Alport syndrome is a genetic disorder of basement membranes caused by mutations in type IV collagen network. It was first identified by Dr. Alport in 1927. Its major clinical manifestations are included: glomerulopathy, sensory hearing loss, anterior lenticonus, and the prevalence of Alport’s gene in general population is about 1 in 5000 and the disease prevalence is 1 in 10000 [1-3]. Here we report ocular, ear and renal manifestations of Alport syndrome in 3 families that living in the east city of Iran, Birjand, and we have followed all of this family for 6 years so far.

In the first family, father (48 years) and mother (45 years) are close relatives but they do not have eye disorders. They have 2 daughters and 1 son. The first child is a 23 years old woman who had complained of decreased visual acuity in eyes, photophobia and hearing loss. The urine analysis was shown hematuria with normal sonographic appearance of the kidneys. The second child is a 19 years old woman, single and a homemaker who had complained of decreased visual acuity in both eyes, no hearing loss, and the third child is a 26 years old man, married and farmer who had complained of decreased visual acuity and hearing loss.

Similarly in the second family parents are closely related and healthy, they have two children the first child was a 16 years old woman who has bilateral hearing loss using hearing aid. Within 3 years her renal failure progression leads to dialysis. The second one is an 18 years old woman who had hearing loss and renal manifestations which leaded to renal failure so she has been undergoing dialysis. She has been candidate for kidney transplantation but due to the lack of donor she died at the age 18 years.

Finally the third family also consists of healthy 40 years old mother and 41 years old father without lenticonus, but in his examinations small fleck retinopathy was seen with normal macula. He did not complain of kidney problems but on laboratory tests hematuria was reported. Nephrocalcinosis was reported by sonography and CT-scan. He did not have hearing loss. Parents are close relatives as the other families.

They have 4 children. Their first child was a 19 years old boy complaining of photophobia and hearing loss of both ears. Laboratory tests showed hematuria. Anterior lenticous in both eyes that leads to cataract surgery in both eyes. The second child was an 18 years old boy who has hearing loss and hematuria in urine analysis. The third child was a 10 years old boy whose eyes are normal; hematuria was reported in urine analysis. The last child was a 3 years old girl who has hearing loss, no hematuria.

Notwithstanding the reports based on rarity of vision disorders associated with Alport syndrome, our patients had retinal lesions and anterior lenticonus, despite the existence of retinal lesion the main reason for decreased visual acuity was lens involvement, removal of the lens and inserting the intraocular lenses is the only treatment of these patients.

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References