



Stria vascularis profile in model of Warrensburg syndrome type 2a

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DESCRIPTION

Warrensburg Syndrome is an abnormal hereditary disorder characterized by at least some congenital hearing loss and pigmentation disorders. However, the genetic pathway affecting the event of stria vascularis isn't fully illustrated. There were 113 genes in tyrosine metabolism, melanin formation, and ion transport, with significant changes in the porcine model and 191 genes in the mouse model. In addition, there have been some species specific gene changes within the stria vascularis within the mouse and porcine models. Expression of tight junction-related genes, including the *Cadm1*, *Cldn11*, *Pcdh1*, *Pcdh19*, and *Cdh24* genes, was significantly higher in the porcine model compared to the mouse model. Stria vascularis-related and ion channel-related genes also showed significant differences between the two species. The expression of the *Col2a1*, *Col3a1*, *Col11a1* and *Col11a2* genes was higher in the porcine model compared to the mouse model, and the expression of the *Col8a2*, *Cd34* and *Ncam* genes was lower (Brokekamp,1986).

Warrensburg Syndrome (WS) is a rare hereditary disorder characterized by congenital deafness and pigmentation changes, at least to some extent. Although mouse models are widely used in the study of disease phenotypes and pathogenic mechanisms of deafness, studies of human genetic diseases have also identified many shortcomings. This can make significant biological differences in anatomy, energy metabolism, and hearing, as there are significant revolutionary differences between mice and humans. For example, the developmental patterns of the auditory organs of mice and humans are different. Human hearing developed prenatally, but mouse hearing did not fully develop until two weeks after birth. Some studies have found it difficult to reproduce human embryonic disease in some mouse models. Therefore, various animal models such as cows, horses, dogs, and pigs were also needed to study genetic diseases. Pigs are a social species with a fully developed auditory system at birth.

Recent studies have also found that the anatomy of the cochlea is very similar to that of humans. Pigs are large animals that are highly reproductive and economically convenient, making them an excellent model for studying genetic disorders of hearing (Fox,1989).

Therefore, various animal models such as cows, horses, dogs, and pigs were also needed to study genetic diseases. Pigs are a social species with a fully developed auditory system at birth. Recent studies have also found that the anatomy of the cochlea is very similar to that of humans. Pigs are large animals that are highly reproductive and economically convenient, making them an excellent model for studying genetic disorders of hearing. Different genes may be involved in the maintenance of EP in mice and pigs, and mutations in the *Mitf* gene are expected to cause different changes in potassium channels. To answer these questions, this study sought to detect changes in the gene profiles of these two species caused by mutations in the *Mitf* gene at the RNA transcriptase level. Since most studies today use only mouse models, this paper further demonstrates the differences in RNA transcription enzymes in the stria vascularis of large animals and mouse models (Njunge,1991).

Stria Vascularis Specific Ion Transport-Related Gene

Ion transport-related genes were extracted from RNA transcriptase data from normal and *mitf-m* mutant pig and mouse samples for cluster analysis. The results showed that many ion transport-related genes were highly expressed in both species by MeV cluster analysis. *Mitf* mutations were affected by the genes *Trpm1*, *Kcnj13*, and *Slc45a2* from both species. There was a significant difference in ion channel regulation between pigs and mice. Expression of the *Kcnn1*, *Cicn2*, and *Trpm4* genes was higher in pigs than in mice, whereas expression of the *Trpm7*, *Kcnq1*, and *Kcnj8* genes was higher in mice than in pigs (Schoeb,1990).

Specific Tight Junction-Related Genes in the Stria Vascularis

Tight junction-related genes were extracted from *mitf* mutants and normal porcine / mouse RNA transcriptase data for cluster analysis. The expression of tight junctions in the two stria vascularis was different. The genes *Cadm1*, *Cldn11*, *Pcdh1*, *Pcdh19*, and *Cdh24* are more highly expressed in pigs than the mouse genes, while the genes *Ncam*, *Cldn6*, *Cldn9*, and *Cldn14* are more highly expressed in mice compared to pigs was done. They found that both structures of the stria vascularis were intact in the two groups. The surrounding nuclei and cell junctions were intact. The three layers of cells were obvious and the basal cells were tightly connected (Vander Gaag, 1984).

Stria Vascularis Specific Vascular Development-Related Genes

Genes associated with vascular development extracted from *mitf* mutants and normal porcine / mouse RNA transcriptase data were used for cluster analysis. There was a significant difference in the vascular development genes of the stria vascularis between these two species. The *Col2a1*, *Col3a1*, *Col11a1*, and *Col11a2* genes were more highly expressed in pigs than in mice, while the *Col8a2*, *Cd34*, and *Ncam* genes were more highly expressed in mice compared to pigs.

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