SBMA Transgenic Mouse Model for Spinal and Bulbar Muscular Atrophy Research

Biomaterial – Mouse

Biomaterial Description

This genetically engineered mouse model expresses a mutant human androgen receptor (AR) containing an expanded polyglutamine (polyQ) tract, specifically in skeletal muscle tissue. The transgene is floxed, enabling cell-type-specific excision of the human AR121Q allele through introduction of Cre recombinase. Designed to model Spinal and Bulbar Muscular Atrophy (SBMA)—a neuromuscular disorder caused by CAG repeat expansion in the AR gene—this model reproduces hallmark features of the disease, including motor neuron degeneration and systemic metabolic dysfunction. It demonstrates that muscle-restricted expression of mutant AR is sufficient to drive both neurological and systemic disease phenotypes.

Applications

-Investigating muscle-to-neuron signaling in SBMA and related disorders

-Evaluating skeletal muscle-specific therapeutic strategies

-Studying systemic metabolic and endocrine dysfunction linked to muscle pathology

-Modeling non-cell-autonomous neurodegeneration with spatial and temporal control

Advantages

-Conditional expression via LoxP-Cre system enables tissue-specific control

-Muscle-specific AR expression isolates peripheral contributions to SBMA

-Recapitulates both neuromuscular and systemic disease phenotypes

-Compatible with a wide range of Cre driver lines for flexible experimental design

-Supports translational research into muscle-directed therapies

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Category

Research Tools/Biological Materials/Mouse

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References

1. Cortes, C. J., Ling, S. C., Guo, L. T., Hung, G., Tsunemi, T., Ly, L., Tokunaga, S., Lopez, E., Sopher, B. L., Bennett, C. F., Shelton, G. D., Cleveland, D. W., La Spada, A. R.(2014) , https://pmc.ncbi.nlm.nih.gov/articles/PMC4096235/, https://www.sciencedirect.com/journal/neuron, 82, 295-307