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Degree of Cajal-Retzius Cell Mislocalization Correlates with the Severity of Structural Brain Defects in Mouse Models of Dystroglycanopathy.

Booler HS¹, Williams JL¹, Hopkinson M¹, Brown SC¹.

Author information

Abstract

The secondary dystroglycanopathies are characterized by the hypoglycosylation of alpha dystroglycan, and are associated with mutations in at least 18 genes that act on the glycosylation of this cell surface receptor rather than the Dag1 gene itself. At the severe end of the disease spectrum, there are substantial structural brain defects, the most striking of which is often cobblestone lissencephaly. The aim of this study was to determine the gene-specific aspects of the dystroglycanopathy brain phenotype through a detailed investigation of the structural brain defects present at birth in three mouse models of dystroglycanopathy-the FKRP(KD), which has an 80% reduction in Fkrp transcript levels; the Pomgnt1null, which carries a deletion of exons 7-16 of the Pomgnt1 gene; and the Large(myd) mouse, which carries a deletion of exons 5-7 of the Large gene. We show a rostrocaudal and mediolateral gradient in the severity of brain lesions in FKRP(KD), and to a lesser extent Pomgnt1null mice. Furthermore, the mislocalization of Cajal-Retzius cells is correlated with the gradient of these lesions and the severity of the brain phenotype in these models. Overall these observations implicate gene-specific differences in the pathogenesis of brain lesions in this group of disorders.

KEYWORDS: Cajal-Retzius cells; congenital muscular dystrophy; dystroglycan; neuronal migration; pial basement membrane; reelin

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