



Australian animals crack genetic riddle

Our marsupials help scientists uncover the mysterious origins of two debilitating human diseases. **TAMARA McLEAN** reports . . .

THE genes of two iconic Australian animals, the kangaroo and the platypus, have been used to uncover the mysterious origins of two debilitating human diseases.

Research by Australian National University scientists is the first to prove that two rare genetic conditions evolved in humans much more recently than first thought.

The team studied the genes that are abnormal in Prader-Willi Syndrome, a complex disorder which causes learning difficulties and obesity, and Angelman Syndrome, which is marked by severe mental retardation and inappropriate laughter.

Both diseases are caused by an error in imprinted genes when only the genes from the mother or the father — not both — work but the relevant gene for each disease is either missing or mutated.

People with PWS have a problematic copy from their father and, for Angelman sufferers,

their mother.

Eminent ANU geneticist, Professor Jenny Graves, led a study by student researchers Rob Rapkins and Tim Hore to investigate the origins of the conditions by studying the genomes of two very different creatures, the kangaroo and the platypus.

Examined together, the two animals offer important clues because they both have very different reproduction systems: kangaroos bear very tiny, live young without foetal development, and the platypus lays eggs.

The researchers were able to find the AS gene in both creatures but, says Prof. Graves, it was not what they expected.

The big surprise was that the AS gene turned out to be next to completely different genes from those nearby in the human genome, she said.

And the problematic gene responsible for the other condition, PWS, couldn't be found in either animal.

This discovery, detailed in the scientific journal *PloS Genetics*, shows that gene imprinting is a relatively recent development in evolutionary terms.

"Imprinting was thought to have evolved when mammals abandoned egg-laying 210 million years ago," Prof. Graves said.

But the absence of the imprinted genes in kangaroos indicates that imprinting of these genes developed in placental mammals much more recently.

She said imprinting seems to have been thrown together from bits and pieces taken from all around the (human) genome.

The research sheds new light on the accidental way that imprinting can come into being and suggests, says Prof. Graves, that evolution of the phenomenon is an ongoing process in all mammals that bear live young, including humans. — AAP



CASE STUDY: Two of our iconic animals, the kangaroo and platypus, offer important clues about gene imprinting