

# Early onset hypertension with primary hyperaldosteronism through mutation in the calcium channel CACNA1H - case report.

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**Introduction:** The genetic causes of primary hyperaldosteronism are still being discovered.

We present the case of a **17 years old girl** who was found by accident with **severe hypertension** (TA 180/100 mmHg, bilateral). Her personal history was unremarkable. Her father had hypertension and a paternal aunt had died at 55 due to a stroke. Both her sisters and mother had normal blood pressure

The clinical exam showed a slightly overweight girl (157.2 cm; 52.9 kg; BMI – 22 kg/m<sup>2</sup>) with normal pubertal development and no signs of virilisation or glucocorticoid excess

Heart US: concentric left ventricle hypertrophy (posterior wall 11 mm; interventricular septum 13 mm) with diastolic dysfunction of delayed relaxation

Abdominal and vascular US: normal (no aortic or renal arteries stenosis)  
Abdominal CT - normal

Normal ionogram (Na- 139 mM/L; K – 4.2 mM/L; Cl – 104 mM/L)  
normal renal function (creatinine – 0.68 mg/dL)  
Normal thyroid function

low normal ACTH (11.17, N 3-66 pg/mL); high normal cortisol (17.35, N -6.2-19 mcg/dL);  
Normal cromogranin A (71, N 20-125 ng/mL);  
Slightly increased serotonin (465, N 80-450 ng/mL)

normal urinary and plasmatic metanephrine and normetanephrine (uMN – 86; N 50-350 mcg/24h; uNMN – 178; N -100-600 mcg/24h; pMN – 69, N 10-90 pg/ML; pNMN – 126, N - 15-180 pg/mL);

**The plasmatic renin was very low** (<0.3; N 4.4-46.1 mcUI/mL), with **high normal aldosterone** (341 pg/mL, N orthostatism 25.2-392pg/mL) and an increased aldosterone/renin ratio (1136, N< 19).

**After 2 days of dexamethasone (0.5 mg every 6 hours) her aldosterone remained high (428) with low renin** and high aldosterone/renin ratio (856).

She was diagnosed with **secondary hypertension due to primary hyperaldosteronism**. Genetic testing showed she was **heterozygous for a mutation in CACNA1H (CACNA1H<sup>M1549V</sup>)** witch causes early onset hyperaldosteronism and hypertension. The mutation was not present in her parents.

The aldosterone secretion is mediated thru calcium channels, like Ca<sub>v</sub>3.2, which is abundant in the human adrenal glomerulosa. The mutation - CACNA1H<sup>M1549V</sup> causes much slower inactivation of the calcium channel and activation at more hyperpolarized potentials thus resulting in calcium channels that remain open longer, higher intracellular calcium and higher aldosterone secretion.

Primary familial hyperaldosteronism determines primary hypertension with early age onset and severe evolution. Rare causes are still being discovered furthering our knowledge of the mechanisms of adrenal function.

There are no conflicts of interest.