

Genome editing reveals role of gene important for human embryo development

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Researchers have used genome editing technology to reveal the role of a key gene in human embryos in the first few days of development. This is the first time that genome editing has been used to study gene function in human embryos, which could help scientists to better understand the biology of our early development.

The team used genome editing techniques to stop a key gene from producing a protein called OCT4, which normally becomes active in the first few days of [human embryo development](#). After the egg is fertilised, it divides until at about 7 days it forms a ball of around 200 [cells](#) called the 'blastocyst'. The study found that [human embryos](#) need OCT4 to correctly form a blastocyst.

"We were surprised to see just how crucial this gene is for human embryo [development](#), but we need to continue our work to confirm its role" says Dr Norah Fogarty from the Francis Crick Institute, first author of the study. "Other research methods, including studies in mice, suggested a later and more focused role for OCT4, so our results highlight the need for human embryo research."

Dr Kathy Niakan from the Francis Crick Institute, who led the research, adds: "One way to find out what a gene does in the developing embryo is to see what happens when it isn't working. Now we have demonstrated an efficient way of doing this, we hope that other scientists will use it to find out the roles of other genes. If we knew the key genes that embryos need to develop successfully, we could improve IVF treatments and understand some causes of pregnancy failure. It may take many years to achieve such an understanding, our study is just the first step."

The research was published in *Nature* and led by scientists at the Francis Crick Institute, in collaboration with colleagues at Cambridge University, Oxford University, the Wellcome Trust Sanger Institute, Seoul National University and Bourn Hall Clinic. It was chiefly funded by the UK Medical Research Council, Wellcome and Cancer Research UK.

The team spent over a year optimising their techniques using mouse embryos and human embryonic [stem cells](#) before starting work on human embryos. To inactivate OCT4, they used an editing technique

called CRISPR/Cas9 to change the DNA of 41 human embryos. After seven days, embryo development was stopped and the embryos were analysed.

The embryos used in the study were donated by couples who had undergone IVF treatment, with frozen embryos remaining in storage; the majority were donated by couples who had completed their family, and wanted their surplus embryos to be used for research. The study was done under a research licence and strict regulatory oversight from the Human Fertilisation and Embryology Authority (HFEA), the UK Government's independent regulator overseeing infertility treatment and research.

As well as human embryo development, OCT4 is thought to be important in [stem cell biology](#). 'Pluripotent' stem cells can become any other type of cell, and they can be derived from embryos or created from adult cells such as skin cells. Human [embryonic stem cells](#) are taken from a part of the developing embryo that has high levels of OCT4.

"We have the technology to create and use [pluripotent stem cells](#), which is undoubtedly a fantastic achievement, but we still don't understand exactly how these cells work," explains Dr James Turner, co-author of the study from the Francis Crick Institute. "Learning more about how different genes cause cells to become and remain pluripotent will help us to produce and use stem cells more reliably."

Sir Paul Nurse, Director of the Francis Crick Institute, says: "This is exciting and important research. The study has been carried out with full regulatory oversight and offers new knowledge of the biological processes at work in the first five or six days of a human embryo's healthy development. Kathy Niakan and colleagues are providing new understanding of the genes responsible for a crucial change when groups

of cells in the very early embryo first become organised and set on different paths of development. The processes at work in these embryonic cells will be of interest in many areas of stem cell biology and medicine."

Dr. Kay Elder, study co-author from the Bourn Hall Clinic, says:
"Successful IVF treatment is crucially dependent on culture systems that provide an optimal environment for healthy embryo development. Many embryos arrest in culture, or fail to continue developing after implantation; this research will significantly help treatment for infertile couples, by helping us to identify the factors that are essential for ensuring that human [embryos](#) can develop into healthy babies."

Dr Ludovic Vallier, study co-author from the Wellcome Trust Sanger Institute and the Wellcome - MRC Cambridge Stem Cell Institute, says:
"This study represents an important step in understanding human embryonic development. The acquisition of this knowledge will be essential to develop new treatments against developmental disorders and could also help understand adult diseases such as diabetes that may originate during the early stage of life. Thus, this research will open new fields of opportunity for basic and translational applications."

More information: Norah M. E. Fogarty et al, Genome editing reveals a role for OCT4 in human embryogenesis, *Nature* (2017). [DOI: 10.1038/nature24033](#)

Provided by The Francis Crick Institute

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