Breakthrough in deafness and ovarian failure syndrome

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(Medical Xpress)—Researchers from Manchester Biomedical Research Centre at Saint Mary's Hospital and the University of Manchester have identified a new gene, which increases our understanding of the rare inherited disorder Perrault syndrome.

Perrault syndrome is an inherited form of deafness that can be particularly distressing for women, as they often require hormone treatment and are unable to conceive naturally due to ovarian failure. Some patients can also have problems with their nerves, which can affect their balance and lead to difficulty with walking.

The team lead by Dr Bill Newman in the Centre for Genetic Medicine

worked with colleagues from major research centres in the United States and Pakistan, to identify the new gene that provides new insight into our understanding of infertility problems and hearing loss.

Using a powerful new genetic technique called next generation sequencing, they were able to find the responsible gene called CLPP, and establish a link between changes in this gene and the incidence of Perrault syndrome in some families. The findings have been published in a major journal, the *American Journal of Human Genetics*.

"Although the syndrome was first described in 1951 by a French doctor called Perrault, understanding the cause has eluded scientists for over 50 years," explains Emma Jenkinson, who worked on the project as part of her PhD at the University of Manchester funded by the Infertility Research Trust. "With the availability of new genetic techniques researchers have now identified four genes that can cause this condition. The genes are all important in the energy bundles in cells called mitochondria."

Provided by University of Manchester