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Role of genetic background in behavioral and cognitive phenotypes of NF1 mice

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Abstract

Neurofibromatosis type 1 (NF1) is a genetically determined neurodevelopmental disorder and tumor syndrome with an incidence of approximately 1/3000 live births. It is caused by loss of function mutations in the neurofibromin gene (NF1) and is estimated to effect 100,000 people in the US. Approximately 50% of the patients inherit the condition in an autosomal dominant pattern; the remaining 50% are the result of de novo mutations. Cognitive deficits have been found in 70% of children with NF1. They include specific problems with attention, visual perception, language, learning, attention, and executive function. These symptoms are observed in the absence of tumors or macroscopic structural abnormalities in the central nervous system. No effective treatments for the behavioral and cognitive disabilities of NF1 exist. It is currently unclear how the parental gene carrier may affect the offspring's cognitive abilities, therefore we tested whether the parental carrier of the mutated NF1 gene and/or genetic background strain affected behavioral and cognitive performance in mice. The experimental mice are on a mixed C57BL6/J x 129/SvJ background where either the mother or the father was a heterozygous NF1 carrier. Preliminary results indicate that the parental carrier does significantly influence the behavioral and cognitive phenotype of the offspring. A progress report on this study will be presented during the Research Week.

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