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## **Exploring the Significance of Japanese 3-Year-Old Urine screening in Diagnosing Alport Syndrome Patients**

**Hideaki Kitakado**, Yuta Inoki, Yuta Ichikawa, Yu Tanaka, Chika Ueda, Atsushi Kondo, Nana Sakakibara, China Nagano, Tomoko Horinouchi, Kandai Nozu  
Department of Pediatrics-Nephrology, Department of Pediatrics, Kobe University Graduate School of Medicine, Japan

**Objectives :** Alport syndrome (AS) is an inherited kidney disease caused by pathological variants in COL4A3/4/5, encoding  $\alpha 3/4/5$  chains of type 4 collagen. The prevalence is estimated at 1 in 5000 individuals, with autosomal dominant (ADAS)/recessive (ARAS) forms and X-linked (XLAS) forms. Symptoms typically begin with hematuria in the early stages, progressing to increased proteinuria and eventual kidney failure. The treatment by Renin-Angiotensin-Aldosterone System Inhibitors can extend kidney prognosis, so cases with proteinuria are strongly recommended to start these medicines. 3-year-old urine screening is available for all children in Japan.

**Methods :** We examined diagnostic events in 337 cases of AS diagnosed by genetic testing from August 2015 to September 2023, under 18 years old.

**Results :** Among 337 cases, 175 were males (52%), and the median age was 6 years. The most common event for detecting urinary abnormalities was 3-year-old urine screening, accounting for 100 cases (30%). Other events included the incidental detection during the examination of other diseases: 80 cases (24%), macroscopic hematuria episodes: 78 cases (23%), school urine screenings: 20 cases (6%), and nursery urine screenings: 18 cases (5%). Among the 100 cases identified by 3-year-old urine screening, the breakdown of each genetic form was XLAS males: 28%, XLAS females: 45%, ADAS: 20%, and ARAS: 7%. Of these, 42 cases showed both hematuria and proteinuria, with XLAS males: 15/28 cases (54%), XLAS females: 15/45 cases (33%), ADAS: 6/20 cases (30%), and ARAS: 6/7 cases (86%).

**Conclusions :** 3-year-old urine screening is the most common diagnostic event for AS, encompassing all genetic forms. The proportion of cases with proteinuria is higher in ARAS and XLAS males but present in all genetic forms. Urinary protein is already observed in over 40% of cases, enabling early intervention. The benefits of 3-year-old urine screening for the early detection of AS are immeasurable.