CNTNAP2 Knock Out Mouse Model for Human Diseases

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Overview

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Cntnap2 (contactin-associated protein-like 2) knockout mice lack detectable mRNA or protein expression in brain tissue and serve as a robust model for Cortical Dysplasia-Focal Epilepsy Syndrome and autism spectrum disorders (ASD).

Homozygous animals exhibit hyperactivity, social and communication deficits, spontaneous and handling-induced seizures, and abnormal cortical neuron organization with reduced parvalbumin-positive interneurons and asynchronous neuronal firing. Despite normal nerve conductance, they show enhanced responses to sensory stimuli, impaired nest building, increased grooming and digging, and reduced ultrasonic vocalizations. Risperidone treatment alleviates some behavioral abnormalities. Cntnap2, a member of the neurexin superfamily, is essential for potassium channel localization at the juxtaparanodal region, and its disruption models key neurodevelopmental and epileptic features observed in humans with CNTNAP2 mutations.

References

Poliak S , et al. 2003, J Cell Biol