components will provide