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| |  |  |  | | --- | --- | --- | | **‘Gene overdose’ causes extreme thinness**   |  |  | | --- | --- | |  |  |   **loading . . .**  http://www3.imperial.ac.uk/newseventsxml/images/ajax-loader.gif | |

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| Scientists have discovered a genetic cause of extreme thinness for the first time, in a new study published in the journal Nature - News release  The research shows that people with extra copies of certain genes are much more likely to be very skinny.  In one in 2000 people, part of chromosome 16 is duplicated, making men 23 times and women five times more likely to be underweight.  Each person normally has a copy of each chromosome from each parent, so we have two copies of each gene.  But sometimes sections of a chromosome can be duplicated or deleted, resulting in an abnormal 'dosage' of genes.  In a study examining the DNA of over 95,000 people, researchers at Imperial College London and the University of Lausanne have identified that duplication of a part of chromosome 16 is associated with being underweight, defined as a a body mass index below 18.5.  Half of all children with the duplication in the study have been diagnosed with a 'failure to thrive', meaning that their rate of weight gain is significantly lower than normal.  A quarter of people with the duplication have microcephaly, a condition in which the head and brain are abnormally small, which is associated with neurological defects and shorter life expectancy.  Last year, the same researchers discovered that [people with a missing copy of these genes are 43 times more likely to be morbidly obese](http://www.nature.com/nature/journal/v463/n7281/full/nature08727.html).  [Professor Philippe Froguel](http://www1.imperial.ac.uk/medicine/people/p.froguel/), from the [School of Public Health](http://www1.imperial.ac.uk/publichealth/) at Imperial College London, who led the study, said: "The dogma is that we have two copies of each gene, but this isn't really true.  The genome is full of holes where genes are lost, and in other places we have extra copies of genes.  In many cases, duplications and deletions have no effect, but occasionally they can lead to disease.  "So far, we have discovered a large number of genetic changes that lead to obesity.  It seems that we have plenty of systems that increase appetite since eating is so important – you can suppress one and nothing happens.  This is the first genetic cause of extreme thinness that has been identified.  "One reason this is important is that it shows that failure to thrive in childhood can be genetically driven.  If a child is not eating, it’s not necessarily the parents' fault.  "It’s also the first example of a deletion and a duplication of one part of the genome having opposite effects.  At the moment we don’t know anything about the genes in this region.  If we can work out why gene duplication in this region causes thinness, it might throw up new potential treatments for obesity and appetite disorders.  We now plan to sequence these genes and find out what they do, so we can get an idea of which ones are involved in regulating appetite."  The part of chromosome 16 identified in the study contains 28 genes.  Duplications in this region have previously been linked with schizophrenia, and deletions with autism.  The study was funded by the [Medical Research Council](http://www.mrc.ac.uk), the [Wellcome Trust](http://www.wellcome.ac.uk), and other sources.  **For further information please contact:**  Sam Wong Research Media Officer Imperial College London Email: [sam.wong@imperial.ac.uk](mailto:sam.wong@imperial.ac.uk) Tel: +44(0)20 7594 2198 Out of hours duty press officer: +44(0)7803 886 248  **Notes to editors:**  1. Journal reference: S. Jacquemont et al. '[Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus](http://www.nature.com/nature/journal/vaop/ncurrent/full/nature10406.html).' Nature, 31 August 2011.  2. About Imperial College London  Consistently rated amongst the world's best universities, Imperial College London is a science-based institution with a reputation for excellence in teaching and research that attracts 14,000 students and 6,000 staff of the highest international quality. Innovative research at the College explores the interface between science, medicine, engineering and business, delivering practical solutions that improve quality of life and the environment - underpinned by a dynamic enterprise culture.  Since its foundation in 1907, Imperial's contributions to society have included the discovery of penicillin, the development of holography and the foundations of fibre optics. This commitment to the application of research for the benefit of all continues today, with current focuses including interdisciplinary collaborations to improve global health, tackle climate change, develop sustainable sources of energy and address security challenges.  In 2007, Imperial College London and Imperial College Healthcare NHS Trust formed the UK's first Academic Health Science Centre. This unique partnership aims to improve the quality of life of patients and populations by taking new discoveries and translating them in to new therapies as quickly as possible.  Website: [www.imperial.ac.uk](http://www.imperial.ac.uk)  3. About the Medical Research Council For almost 100 years the Medical Research Council has improved the health of people in the UK and around the world by supporting the highest quality science. The MRC invests in world-class scientists. It has produced 29 Nobel Prize winners and sustains a flourishing environment for internationally recognised research. The MRC focuses on making an impact and provides the financial muscle and scientific expertise behind medical breakthroughs, including one of the first antibiotics penicillin, the structure of DNA and the lethal link between smoking and cancer. Today MRC funded scientists tackle research into the major health challenges of the 21st century. [www.mrc.ac.uk](http://www.mrc.ac.uk)  4. About the Wellcome Trust  The Wellcome Trust is a global charitable foundation dedicated to achieving extraordinary improvements in human and animal health. It supports the brightest minds in biomedical research and the medical humanities. The Trust's breadth of support includes public engagement, education and the application of research to improve health. It is independent of both political and commercial int erests. Website: www.wellcome.ac.uk |