

Male fertility genes discovered

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A new study has revealed previously undiscovered genetic variants that influence fertility in men. The findings, published by Cell Press on May 24th in the *American Journal of Human Genetics*, shed much-needed light on human reproduction and might provide answers for countless men suffering from infertility.

Despite its high incidence, [infertility](#) remains a sensitive topic. Some of the stigma surrounding infertility arises from a lack of known scientific causes. In fact, nearly a quarter of reported infertility cases remain unexplained. Research regarding the genetics of fertility has come primarily from studies involving infertile subjects. "Such studies have not been able to identify genes or pathways contributing to variation in natural human fertility," remarks Carole Ober, the lead author of the study. This is because numerous non-genetic factors, such as alcohol and [tobacco use](#), certain medications, and disease history, can contribute to infertility.

Ober and her graduate student, Gülüm Kosova, at the University of Chicago have taken a different approach. By studying a founder population, the Hutterites, Ober's research maximizes genetic influences and minimizes non-genetic ones. The Hutterites are a branch of Anabaptists who conscribe to a common set of religious and social beliefs. "Hutterites [forbid] contraception and uniformly desire large families, providing an outstanding population in which to study the genetics of normal human fertility," explains Ober. Rather than studying infertile subjects, the team included Hutterite men who had one or more child, and it took both family size and birth rate into consideration.

The study uncovered more than 40 genetic regions that influence fertility in Hutterite men. Nine of these regions were additionally found to impact sperm quality in non-Hutterites. These regions harbor genes involved in several essential biological processes, including protein regulation, nucleotide binding, and immunity, and shed light on the complexity of human fertility. Ultimately, says Ober, further studies might find that mutations in these genes underlie some of the currently unexplained cases of male infertility.

More information: Kosova et al.: “Genome-wide Association Study Identifies Candidate Genes for Male Fertility Traits in Humans.” *American Journal of Human Genetics* (2012).

Provided by Cell Press

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