



Better understanding of the all-consuming Prader-Willi Syndrome

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Australian researchers have uncovered facts that take us a step closer towards understanding Prader-Willi Syndrome, a harrowing genetic disorder that causes insatiable appetite for life.

One in 25,000 babies will be born with Prader-Willi Syndrome. Small and floppy at birth, within two years the children start to become fatter because they feel constantly hungry, no matter how much they eat.

Prader-Willi children have such voracious appetites that parents are forced to put locks on refrigerators and place food out of sight and out of reach. The children grow into ravenous obese adults with cardiovascular problems and a short lifespan, some not surviving past their 20s or 30s.

Dr Alex Viardot and Professor Lesley Campbell from Sydney's Garvan Institute of Medical Research undertook a study comparing 12 patients from Royal Prince Alfred Hospital's Prader-Willi clinic with 12 obese people, matched exactly for fat mass. Accurate fat matching was necessary to determine which detrimental effects were caused by the disease itself, rather than by obesity alone. Both groups were compared with 10 lean people.

It is well known that abdominal fat activates the immune system, causing unwanted inflammation. It was especially surprising, therefore, that despite having the same amount of abdominal fat, the Prader-Willi group showed much more activation of their innate immune systems, the body's first line of defence.

Notably, significantly higher numbers of the Prader-Willi group had sleep apnoea, a possible reason for their overactive immune response.

These findings are now published online in the *Journal of Clinical Endocrinology and Metabolism*.

"We did this study to find out more about Prader-Willi and to give us better insight into obesity and appetite control generally," said project leader Professor Campbell.

"Identifying possible causes of the immune activation in this disease will provide targets for treatment, which in turn will improve early mortality."

"Prader-Willi is a particularly distressing, life-diminishing disease that can involve developmental delay, growth hormone deficiency and sex hormone deficiency as well as insatiable appetite."

"Although it is the most common known genetic cause of obesity, we still know very little about it."

“Obviously we want to discover as much as we can about Prader-Willi Syndrome and find treatments that reduce its torment for patients and their carers.”

“The main message from this project was that the Prader-Willi group had more activation of their immune system than the obese-only group.”

“And while there is always low-level inflammation associated with obesity, there seemed to be an intrinsic inflammatory process going on in the Prader-Willi group that was excessive.”

“Five members of the Prader-Willi group had sleep apnoea, in contrast to one from the obese group, and that may have contributed towards their fired-up immune system.”

“We believe that if we treat sleep apnoea, we may at least partially suppress the heightened immune response and decrease associated cardiovascular risk.”

“With every small piece of knowledge, such as this study provides, comes hope that we will eventually find the critical clue – the thing that will allow us to turn the appetite switch off.”

ABOUT GARVAN

The Garvan Institute of Medical Research was founded in 1963. Initially a research department of St Vincent's Hospital in Sydney, it is now one of Australia's largest medical research institutions with nearly 500 scientists, students and support staff. Garvan's main research programs are: Cancer, Diabetes & Obesity, Immunology and Inflammation and Neuroscience. Garvan's mission is to make significant contributions to medical science that will change the directions of science and medicine and have major impacts on human health. The outcome of Garvan's discoveries is the development of better methods of diagnosis, treatment, and ultimately, prevention of disease.

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