B6-*Dmd* Del51

Strain Name： C57BL/6JGpt-Dmd^{em2Cd1406}/Gpt
Strain Type： Knock-out
Strain Number： T056004
Background： C57BL/6JGpt

Description

Duchenne muscular dystrophy (DMD) belongs to the X-linked recessive genetic disease. The disease is characterized by progressive atrophy of proximal skeletal muscles of the extremities, and pseudo hypertrophy of the calf gastrocnemius [1], which also involves the myocarid and respiratory muscles, leading to early death [2]. This disease also affects the development of central nervous systems and organs. The DMD gene encodes a large rod-shaped cytoskeleton protein (Dystrophin), the protein is mainly distributed in the inner surface of the bone and myocardial muscle fibers, which helps muscle fibers maintain their integrity and elasticity during contraction, it is also a vital component of the muscular dystrophin complex and acts as a very important bridge to maintain the cytoskeleton[3].

The main cause of DMD disease is the truncation of the Dystrophin caused by DMD gene mutation or deletion, which lead to the functional loss of Dystrophin. There are many kinds of DMD gene mutation and deletion, mainly occurring in the exon 45-55 region. At present, there is no effective cure for the disease, several gene therapise are in progress, among them, and exon skipping therapy is one of the most popular treatment[4]. Based on the high-frequency deletion region of DMD gene in patients, GemPharmatech constructed a B6-*Dmd* Del51 mouse model of by deleting exon 51 of the mouse *Dmd* gene. B6-*Dmd* Del51 mice did not express DMD protein in the heart and muscle. B6-*Dmd* Del51 mice can be used to exon skipping therapy screening and optimization and the study of the pathogenesis of DMD disease.

Strategy

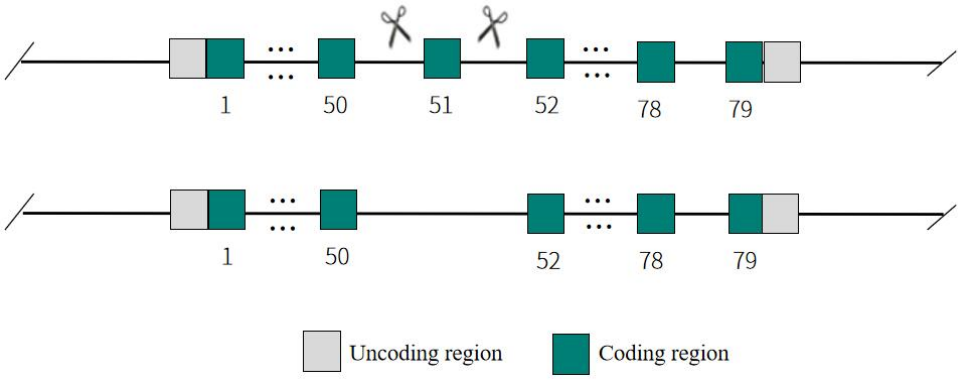

Fig 1. Schematic diagram of B6-*Dmd* Del51 model stratery

Application

1. Screening of drugs for muscular dystrophy
2. Pathophysiological study on muscular dystrophy

Data support

1.Expression of Dystrophin

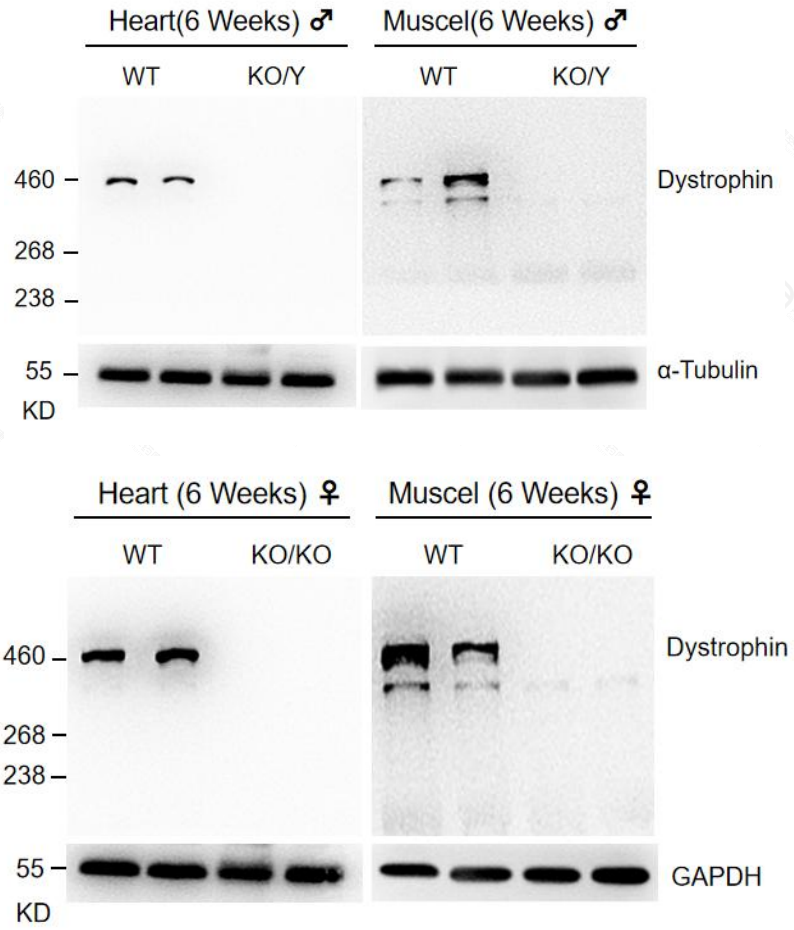

Figure 2. The expression of Dystrophin protein in B6-*Dmd* Del51 mice is deficient

The expression of DMD protein in heart and muscle of 6-week-old WT mice and B6-*Dmd* Del51 mice were detected by western blot analysis. As shown in Figure 2, B6-*Dmd* Del51 mice did not express DMD protein in the heart and muscle.

Reference：

1. Sacco, Alessandra, et al. "Short telomeres and stem cell exhaustion model Duchenne muscular dystrophy in mdx/mTR mice." Cell 143.7 (2010): 1059-1071.
2. Mourkioti, Foteini, et al. "Role of telomere dysfunction in cardiac failure in Duchenne muscular dystrophy." Nature cell biology 15.8 (2013): 895-904.
3. Elangkovan, Nertiyan, and George Dickson. "Gene therapy for Duchenne muscular dystrophy." Journal of Neuromuscular Diseases 8.s2 (2021): S303-S316.
4. Sheikh, Omar, and Toshifumi Yokota. "Developing DMD therapeutics: a review of the effectiveness of small molecules, stop-codon readthrough, dystrophin gene replacement, and exon-skipping therapies." Expert opinion on investigational drugs 30.2 (2021): 167-176.