

Animal Model of Brittle Bone Disease

Published date: Aug. 22, 2017

Technology description

Osteogenesis imperfecta (OI) or brittle bone disease is an incurable genetic disorder that causes fragile bones and a lifetime of breaks and fractures. Recently, mutations in the gene for bone morphogenetic protein 1 (BMP1) were reported in two recessive OI families.

BMP1 and a closely related proteinase called mammalian tolloid-like 1 (TLL1) are coexpressed in various tissues including bone, and have overlapping activities related to collagen development. Studying their roles of these genes in bones and other tissues could lead to greater understanding of OI and possible therapeutic interventions.

Until now, early lethality of BMP1- and TLL1-null mice has precluded the use of such models. A UW–Madison researcher and collaborator have developed a mouse strain that is conditionally null for BMP1 and mTLL1. The model exhibits the hallmark features of OI and provides a new means to screen drugs and study mechanism.

The mice exhibit dramatically weakened and brittle bone with spontaneous fractures – defining features of OI. Additional skeletal features include osteomalacia, thinned/porous cortical bone, reduced processing of procollagen and dentin matrix protein 1, remarkably high bone turnover and defective osteocyte maturation.

WARF also owns and can license one of the parental lines (a mouse strain with a floxed TLL1 allele). The Wisconsin Alumni Research Foundation (WARF) is seeking commercial partners interested in BMP1 and TLL1 double-knockout mice for *in vivo* analysis and screening for potential treatments of osteogenesis imperfecta.

Additional Information

Muir A.M., Ren Y., Butz D.H., Blank R.D., Birk D.E., Lee S.J., Rowe D., Feng J.Q. and Greenspan D.S. 2014. Induced Ablation of BMP1 and TLL1 Produces Osteogenesis Imperfecta in Mice. *Hum Mol Genet.* 23, 3085-3101.

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Application area

Animal model for mechanistic studies of OI and drug screening

Advantages

First mouse model of its kind

Provides exciting new opportunity to study and potentially treat OI

Biomaterial is available for licensing.

Institution

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