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Researchers link 'housekeeping' gene with male infertility

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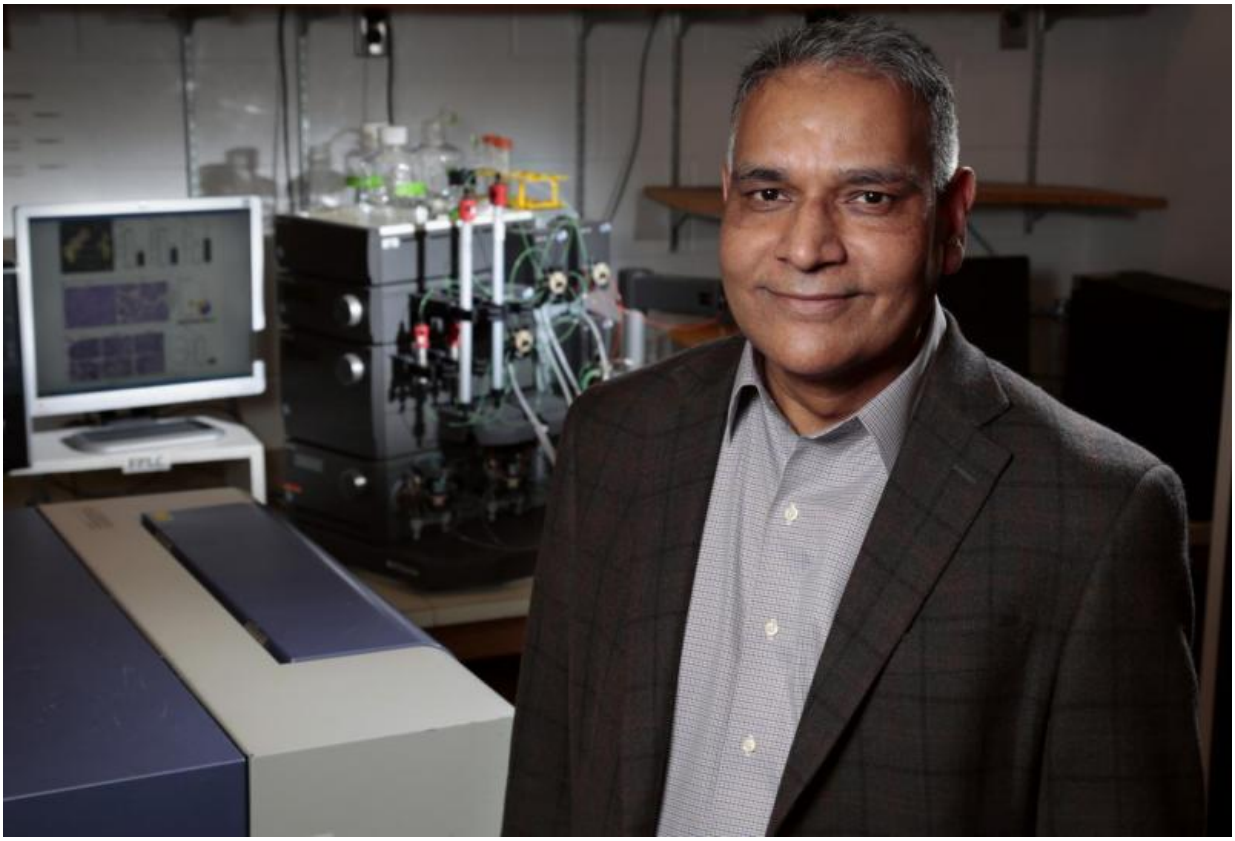

Ravindra Singh has studied SMN deficiency for years. Newly published research from his group indicates a link between SMN deficiency and male infertility in mice. Credit: Christopher Gannon.

Researchers at Iowa State University have found evidence that a

"housekeeping" gene present in every cell of the body may have a link to male infertility.

Ravindra Singh, a professor of biomedical sciences in the ISU College of Veterinary Medicine, has studied the survival motor neuron (SMN) gene for years. A deficiency in the gene, known as a "housekeeping" gene because its presence is essential for basic cellular function, can lead to neurological problems such as spinal muscular atrophy.

But Singh's laboratory discovered a link between SMN and male infertility, making it one of only a handful of genes suspected to have such a connection. The findings appeared recently in *Scientific Reports*, a peer-reviewed academic journal published by *Nature*.

Singh's group conducted a genome-wide study of SMN deficiency in mice and found surprising correlation between low levels of the gene expression and testicular size and low sperm count in male specimens.

"We need to have housekeeping genes for normal function," Singh said. "Every cell in the body requires them. Our findings seemed to uncover a new function of the gene and suggest SMN plays a role in testicular development. Mice with deficient levels of the gene had lower sperm count and more instances of infertility."

Singh said around 5 percent of men deal with infertility concerns, and little is known about the intersection of genetics and infertility. He said genome-wide association studies have linked only around six genes in the human genome to male infertility, potentially making SMN another such gene. However, Singh cautioned that further human-based research is needed to validate his group's novel findings, since these findings are based on observations in rodents.

The results also suggest SMN deficiencies could have different effects in

males and females, Singh said. Further developing that knowledge may lead physicians to take into account a patient's sex when determining treatments for SMN deficiencies.

He said the next step in the research will be to determine how early SMN deficiency can change testicular development in mice and what particular cells are targeted.

This kind of research guides new ways of thinking about medical therapies and how they interact with genetics, Singh said.

"We're heading toward an age of molecular medicine, where treatments can depend on the individual genetic differences in patients," he said. "What mutations you have could impact what treatments you receive."

More information: Eric W. Ottesen et al. Severe impairment of male reproductive organ development in a low SMN expressing mouse model of spinal muscular atrophy, *Scientific Reports* (2016). DOI: 10.1038/srep20193

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