A Novel Diagnostic for the Identification of Surgically Correctable Hypertension and Potassium Channel Antibody Therapy

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Description:

- Hypertension affects more than 1.2 billion people worldwide with over 76 million individuals in the US.
- The majority of adult hypertension diagnoses are considered:
  - “primary” — having no obvious medical cause
  - 5–10% of cases are considered “secondary” due to primary aldosteronism (PA)
  - Of this population, an estimated 1-5% of patients have a tendency to develop severe, difficult to treat hypertension caused by aldosterone-producing adenomas
- In a collaborative study, Yale researchers have developed a novel DNA-based diagnostic that detects mutations in ion channels in patients presenting with hypertension due to PA.
- Somatic mutations, as illustrated in the figure, cause increased sodium ion conductance, cell depolarization, and intracellular calcium levels have been revealed in 47% of sporadically occurring aldosterone-producing adenomas.
- This genetic test:
  - can eliminate the requirement for the conventional, invasive adrenal vein sampling and streamline the diagnostic evaluation of severe hypertension
  - indicate a direct diagnosis for a surgically correctable form of the disease
  - can be used as a stand-alone or a companion diagnostic in all patients with worsening or difficult-to-treat hypertension.

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Publications:

K⁺ channel mutations in adrenal aldosterone-producing adenomas and hereditary hypertension, Science. 2011 Feb 11;331(6018):768-72.

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