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Australian animals solve riddle of genetic disease

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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 (ANU) 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 (PWS), a complex disorder which causes learning difficulties and obesity, and Angelman Syndrome (AS), 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 humans.