# Further potential insight into the complex neuropathology of Down's syndrome

Researchers at the University of Bristol have revealed new insight into the function of a key protein attributed to impaired learning and memory in Down's syndrome. The findings, published online in Nature Cell Biology, offer further molecular insight into how the reduced level of this key protein termed 'sorting nexin-27' [SNX27] may contribute to learning and memory problems associated with Down's syndrome.

The Bristol-based team now reveal how SNX27 forms the core component of an ancient protein complex which functions to control the abundance of a select group of proteins at the surface of cells. Included among these proteins are numerous transporters that regulate the cell's ability to take up various nutrients, including glucose and metal ions such as zinc and copper. In cells lacking SNX27, the level of these transporters is reduced and the cell's ability to take up nutrients is adversely perturbed.

Peter Cullen, Professor of Biochemistry from the University's School of Biochemistry and senior author of the Wellcome Trust-funded study, said: "Besides the previously recognised role of SNX27 in regulating the synaptic activity of neurones, our study suggests that the lack of SNX27 expression observed in Down's syndrome may also lead to a reduced metabolic activity that may adversely affect neuronal development and cognitive function.

"Further analysis of the effect of reduced SNX27 expression on the synaptic and metabolic activity of specific neuronal populations will certainly provide much needed molecular insight into the complex neuropathology of Down's syndrome as well as other neurological conditions."

**Further information:**

Paper

The Wellcome Trust-funded study entitled "A global analysis of SNX27-retromer assembly and cargo specificity reveals a function in glucose and metal ion transport" by Florian Steinberg (1), Matthew Gallon (1), Mark Winfield (2), Elaine Thomas (1), Amanda J. Bell (1), Kate J. Heesom (3), Jeremy M. Tavaré (1) and Peter J. Cullen (1,4).


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Source: Bristol University