

Scientists identify gene that regulates body weight in humans and mice

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"Starting with gene discovery in a single family with morbid obesity, these studies led to the identification a gene that seems to be fundamental to regulating nutritional status," says one of the senior authors, Dr. John Martignetti of the Icahn School of Medicine at Mount Sinai in New York City. "This gene is shown to be present not only in humans and mice but also in the simplest known single-cell animal. Nature considers this gene so important that it has preserved its structure for more than 700 million years."

Drs. Martignetti and Adel Shalata, of the Ziv Medical Center Safed, Israel, and their team analized a large Israeli Arab family affected by autosomal-recessive morbid obesity and identified a truncating mutation in the gene that recently was found to encode CEP19, a ciliary protein. When the investigators deleted the Cep19 gene in mice, the animals became obese and diabetic and had increased appetites, decreased energy expenditure, and impaired fat metabolism.

"The mouse models we have generated, which can be more than twice as heavy as other mice and are insulin resistant, represent important research tools for basic biology and clinical testing," says Dr. Martignetti.

The researchers note that the <u>role</u> of this ciliary protein in maintaining a



balance between leanness and obesity remains unknown. Additional studies are needed if researchers are to determine the mechanisms behind CEP19's effects on appetite control, energy expenditure, and insulin signaling and sensitivity.

Uncovering the details behind the pathways that control body weight will only become more pressing with time. "Obesity is a global epidemic, affecting almost all areas of human health, from heart disease to cancer, and impacting upon most of the major causes of preventable death," says Dr. Martignetti. "Moreover, obesity rates are rising dramatically worldwide. If we are going to combat this disease, we need to understand its medical basis."

More information: *AJHG*, Shalata et al.: "Morbid obesity in humans and mice resulting from inactivation of the ciliary protein MO1/CEP19.." <u>dx.doi.org/10.1016/j.ajhg.2013.10.025</u>

Provided by Cell Press

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