These mice harbor a Dhcr24-knockout allele that disrupts catalysis of the last step of cholesterol biosynthesis (the conversion of desmosterol to cholesterol), and may be useful in studying cholesterol biosynthesis, epidermal development, keratinocyte differentiation, and epidermal barrier function, as well as Alzheimer's disease and tumor biology. In addition, the Dhcr24 gene (also called selective Alzheimer's disease indicator-1 (Seladin-1)) is reported to be a liver X receptor (LXR) target gene.

Donating Investigator
Shailendra B Patel, Medical College of Wisconsin
Details

Detailed Description

Homozygous mice on the C57BL/6 genetic background die within the first postnatal day with features of lethal restrictive dermopathy; including taut, wrinkle-free, shiny skin, severe defects in epidermal maturation/epidermal barrier function (impaired epidermal development), and increased presence of hyperproliferative immature keratinocytes (defective keratinocyte differentiation). The increased transepidermal water loss/increased epidermal water content is associated with increased aquaporin-3 (AQP3) expression throughout the epidermis. Mice heterozygous for this allele are viable and fertile, with no overt phenotype. As the Dhcr24 protein functions to catalyze the last step of cholesterol biosynthesis (the conversion of desmosterol to cholesterol), homozygous disruption of this locus results in desmosterolosis: almost no cholesterol in plasma and tissues with ~99% of total sterols in the form of desmosterol. LacZ expression from the mutant allele is not characterized.

In an attempt to offer alleles on well-characterized or multiple genetic backgrounds, alleles are frequently moved to a genetic background different from that on which an allele was first characterized. It should be noted that the phenotype of Dhcr24-mutant mice was originally described on a mixed C57BL/6;129S5 genetic background. We will modify the strain description if necessary as published results become available.

Development

Expression Data

Control Suggestions

Selected References

Genetics

Dhcr24tm1Lex
Genotyping Protocols
Separated PCR: Dhcr24
Genotyping resources and troubleshooting

Breeding Considerations

When maintaining a live colony, heterozygous mice may be bred together, to wildtype siblings or to C57BL/6J inbred mice (Stock No. 000664). Homozygous mice on the C57BL/6J genetic background die immediately after birth.

Additional Breeding and Husbandry Support

Citation

When using the B6.129S5-Dhcr24<sup>tm1Lex</sup>/SbpaJ mouse strain in a publication, please cite the originating article(s) and include JAX stock #012564 in your Materials and Methods section.

Animal Health Reports
Facility Barrier Level Descriptions

Production of mice from cryopreserved embryos or sperm occurs in a maximum barrier room, G200

Pricing & Availability

Typically mice are recovered in 10-14 weeks. Contact Customer Service to place an order or for more information.
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