Docket #: S98-194

Mouse Model for Progressive Myoclonus Epilepsy (EPM1)

Myers, et al previously discovered that specific loss-of-function mutations in the human cystatin B gene on chromosome 21 cause the human genetic disease Progressive Myoclonus Epilepsy (EPM1). They have now created a mouse model for EPM1--the first mouse model of a human epilepsy.

This mouse model for the EPM1 may prove effective in development of therapies for EPM1 and other Progressive Myoclonic Epilepsies. Additionally, it may serve as a model to uncover the mechanisms of action of dilantin and of cysteine proteases and their inhibitors, and clarify the role of cystatin B in apoptosis and corneal defects.

Applications

- To develop better therapies for treating EPM1
- To develop therapies that may be relevant in treating other Progressive Myoclonic Epilepsies
- To understand the mechanism of action of dilantin (phenytoin), which in EPM1 patients, causes worse symptoms than no treatment at all. Dilantin is one of the most commonly prescribed antiepileptic drugs, but its mechanism of action is unknown and its affects vary from patient to patient.
- To study apoptosis in vivo . These knockout mice have widespread granule cell death in the cerebellum indicating cystatin B has an essential role in preventing apoptosis.
- To study corneal defects. Mice lacking cystatin B develop ocular opacities as a result of corneal lesions.
- To study cysteine proteases and their inhibitors

Advantages

- This is the first mouse model of human epilepsy and it may contribute greatly to the development of future epilepsy mouse models and in discovery of therapeutic targets for epilepsy

Publications

- Pennacchio, L. A., Bouley, D. M., Higgins, K. M., Scott, M. P., Noebels, J. L., and Myers, R. M. (1998). Progressive ataxia, myoclonic epilepsy, and cerebellar apoptosis in cystatin B-deficient mice. Nature Genet. 20: 251-258.

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