KIT gene mutation causes deafness and hypopigmentation in Bama miniature pigs

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Abstract

Background: Waardenburg syndrome (WS), a common type of syndromic hearing loss. A large group of patients affected by WS were found no mutations in the above gene panel, indicating that there are still potential genes responsible for WS yet to be detected. Methods: In our previous study, we established a KITD806E/+ pig pedigree with an autosomal dominant inheritance model. This model presented congenital bilateral severe sensorineural hearing loss with hypopigmentation, exact the same as human WS. Results: Histological analysis of the KITD806E/+ cochlea showed nearly normal structures of the organ of Corti, stria vascularis and spiral neuron ganglions at E85. Scanning electron microscopy (SEM) exhibited the auditory hair cells began to degenerate at E100, and totally gone at P1. Transmission electron microscope (TEM) showed disorganization of stria vascularis (SV) and intermediate cells in the middle layer of SV had gone. The absence of endocochlear potentials also demonstrated the dysfunction of stria. Conclusions: KITD806E/+ mutation interrupted the development of melanocytes in cochlea, which led to the malformation and dysfunction of SV, resulting in degeneration of hair cells and finally hearing loss. Therefore, KIT was highly supposed to be a newly found gene associated with WS.

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