Scientists discover human sperm gene is 600 million years old

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Just as styles in sexy clothes or fashion change from year to year and culture to culture, "sexy" genes, or genes specific to sex, also change rapidly. But there is one sex-specific gene so vital, its function has remained unaltered throughout evolution and is found in almost all animals, according to new research from Northwestern University Feinberg School of Medicine.

The gene, called Boule, is responsible for sperm production. Northwestern scientists also discovered in their research that Boule appears to be the only gene known to be exclusively required for sperm production from an insect to a mammal.

"This is the first clear evidence that suggests our ability to produce sperm is very ancient, probably originating at the dawn of animal evolution 600 million years ago," said Eugene Xu, assistant professor of obstetrics and gynecology at Feinberg. "This finding suggests that all animal sperm production likely comes from a common prototype."

Xu is senior author of a paper on the study that will be published July 15 in PLoS Genetics.

The discovery of Boule's key role in perpetuating animal species offers a better understanding of male infertility, a potential target for a male contraceptive drug and a new direction for future development of pesticides or medicine against infectious parasites or carriers of germs.

"Our findings also show that humans, despite how complex we are, across the evolutionary lines all the way to flies, which are very simple, still have one fundamental element that's shared," Xu said.

"It's really surprising because sperm production gets pounded by natural selection," he said. "It tends to change due to strong selective pressures for sperm-specific genes to evolve. There is extra pressure to be a super male to improve reproductive success. This is the one sex-specific element that didn't change across species. This must be so important that it can't change."

Boule is likely the oldest human sperm-specific gene ever discovered, Xu said. He originally discovered the human gene in 2001.

Prior to the new findings, it was not known whether sperm produced by various animal species came from the same prototype. Birds and insects both fly, for example, but the fly wing and bird wing originated completely independently.

For the study, Xu searched for and discovered the presence of the Boule gene in sperm across different evolutionary lines: human, mammal, fish, insect, worm and marine invertebrate.

In order to search for Boule's presence across the spectrum of evolutionary development, Xu had an interesting shopping list. He needed sperm from a sea urchin, a rooster, a fruit fly, a human and a fish. The fish proved to be the most difficult.

Xu purchased a rainbow trout at a Chicago fish market, unwrapped it and was dismayed to discover it had been gutted. "I need the testicles!" he exclaimed to the seafood salesman. Xu decided he'd have to catch his own. He cast a fishing line into a recreational pond stocked with trout and reeled in a rainbow trout.

Discovery of this common gene involved in sperm production could have many practical uses for human health, including male contraception. When Xu's research group knocked out the Boule gene from a mouse, the animal appeared to be healthy but did not produce sperm.

"A sperm-specific gene like Boule is an ideal target for a male contraceptive drug," Xu noted.

Boule also has the potential to reduce diseases caused by mosquitoes and parasites such as worms.

"We now have one strong candidate to target for controlling their breeding," Xu said. "Our work suggests that disrupting the function of Boule in animals most likely will disrupt their breeding and put the threatening parasites or germs under control. This could represent a new direction in our future development of pesticides or medicine against infectious parasites or carriers of germs."

To further support his hypothesis that Boule is widespread across all animals producing sperm and eggs, Xu also examined the genome of one of the most primitive animals, a sea anemone, for the presence of Boule. He looked at its genome because the sperm of the sea anemone is difficult to find and few labs study the animal. When Xu identified Boule in the sea anemone genome, his theory was clinched.

Provided by Northwestern University