

High blood pressure may be caused by mutation in adrenal gland

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High blood pressure may in some cases be caused by benign hormoneproducing tumours of the adrenal cortex. A joint Swedish-American research effort has now uncovered a genetic cause behind the occurrence of such tumours. The findings were published today in the journal *Science*.

Approximately 5 per cent of patients with elevated blood pressure have benign endocrine tumours in their <u>adrenal gland</u>. The tumours produce abnormally high levels of the hormone aldosterone (the condition is known as primary aldosteronism), which in turn causes blood pressure to rise. Why the tumours arise has thus far been unknown.

Researchers at the Endocrine Surgery Unit at the Department of Surgical



Sciences at Uppsala University Hospital, in collaboration with colleagues at the Yale School of Medicine, New Haven, Connecticut, USA, have now identified a causal mechanism.

The genetic codes of the relevant genes in tumour and normal tissue were analyzed by means of exome sequencing, a new technique. The results showed that mutation in a specific potassium channel (KCNJ5) – which has a role in the passage of molecules into and out of cells – results, in a large number of cases, in tumour growth and overproduction of the hormone aldosterone. This leads to increased levels of potassium and water in the blood, which raise the blood pressure. The same mutation turns out to underlie a rare genetic disease characterised by a difficult-to-treat high blood pressure condition.

"The discovery may help to improve diagnostics in connection with primary aldosteronism and cases of severe blood pressure elevation," says Peyman Björklund, a researcher at the Department of Surgical Sciences at Uppsala University. "The mutated potassium channel also represents a potential target molecule for treatment of the tumours in question."

Provided by Uppsala University

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