'Teashirt' gene links autism and kidney problems, new study finds

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A gene dubbed the 'Teashirt' by its discoverers has been identified as a link between children with kidney problems and autism, in a new study which has implications for how doctors working on both conditions

administer tests to their patients.

The new paper, published in the journal *Nature Genetics*, was led by the Developmental Biology Institute of Marseille, collaborating with The University of Manchester, and it describes the effects of mutations of Teashirt in people and mice.

The gene, formally named Tshz3, had already been implicated by the joint research team in 2008 as being essential for development of smooth muscle in the wall of the ureter. Mutant mice were born with 'blown-up' kidneys because their ureters failed to actively propel urine down to the bladder.

Professor Adrian Woolf from The University of Manchester, then working as a children's consultant in London, discovered that one of his patients born with abnormal kidneys had a deleted Tshz3 gene and also displayed characteristics of autistic spectrum disorder.

The French team also realised that mice with Tshz3 mutation not only had kidney problems but also displayed learning difficulties.

The findings sparked a global search of other kidney clinics, which returned ten more patients with similar symptoms. After genetic testing, it was confirmed that the same gene was missing in all of them – findings which are published in the new paper.

Professor Woolf said: "The mutant mouse kidney looks just like 'hydronephrosis', the distended kidney seen in about 1 in 1,000 individuals when they are screened by sonar scans as unborn babies. It now appears that this gene is linked to at least some of these cases and that it also has implications for how our brains work in childhood."

The research was led by Professor Laurent Fasano in Marseille who

discovered the teashirt gene in fruit flies in 1991. He said: "The sooner the better; early detection of this new condition will favour early behavioural therapies, which is good for the kids and their family."

The link between the two diseases has implications for how doctors work with patients who display either kidney or learning problems.

Professor Woolf, who is also a consultant at the Royal Manchester Children's Hospital where he runs a renal genetics clinic, added: "A fairly simple genetic test on patients being treated for either kidney problems or autistic spectrum disorder could identify whether the Teashirt gene is missing and also highlight that the patient may need investigation for the other condition. Time will tell whether TSHZ3 plays a role in many more cases than we've currently been able to identify."

More information: Xavier Caubit et al. TSHZ3 deletion causes an autism syndrome and defects in cortical projection neurons, *Nature Genetics* (2016). DOI: 10.1038/ng.3681

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