A Long Follow-Up of a Juvenile Case with Adrenal Cortical and Medullary Hyperplasia

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Introduction: Adrenal cortical and medullary hyperplasia combination is a rare condition in where medullary or cortical hyperfunction may be a major presenting symptom and its etiology and genetic basis are unknown. The *PRKAR1A* gene codes protein kinase-A-type1-α regulatory subunit, and its mutation causes adrenocortical tumors. Mitochondrial succinate dehydrogenase (SDH) complex's B, C, D subunit mutations are associated with pheochromocytomas and paragangliomas.

Case: An eleven-year-old male patient applied with stomachache. Anthropometric and systemic examinations were normal. Abdominal ultrasonography, computed tomography (CT) and suprarenal magnetic resonance imaging revealed a 25x18x20 mm sized lesion at medial cruris of the left suprarenal gland. On his 24-hour blood pressure monitoring, 44% systolic hypertension and hypertension attacks high as 187/159 mmHg were detected. When his suprarenal cortex hormones, adrenaline, noradrenaline, metanephrine levels were normal, 24-hour urine normetanephrine level was high as 248.9 ug/day (47-176). There was not any pathologically increased metabolic activity of the left suprarenal on tumor 18F-FDG PET/CT. With preliminary diagnosis of pheochromocytoma, left surrenalectomy was performed after a pre-operative alpha- and beta-blocker therapy. In pathological examination, it was seen that cortex was extremely thick with nodularities in some places; other than cortical hyperplasic sites, medullary thickness has reached the cortex thickness in some places and was accompanied by focal medullary hyperplasia. The patient had to use antihypertensive drugs for post-operative 6 months. On his 5-year follow-up, repetitive 24-hour blood pressure monitoring, suprarenal hormones, catecholamine and catecholamine metabolites levels were normal. On periodic imaging, the right suprarenal was normal and there were postoperative changes in the left suprarenal. On genetic analysis, there was not any *PRKAR1A* gene mutation, deletion, or duplication. SDH subunit mutation analysis is still going on.

Result: It is important to know the pathophysiology and specific genetic basis in adrenal cortical and medullary nodular hyperplasia cases which have not been defined in childhood, by means of therapy strategies and long-term follow-up.

Key words: Adrenal corticomedullary hyperplasia, *PRKAR1A* gene, SDHB, SDHC, SDHD