Breakthrough in genetics of fibroids

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Uterine leiomyomas, also called fibroids, cause a very significant burden to women's health. They are benign tumors that occur in 60% of women by the age of 45 years and cause symptoms such as abdominal pain and discomfort, and abnormal bleeding, in about half of the cases. Fibroids are also an important cause of infertility. These tumors are the most common medical reason for hysterectomy.

Considering the clinical importance of fibroids, relatively little has been known about the mechanisms of tumorigenesis involved.

A research team at the University of Helsinki, Finland, set out to study the genetic structure of fibroids by determining the sequence of all the more than 20 000 human genes, in a series of 18 tumors. This, and further validation in over 200 fibroids, revealed very specific mutations in a gene called MED12, in as many of 70% of the studied tumors.

MED12 protein, the product of the MED12 gene, is known to play a role in regulation of general gene transcription. The observed pattern of mutations suggests that MED12 function is disrupted in a very specific manner in the majority of fibroids. The research will be published in the highly valued scientific journal *Science*.

That such a large proportion of fibroids display such specific mutations provides some hope that targeted therapies could in the future be designed based on the findings. As professor Lauri Aaltonen, the leader of the research team, comments: "This is a giant step towards understanding why fibroids arise, but towards design of targeted

therapies it is a very early step. Let's hope that this journey has begun."

Provided by University of Helsinki