KSN 2019
Seoul, Korea · May 23 - 26

Abstract Type : Oral
Abstract Submission No. : OR-1030

Features of Autosomal Recessive Alport Syndrome: A Systematic Review

Jiwon Lee[1], Kandai Nozu[2], Dae Eun Choi[3], Hee Gyung Kang[4], Hae Il Cheong[4]
[1]Department of Pediatrics, Chungnam National University Hospital, Korea, Republic of
[2]Department of Pediatric Nephrology, Kobe University Graduate School of Medicine, Kobe, Japan, Japan
[3]Department of Internal Medicine-Nephrology, Chungnam National University Hospital, Korea, Republic of
[4]Department of Pediatrics-Nephrology, Seoul National University College of Medicine, Korea, Republic of

Objectives: Alport syndrome is a major hereditary nephritis leading to end-stage renal disease (ESRD). Although X-linked inheritance is the most common form, cases with autosomal recessive inheritance with mutations in *COL4A3* or *COL4A4* are being increasingly recognized.

Methods: A systematic review was conducted on autosomal recessive (ARAS, OMIM 203780) form of Alport syndrome. Electronic databases were searched using related terms (till Oct 10th, 2018). Information regarding clinical manifestation and genotypes was collected. The results were peer-reviewed.

Results: From 1601 articles searched, there were 26 eligible studies with 148 patients. Female and male patients were equally affected. About 62% of patients had ESRD, 64% had sensorineural hearing loss (SNHL) and 17% had ocular manifestation. The median at onset was 2.5 years for hematuria, 21 years for ESRD, and 13 years for SNHL. Patients with no missense mutations had more grave outcomes at earlier ages, while those who had one or two missense mutations had delayed onset of age and lower prevalence of extrarenal manifestations. Of 49 patients available for electron microscopy pathology, 42 (86%) had typical glomerular basement membrane (GBM) changes, while 5 (10%) patients showed GBM thinning only.

Conclusions: Autosomal Alport syndrome requires clinical suspicion and close follow-up. SNHL seem to develop earlier than previously reported. There was a genotype-phenotype correlation according to the number of missense mutations. Possessing missense mutations had delayed onset of age and lower prevalence of extrarenal manifestations.

