

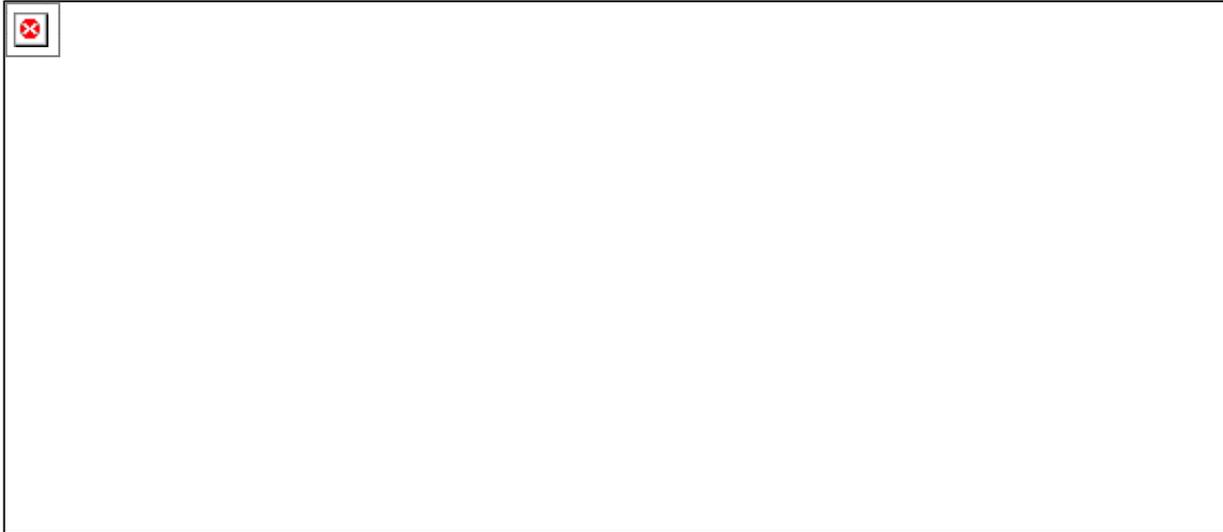
"Mighty mouse" gene found in humans

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By studying the genes of an unusually muscular child, scientists have identified a gene in humans which has also been used to create "mighty mice" in the lab.

The discovery means that successful therapies for degenerative muscle diseases in mice that target this gene might be also be effective in humans.

The strapping German boy, whose mother is a professional sprinter, is so strong that at the age of just four and a half he can hold a three kilogram weight in each hand with his arms outstretched horizontally.

The international team of scientists has shown that the boy has a single mutation in the gene for a protein called myostatin. This gene has previously been examined in mice, and blocking it can make mice twice as brawny as normal. However, until now no one knew its effects in humans.

"The idea was if we could find ways to interfere with myostatin function, perhaps we could slow down muscle loss in muscle wasting diseases and hopefully increase lifespan," explains Se-Jin Lee at Johns Hopkins University, Maryland, US, and one of the team.

"But a major question was – will this work in humans?" he says. The new study confirms that the so-called "mighty mouse" gene does have the same effects in humans.

Potential for abuse

However, the discovery also suggests a substantial "potential for abuse outside the medical arena", notes Elizabeth McNally, at the University of Chicago, in a perspective article accompanying the paper in the *New England Journal of Medicine*.

"Myostatin blockade will probably work its way into professional and amateur athletics, as well as into the ever-growing business of physical enhancement," she says.

Lee agrees the potential exists, but adds: "It would be a shame if the potential for abuse stood in the way of developing treatments for people in dire need." Lee has declared a financial interest in the research, in that he would be entitled to a share of the royalties on any commercial therapies.

The super-strong child was examined by Markus Schuelke, a neurologist at the University Medical Centre Berlin, Germany, just after birth. He was struck by the baby's protruding thigh and upper arm muscles, and

suspected a myostatin mutation.

Sequencing the myostatin gene of the boy and his mother revealed both had a single change in the gene's DNA sequence. But where the mother had only one copy of the mutant gene, the boy had two.

Splicing error

The mutation affects the way that the RNA made from the gene is spliced. This gives a misformed protein says Lee, whose team were involved in the molecular analysis. Researchers from pharmaceutical company Wyeth, in Cambridge, Massachusetts, then confirmed that there was no myostatin in the boy's blood.

Myostatin is normally made by muscle cells and circulates in the blood, acting on muscle only. The current view is that it may act on a group of muscle stem cells called satellite cells. These cells are activated in response to injury and regenerate muscle cells. "Myostatin acts to keep them in some kind of quiescent state," says Lee.

Jenny Versnel, head of research at the UK's Muscular Dystrophy Campaign, told **New Scientist**: "There's definitely sufficient evidence to demonstrate there might be a therapeutic benefit."

However, she notes that blocking myostatin, without tight regulation, in young children with Duchenne muscular dystrophy (DMD) might use up the satellite cells too early.

In early DMD, muscle breaks down, but is also regenerated by the satellite cells, so it is crucial they are not depleted. Also, a protein called dystrophin is missing in DMD. This has a structural and signalling role under the muscle membrane, so only increasing muscle mass may not solve the problem, says Versnel.

Journal reference: *New England Journal of Medicine* (vol 350, p 2682)

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