Scientists identify Down's Syndrome gene

August 9 2016

Geneticists have identified an enzyme which regulates the production of sperm and egg cells in human reproduction.

The discovery furthers our understanding of a process which can often go wrong, resulting in miscarriage or infants born with Down's Syndrome, Edwards's Syndrome and other chromosomal irregularities.

Writing in the US journal *Nature Scientific Reports*, Dr Gary Kerr and colleagues stated: "Understanding how meiosis is regulated is of great importance to understanding the causes of aneuploidy and genetic disorders in humans."

The scientists looked at a process known as meiosis, which unlike normal cell division (mitosis) has two rounds of nuclear division, to ensure that when sex cells fuse with each other, they have two copies of each chromosome – one from each parent!

Miscarriage

When the cells have too many or too few (aneuploidy), babies are born with Down's Syndrome (three copies of chromosome 21); Patau syndrome (three copies of chromosome 13) and other conditions. Aneuploidy is also a leading cause of miscarriage, and with an estimated 1 in 7 pregnancies resulting in miscarriage.

In order to identify the 'genetic switch' which regulates segregation and mis-segration, the team investigated PP2ACdc55, an enzyme involved

in several cellular processes. Using flourescent tagging, they tracked the enzyme's presence on yeast models – which offer a number of commonalities in the processes of meiosis in humans.

Prior studies by Kerr el al, showed that PP2ACdc55 played an essential role in controlling the timing of metaphase to anaphase during meiosis, in other words preventing cells from prematurely exiting meiosis.

By creating random mutations in the Cdc55 gene, the team analysed the resulting 987 mutant yeast strains, characterized the mutations and worked backwords to identify the role of the gene by looking at the effect of the mutations on the resulting colonies.

Pivotal role

The resulting data suggests that PP2ACdc55 plays a pivotal role in chromosome segregation, although we are still a way from knowing how the processes go wrong.

More information: Gary W. Kerr et al. PP2ACdc55's role in reductional chromosome segregation during achiasmate meiosis in budding yeast is independent of its FEAR function, *Scientific Reports* (2016). DOI: 10.1038/srep30397

Provided by University of Salford