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Search for blood pressure secrets reveals a surprising new syndrome

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Yale researchers investigating the genetic causes of blood pressure variation have identified a previously undescribed syndrome associated with seizures, a lack of coordination, developmental delay and hearing loss.

The findings, published this week in Proceedings of the National Academy of Sciences, illustrate the power of genetic studies not only to find causes of chronic ailments, but also to identify a common cause in a seemingly unrelated set of symptoms in different parts of the body.

"Our ability to unequivocally and rapidly define new syndromes and their underlying disease genes has progressed dramatically in recent years," said Richard Lifton, chair of the Department of Genetics at the Yale School of Medicine and senior author of the study. "A study like this would have taken years in the past, but was accomplished in a few weeks by a single fellow in the lab."

The discovery of the new syndrome was made by Ute Scholl, a post-doctoral fellow in Lifton's lab, who was conducting a genetic analysis of 600 patients for causes of salt-handling defects of the kidney, which lead to high or low blood pressure. She identified a group of five patients from four families in Afghanistan, Turkey, Great Britain and Canada who had, in addition to a salt-handling defect, diverse neurologic problems. The similar clinical features of these patients suggested that all might be caused by a single defect, and in a matter of weeks she found that all five had inherited mutations in the gene KCNJ10, a

potassium channel that is expressed in the brain, inner ear and kidney.

Previous studies in mice had implicated this channel in the brain; however the human findings have clearly established its essential role in renal salt handling. In the brain, the mutation apparently interferes with the ability to clear neurotransmitters and potassium from synapses, leading to seizures. The same channel is required for sound transduction in the inner ear. The new findings implicated this channel in maintenance of the activity of the sodium pump in the kidney, the major driver of salt reabsorption.

The authors call this new syndrome SeSAME because of the clinical features of seizures, sensorineural deafness, ataxia, mental retardation and electrolyte imbalance.

Lifton said he hopes the research will not only help doctors identify people with the new syndrome but also lead to greater recognition that patients with apparently complicated syndromes may often have simple underlying defects that can be understood.

Source: Yale University (news : web)