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## **Renovascular Hypertension in a Girl with Williams-Beuren Syndrome**

**Se Jin Park**<sup>1</sup>, Ki Soo Pai<sup>2</sup>, Jae Il Shin<sup>3</sup>

<sup>1</sup>Department of Pediatrics-Nephrology, Geje Children's Hospital, Korea, Republic of

<sup>2</sup>Department of Pediatrics-Nephrology, Ajou University Hospital, Korea, Republic of

<sup>3</sup>Department of Pediatrics-Nephrology, Severance Hospital, Korea, Republic of

### **Case Study:**

Williams-Beuren syndrome (WBS), also known as Williams syndrome, is a multisystem genetic disorder caused by hemizygous deletion of 1.5 to 1.8 Mb on chromosome 7q11.23, encompassing about 28 genes including the elastin gene, ELN. Williams-Beuren syndrome affects between 1 in 7,500 to 1 in 20,000 people at birth. This condition is characterized by mild to moderate intellectual disability or learning problems, unique personality characteristics, distinctive facial features, and heart and blood vessel (cardiovascular) problems such as supravalvular aortic stenosis (SVAS).

People with WBS may also have hypercalcemia in infancy, developmental delays, problems with coordination, and short stature. Additional signs and symptoms involving the eyes and vision, the digestive tract, and the urinary system are also possible. The diagnosis is typically suspected based on symptoms and confirmed by genetic testing: micro-array analysis or the fluorescent in situ hybridization (FISH) test. Treatment includes special education programs and various types of therapy such as surgery to correct heart and blood vessel problems.

Although SVAS and pulmonary artery stenosis are the most common vascular anomalies noted in patients with WBS, renal artery stenosis and stenotic anomalies of the abdominal aorta can also occur, resulting in high blood pressure. Here, we present our experience regarding renovascular hypertension in a 10-year-old girl with WBS.