CONDITIONAL Fmr1 Knockout Mouse Model For Fragile X Syndrome

ERASMUS MC has developed a conditional Fmr1 KO model that allows the creation of null allele in specific cell types and at specific time points.

THE CHALLENGE

Fragile X syndrome (FXS) is the most frequent form of inherited intellectual disability with a prevalence of 1 in 4000 males and 1 in 6000 females. The main primary cause of this disease is an impediment in the transcription of FMR1 gene, leading to a loss of FMR1 gene function. To further understand the role of Fmrp during development and in FXS, true null Fmr1 knockout animal models that allow to limit the expression of Fmrp protein, both spatially and temporally, are needed.

THE TECHNOLOGY

ERASMUS MC has developed a conditional Fmr1 knock-out (Fmr1 CKO) mouse model by flanking the promoter and first exon of Fmr1 with bacteriophage P1-derived loxP sites. This enables the creation of a null allele in specific cell types and at specific time points by crossing Fmr1 CKO mice with tissue specific or inducible cre-recombinase expressing mice.

STAGE OF DEVELOPMENT

The Fmr1 CKO mouse model is ready to use and its utility has been demonstrated by developing a Purkinje cell-specific null for Fmr1 mice. This mice were successfully generated by crossing Fmr1 CKO with a Purkinje cell-specific (L7/PCP2) cre-recombinase expresser, which resulted in mice lacking Fmrp expression only in Purkinje neurons (Figure 1).

ADVANTAGES & APPLICATIONS

- Research tool for neurological studies
- Research tool for determining the role of Fmrp in brain and testis development
- The Fmr1 CKO mouse model may ultimately allow determining the role of Fmrp in FXS and the underlying molecular mechanisms of this disorder.

COMMERCIAL OPPORTUNITY

ERASMUS MC offers this technology for out-licensing or on a contract research basis. Interested parties are kindly invited to contact ERASMUS MC Technology Transfer Office (please see contact details below) to further explore the possibilities of partnership.

REFERENCES


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