MedicalXpress

Animal study finds link between MAP2 mutation and hereditary hair diseases

November 21 2019

The genetic mechanism of hereditary human hair diseases, such as alopecia and thinning hair, has drawn much attention in human genetics research, yet many questions around this mechanism persist. A recent animal study in *The FASEB Journal* revealed that a mutation in the gene that encodes a protein called MAP2 (for "microtubule-associated protein 2") may be an essential component of the hairless phenotype.

Researchers from China Agricultural University (CAU) selected two groups (i.e., an adult group and a newborn group) of normal and hairless pigs. Pigs were chosen as an appropriate model for the study due to their high homology with humans. Researchers first examined differences in hair follicles and skin between the normal and hairless pigs. They found that the hair follicle densities of the normal pigs were much higher than those of the hairless pigs.

Next, the researchers used sequencing and molecular experiments to examine genetic differences between the normal and hairless pigs. Their findings revealed that protein coding gene MAP2 was strongly associated with the hairless trait. Further, they found that a missense mutation (i.e., the substitution of one amino acid for another in a DNA base pair) in the MAP2 gene led to decreased hair follicle density in the embryonic stage, which resulted in hair follicles not forming normally.

"To the best of our knowledge, we believe this is the first time MAP2 has been associated with hair follicle morphogenesis," said Xiangdong Ding, Ph.D., a researcher in CAU's Department of Animal Breeding and

Genetics in Beijing. "Further exploration of this mechanism could result in a potential cure for hereditary hair diseases through gene editing."

Outside of the main finding in this study, the study also demonstrated that hairless pigs can serve as an ideal animal model for human hair diseases.

"How fascinating it is when an unanticipated gene is implicated in a particular trait—this one encoding a cytoskeleton protein defined in the 1970's," said Thoru Pederson, Ph.D., Editor-in-Chief of *The FASEB Journal*. "Discoveries such as the present one resonate with our appetite for surprise."

Provided by Federation of American Societies for Experimental Biology