Docket #: S19-315

16p11.2 deletion mice - Jackson Labs stock no. 025330

Our researcher has developed a mouse model of 16p11.2 deletion syndrome. A copy number variation on human chromosome 16p11.2 is among the most common genetic variations found in autism spectrum disorders. 16p11flx mice are a Cre or FLP recombinase-inducible mouse model of 16p11.2 deletion that has several loxP and frt sites flanking the corresponding 440 kbp region on mouse chromosome 7F3. These mice may be useful in studying basal ganglia circuitry and the pathophysiology of autism.. Several papers using these mice have been published. The mice were also deposited at JAX and are available for use by academic and industrial groups, stock no. 025330

Applications

- Mouse models are an important tool for identifying the underlying pathological mechanisms of neurodevelopmental and psychiatric diseases. They are also important for testing potential new therapies and drugs preclinically. 16p11.2 deletion syndrome is a neurodevelopmental disorder characterized by mild intelectual disability, ADHD, obesity, autism and other symptoms. The deletion is one of the most common genetic causes of neurodevelopmental disease. The mouse is a cellular and molecular phenocopy of the human disease. It has and several anatomical and behavioral phenotypes that can be used to develop preclinical assays for developing therapies for autism and other diseases. Neurons and other cells from these mice can also be used to develop cellular assays that are useful for drug target validation, for screening and drug optimization assays.

Publications

- Portmann T; Yang M; Mao R; Panagiotakos G; Ellegood J; Dolen G; Bader PL; Grueter BA; Goold C; Fisher E; Clifford K; Rengarajan P; Kalikhman D; Loureiro D; Saw NL; Zhengqui Z; Miller MA; Lerch JP; Henkelman RM; Shamloo M; Malenka RC; Crawley JN; Dolmetsch RE. 2014. Behavioral abnormalities and circuit defects in the Basal Ganglia of a mouse model of 16p11.2 deletion syndrome. Cell Rep 7(4):1077-92PubMed: 24794428MGI: J:210018

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