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Scientists find new genes on male sex chromosomes

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Scientists are a step closer to discovering what determines the sex of Australia's iconic platypus and echidna, after an international study involving researchers from the University of Adelaide and UNSW Australia unravelled new genes contained on mammalian Y chromosomes.

The findings of the study, which was led by the University of Lausanne in Switzerland, were today published in the prestigious journal *Nature*.

The Y chromosome is only found in males and plays a key role in determining male sex. Despite its importance for sex determination, gene content and evolution of the Y chromosome has long been a mystery in most mammals, particularly in monotremes (the platypus and echidna).

Dr Frank Grützner, genetics lecturer and ARC research fellow with the University of Adelaide's School of Molecular and Biomedical Science, says the study saw researchers analyse billions of genetic sequences from 15 mammal species and revealed, for the first time, new gene repertoires for all major mammal groups, tracing the evolution of the Y chromosome in unprecedented detail.

"Unravelling genes on Y chromosomes has always been a challenging task, and little was known about Y chromosome genes in most mammal species," he says.

"Finding these new genes is a major breakthrough for us. It finally reveals the gene content of the two different Y chromosome systems that evolved in mammals," says Dr Grutzner, who has led research into monotreme sex chromosomes for more than 10 years.

Dr Paul Waters, an ARC fellow in the School of Biotechnology and Biomolecular Sciences at UNSW Australia, says that the X and Y chromosomes started their existence as normal autosomes (non-sex chromosomes) harbouring the same genes. "As the Y chromosome evolved, it withered away, losing most of the 1000 genes that are found on today's X chromosomes. The preserved genes were then recruited into male-specific functions," he says.

University of Adelaide School of Molecular and Biomedical Science PhD student, Deborah Toledo-Flores, says: "the most important aspect of this work for us was to identify more genes on platypus Y chromosomes to reveal new leads about potential sex determining genes in these animals."

"The next step will be to discover which gene on the Y chromosomes determines sex in the platypus and echidna.

"Y chromosomes are vital for male development and fertility and it will be fascinating to gain a better insight into the function that these newly discovered genes have in different species," she says.

More information: Origins and functional evolution of Y chromosomes across mammals, *Nature*, dx.doi.org/10.1038/nature13151

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