Loss or gain of genetic material: A weighty issue

0 01/09/2011 03:24:00

Share / Save

Researchers Xavier Estivill, Mónica Gratacós and Sergi Villatoro, from the Genes and Disease programme of the Centre for Genomic Regulation (CRG) in Barcelona, have taken part in a scientific study identifying an area of the genome having opposing effects with respect to body weight.

The study, to be published on 1st September in the journal Nature, involved more than 100 research centres worldwide, five of which are Spanish, and included more than 95,000 individuals with intellectual or developmental disability and psychiatric disorders as well as individuals from the general populace.

The researchers identified 132 subjects with a duplication of one region of chromosome 16p11.2, among who was a high frequency of individuals with low birth weight and low body mass index, in many cases accompanied by selective and restrictive feeding behaviour. Adult carriers of the duplication, and particularly men, were 24 times more likely to be clinically underweight.

Previous studies had found that when this same region on chromosome 16p11.2 is deleted, it is associated with a 43-times greater risk of suffering morbid obesity with hyperphagia and mental retardation. The fact that opposing changes (loss or gain of genetic material) in the same region of the genome give rise to opposite phenotypes (being obese or underweight), suggests that this region may contain genes that regulate energy balance and affect feeding behaviour, being able to manifest as hyperphagia (increased intake) or anorexia (refusal to feed) depending on the number of copies of the genes that an individual has.

The CRG researchers Xavier Estivill, Mónica Gratacós and Sergi Villatoro led the genetic studies into the samples from the Spanish subjects.

http://www.crg.eu

Full bibliographic information Jacquemont, S. et al., "Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus". Nature, Sept 1, 2011, doi:10.1038/nature10406