Mammalian cells with single chromosome set created

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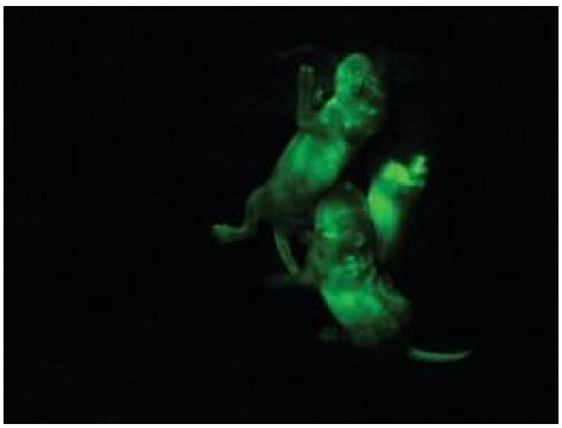

Scientists at the University of Cambridge bred mice with fluorescent green cells derived from haploid (single chromosome set) embryonic stem cells. Credit: Anton Wutz and Martin Leeb, University of Cambridge/Nature

Researchers have created mammalian cells containing a single set of chromosomes for the first time in research funded by the Wellcome Trust and EMBO. The technique should allow scientists to better establish the relationships between genes and their function.

Mammal cells usually contain two sets of chromosomes – one set inherited from the mother, one from the father. The genetic information contained in these chromosome sets helps determine how our bodies develop. Changes in this genetic code can lead to or increase the risk of developing disease.

To understand how our genes function, scientists manipulate the genes in animal models – such as the fruit fly, zebrafish and mice – and observe the effects of these changes. However, as each cell contains two copies of each chromosome, determining the link between a genetic change and its physical effect – or 'phenotype' – is immensely complex.

Now, in research published today in the journal *Nature*, Drs Anton Wutz and Martin Leeb from the Wellcome Trust Centre for Stem Cell Research at the University of Cambridge report a technique which enables them to create stem cells containing just a single set of chromosomes from an unfertilised mouse egg cell. The stem cells can be used to identify mutations in genes that affect the cells' behaviour in culture. In an additional step, the cells can potentially be implanted into the mouse for studying the change in organs and tissues.

The technique has previously been used in zebrafish, but this is the first time it has been successfully used to generate such mammalian stem cells.

Dr Wutz, a Wellcome Trust Senior Research Fellowship, explains: "These embryonic stem cells are much simpler than normal embryonic mammalian stem cells. Any genetic change we introduce to the single set of chromosomes will have an easy-to-determine effect. This will be useful for exploring in a systematic way the signalling mechanisms within cell and how networks of genes regulate development."

The researchers hope that this technique will help advance mammalian genetics and our understanding of the gene-function relationship in the same way that a similar technique has helped geneticists understand the simpler zebrafish animal model.

Understanding how our genetic make-up functions and how this knowledge can be applied to improve our health is one of the key strategic challenges set out by the Wellcome Trust. Commenting on this new study, Dr Michael Dunn, Head of Molecular and Physiological Sciences at the Wellcome Trust, says:

"This technique will help scientists overcome some of the significant barriers that have so far made studying the functions of genes so difficult. This is often the first step towards understanding why mutations lead to disease and, ultimately, to developing new drugs treatments."

More information: Martin Leeb & Anton Wutz. *Derivation of haploid embryonic stem cells from mouse embryos*. *Nature*; e-pub 7 September 2011

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