TRM: Two Mutant Mice Strains for the Study of Miller–Dieker syndrome (MDS)

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BACKGROUND

Miller–Dieker syndrome (MDS), or 17p13.3 deletion syndrome results in human neuronal migration disorders characterized by type 1 lissencephaly sequence (ILS), severe mental retardation and reduced life expectancy. The understanding of these syndromes is often incomplete and is the subject of active research. Researchers have demonstrated that the gene encoding 14-3-3ε (*YWHAE*), one of a family of ubiquitous phosphoserine/threonine–binding proteins, is always deleted in individuals with MDS. Mice deficient in *Ywhae* have defects in brain development and neuronal migration, similar to defects observed in mice heterozygous with respect to *Pafah1b1*.

Gene specific transcriptional activation or repression is regulated by a complex network of transcription factors designated the Myc/Max/Mad network. MNT (max binding protein) binds DNA and a heterodimer with MAX and represses transcription and acts as an antagonist of Myc-dependent transcriptional activation and cell growth.

Described below are two mice strains that may be useful in studies of Miller-Dieker Lissencephaly Syndrome generated by the same researcher.

TECHNOLOGY DESCRIPTION

Mice carrying a targeted mutation of *Ywhae* (tyrosine 3-monooxygenase/tryptophan 5-monooxygenase activation protein, epsilon polypeptide) exhibit hippocampal defects, neuronal migration abnormalities, and increased apoptosis in the brain (Cat. No. 008715).

The Mnt^{cko} (*Mnt*, max binding protein) mutant mice may also be useful in generating conditional mutations for studies of Miller-Dieker syndrome, embryonic development and craniofacial defects. (Cat. No. 009084)

APPLICATIONS

Both mutant mouse strains may be useful in studies of Miller-Dieker Lissencephaly Syndrome.

STATE OF DEVELOPMENT

The mice are designated Tangible Research Material (TRM). A complete description, including genotyping, phenotyping, etc is found at The Jackson Lab cat. No. 008715, https://www.jax.org/strain/008715

Information available for the Mnt^{cko} mice see Cat. No. 009084, https://www.jax.org/strain/009084.

INTELLECTUAL PROPERTY INFO

Academic and non-profit institutions please order directly from The Jackson Laboratory. Commercial entities require a license from UC San Diego contact (https://innovation.ucsd.edu/contact/).

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RELATED MATERIALS

- Toyo-oka K, Shionoya A, Gambello MJ, Cardoso C, Leventer R, Ward HL, Ayala R, Tsai LH, Dobyns W, Ledbetter D, Hirotsune S, Wynshaw-Boris A. 14-3-3epsilon is important for neuronal migration by binding to NUDEL: a molecular explanation for Miller-Dieker syndrome. Nat Genet. 2003 Jul;34(3):274-85. - 06/08/2003

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OTHER INFORMATION

KEYWORDS

Miller–Dieker syndrome, YWHAE, 14-3-3e, k/o mice, neurological abnormalities, microdeletions, lissencephaly

CATEGORIZED AS

- **Medical**
  - Disease: Genetic Diseases and Dysmorphic Syndromes
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