

New syndrome causing obesity and intellectual disability identified

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Scientists at The University of Manchester have discovered a rare new genetic syndrome of obesity, over-eating, mental and behavioural problems in six families, from across the world.

Dr Siddharth Banka, Clinical Senior Lecturer at the Manchester Centre for Genomic Medicine, who led the study, explained: "Our team has identified that this new syndrome is caused by a small deletion on chromosome 6 that affects the function of hypothalamus, a region of the brain that plays a number of important roles in the body."

Working in collaboration with Dr Eric Glasgow of the Georgetown University Medical Center in Washington D.C., the two teams used zebrafish models to study the consequences of chromosome 6 deletion and showed that the deletion has an effect on specific cells in the hypothalamus that produce a hormone called oxytocin.

This could help explain why sufferers are often severely obese, find it difficult to control their appetites and are prone to mood swings and being withdrawn, the researchers say.

The study, published in the latest issue of the *American Journal of Human Genetics* represents an important step in our understanding of how the hypothalamus and oxytocin control appetite and behaviour, says Dr Banka, who is also a Consultant Clinical Geneticist at Saint Mary's Hospital, in Manchester. "This finding demonstrates the power of human genetic study of rare conditions."

Although at an early stage, the researchers hope that the findings will help form part of a picture of how the [hypothalamus](#) works and may lead to new treatments in the future.

The paper, "Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability" was published Jan. 28 in the *American Journal of Human Genetics*.

More information: Paul R. Kasher et al. Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability, *The American Journal of Human Genetics* (2016). [DOI: 10.1016/j.ajhg.2015.12.014](https://doi.org/10.1016/j.ajhg.2015.12.014)

Provided by University of Manchester

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