Figure 1. Flow chart of literature search

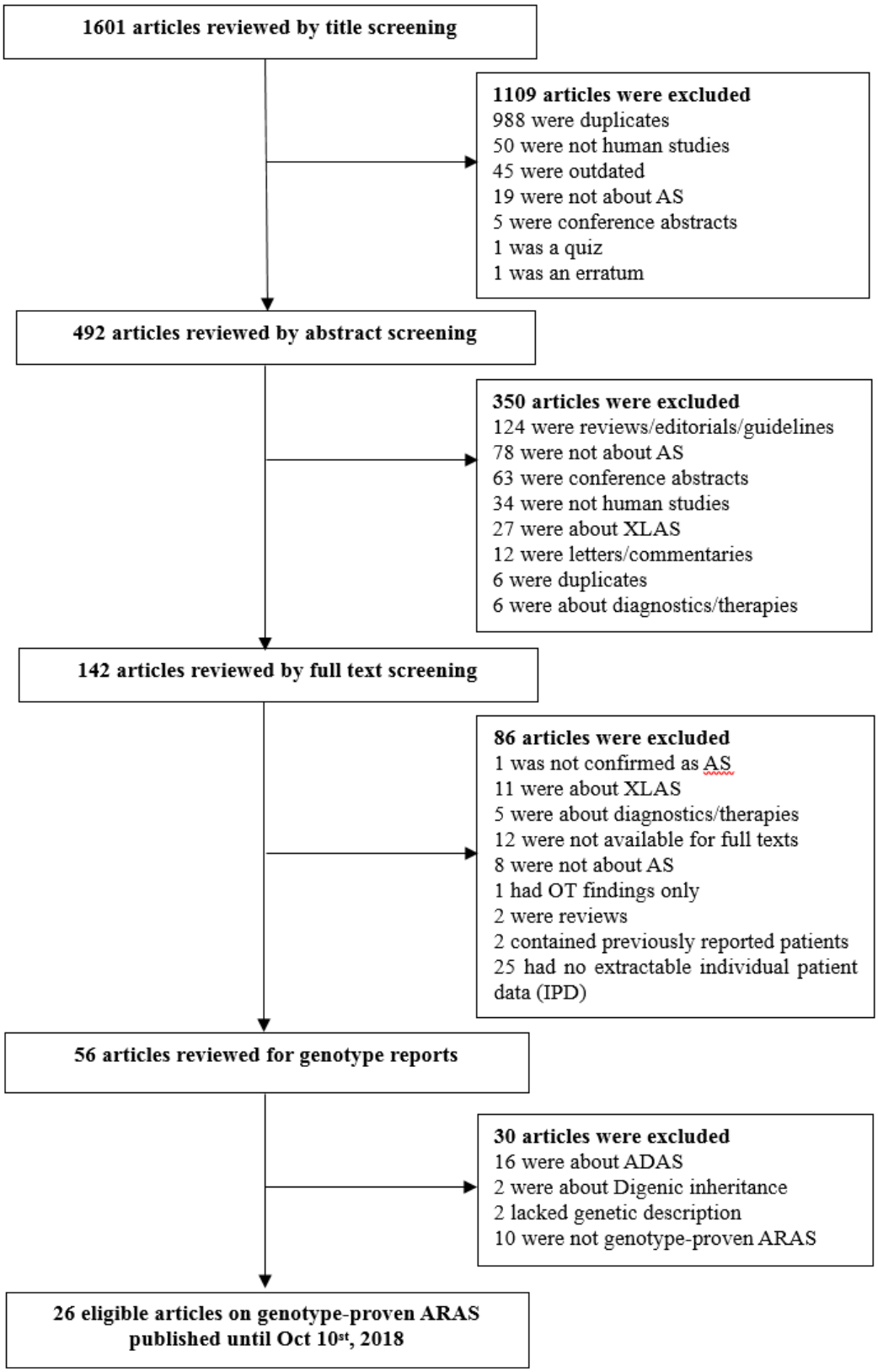

Figure 1. Flow chart of literature search

Figure 2. ESRD-free survival of ARAS patients according to (A) presence of missense mutation and (B) number of missense mutations.

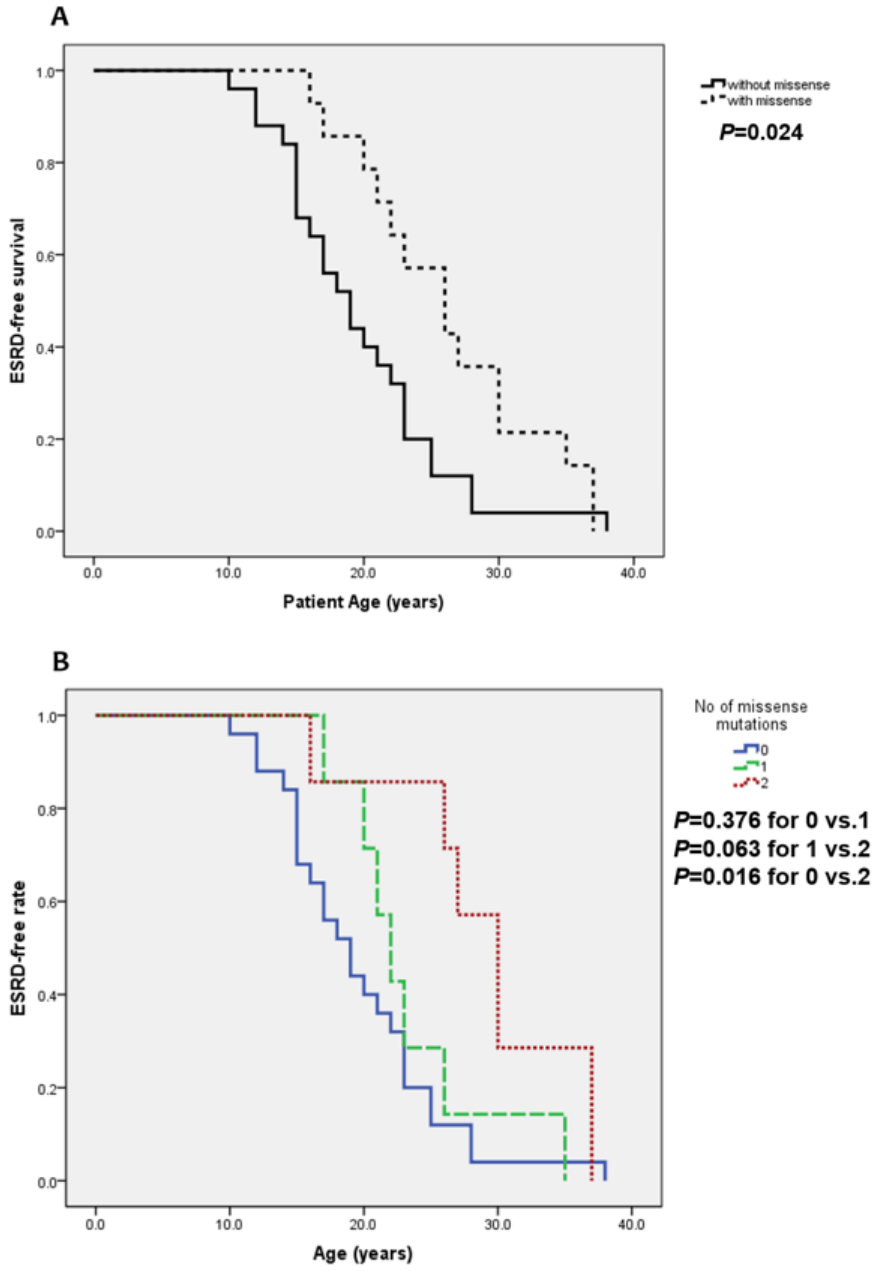

Figure 2. ESRD-free survival of ARAS patients according to (A) presence of missense mutation and (B) number of missense mutations.