

Genetic studies reveal new causes of severe obesity in childhood

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Scientists in Cambridge have discovered that the loss of a key segment of DNA can lead to severe childhood obesity. This is the first study to show that this kind of genetic alteration can cause obesity. The results are published today in *Nature*.

The study, led by Dr Sadaf Farooqi from the University of Cambridge and Dr Matt Hurles from the Wellcome Trust Sanger Institute, looked at 300 children with severe obesity.

The team scanned each child's entire genome looking for types of mutation known as copy number variants (CNVs). CNVs are large chunks of DNA either duplicated or deleted from our genes. Scientists believe this type of mutation may play an important role in genetic diseases.

By looking for CNVs that were unique in children with severe obesity, compared with over 7,000 controls (apparently healthy volunteers from the Wellcome Trust Case Control Consortium 2), they found that certain parts of the [genome](#) were missing in some patients with severe obesity.

According to Dr Farooqi: "We found that part of chromosome 16 can be deleted in some families, and that people with this deletion have severe obesity from a young age.

"Our results suggest that one particular gene on chromosome 16 called SH2B1 plays a key role in regulating weight and also in handling blood

sugar levels. People with deletions involving this gene had a strong drive to eat and gained weight very easily."

Dr Matt Hurles adds: "This is the first evidence that copy number variants have been linked to a metabolic condition such as obesity. They are already known to cause other disorders such as autism and learning difficulties."

The findings also have implications for diagnosing severe [childhood obesity](#), which has on occasion been misattributed to abuse. Some of the children in the study had been formally placed on the Social Services 'at risk' register on the assumption that the parents were deliberately overfeeding their children and causing their severe obesity. They have now been removed from the register.

"This study shows that [severe obesity](#) is a serious medical issue that deserves scientific investigation," says Dr Farooqi. "It adds to the growing weight of evidence that a wide range of genetic variants can produce a strong drive to eat. We hope that this will alter attitudes and practices amongst those with professional responsibility for the health and well-being of children."

Obesity is increasing throughout the world and is now recognised as a major global public health concern. Although the increased prevalence of [obesity](#) over the past 30 years is undoubtedly driven by environmental factors, genetic factors play a major role in determining why some people are more likely to gain weight than others.

More information: The paper, Elena G. Bochukova et al, 'Large, rare chromosomal deletions associated with severe early-onset obesity' is published in *Nature* on 6 December 2009.

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