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Sir,
Alport syndrome with phenotypic marfanoid habitus: atypical case series

Classically, Alport syndrome is described as a hereditary, multisystem progressive disorder affecting visual, auditory, and renal systems. The disease was initially described by Alport (1927) and then by Arnott.[1,2] In Alport syndrome, posterior lenticonus can coexist along with anterior lenticonus. The reported incidence of this coexistence is around (2.6%).[3] The presence of posterior lenticonus along with anterior lenticonus can occur either unilaterally or bilaterally.

Case reports

In this case series, we report the bilateral occurrence of anterior lenticonus and posterior lenticonus in five cases. In addition to the presence of the rare ocular finding, all our cases have marfanoid habitus.[4] This concurrence adds to the rarity of the present clinical series.

The clinical details of the cases have been summarized in Tables 1 and 2.

Discussion

Ocular manifestations in Alport syndrome are variable and incidence ranges from 11 to 92%. The coexistence of

Table 1 Clinical data of patients that led to clinical diagnosis of Alport syndrome

Case no.	*Age (years)*	*Sex*	*Ocular findings*	*Auditory features*	*Renal disease*[a]	*Visual acuity*		*Electron microscopy*[b]
						RE	*LE*	
1	19	M	ALC + PLC	SNHL (higher frequencies)	Present	6/36, N12	6/36, N12	Hereditary nephritis
2	25	F	ALC + PLC, posterior subcapsular cataract (BE)	Moderate SNHL	Present, renal transplant	6/18, N8	6/12, N8	NIL
3	15	M	ALC + PLC	Mixed hearing loss	Present	6/12p, N36	6/18, N18	NIL
4	33	F	ALC + PLC, posterior subcapsular cataract (BE)	Moderate SNHL	Present, deceased sibling (renal failure)	6/18p, N8	6/12, N10	Hereditary nephritis
5	17	M	ALC + PLC	Mixed hearing loss	Present	6/12, N8	6/12, N8	NIL

Abbreviations: ALC, anterior lenticonus; BE, both eyes; N, near vision; PLC, posterior lenticonus; SNHL, sensorineural hearing loss.
All patients underwent cardiac evaluation/echocardiography and were found to be normal.
Cases 2 and 4 underwent uneventful phacoemulsification with good post-operative visual recovery. Capsulorhexis was started at midperiphery and hydrodissection was avoided.
In case 1, fundus was found to be normal clinically. ERG (electroretinogram) was suggestive of cone rod dystrophy.
[a] Presence of proteinuria, hematuria, and RBC casts in urine.
[b] Renal biopsy samples of cases 1 and 4 (Figure 1 e and f) showed thickening and thinning of capillary loops suggestive of hereditary nephritis consistent with Alport syndrome.

Table 2 Clinical data suggestive of marfanoid habitus while ruling out Marfan syndrome

Features	*Case 1*	*Case 2*	*Case 3*	*Case 4*	*Case 5*
Height	176 cm	181 cm	179 cm	172 cm	175 cm
Hand length	20 cm	21 cm	22 cm	21 cm	21 cm
Foot length	27 cm	30 cm	28 cm	27 cm	28 cm
Arm span	184 cm	187 cm	185 cm	180 cm	184 cm
US: LS	0.84	0.86	0.84	0.81	0.84
High arched palate	Present	Present	Present	Present	Present
Jaw deformity	Absent	Absent	Present (overcrowded teeth, mild retrognathia)	Absent	Absent
Sternal deformties	Absent	Absent	Absent	Absent	Absent
Wrist sign	Present	Present	Present	Present	Present
Thumb sign	Present	Absent	Absent	Present	Present
Scoliosis	<200	<200	Absent	Absent	<200
Ectopia lentis	Absent	Absent	Absent	Absent	Absent
Axial myopia	Absent	Absent	Absent	Absent	Absent
Keratometric reading (flattest meridian)	>41 D	>41 D	>41 D	>41 D	>41 D
Pulmonary/cardiac features	None	None	None	None	None
Cutaneous features	None	None	None	None	None

Skeletal features, cardiac features, pulmonary features, ocular features, and cutaneous features—none corresponding to Ghent criteria[6] for diagnosis of Marfan syndrome. None of the features correspond to 'revised Ghent nosology for the Marfan syndrome'.[7]

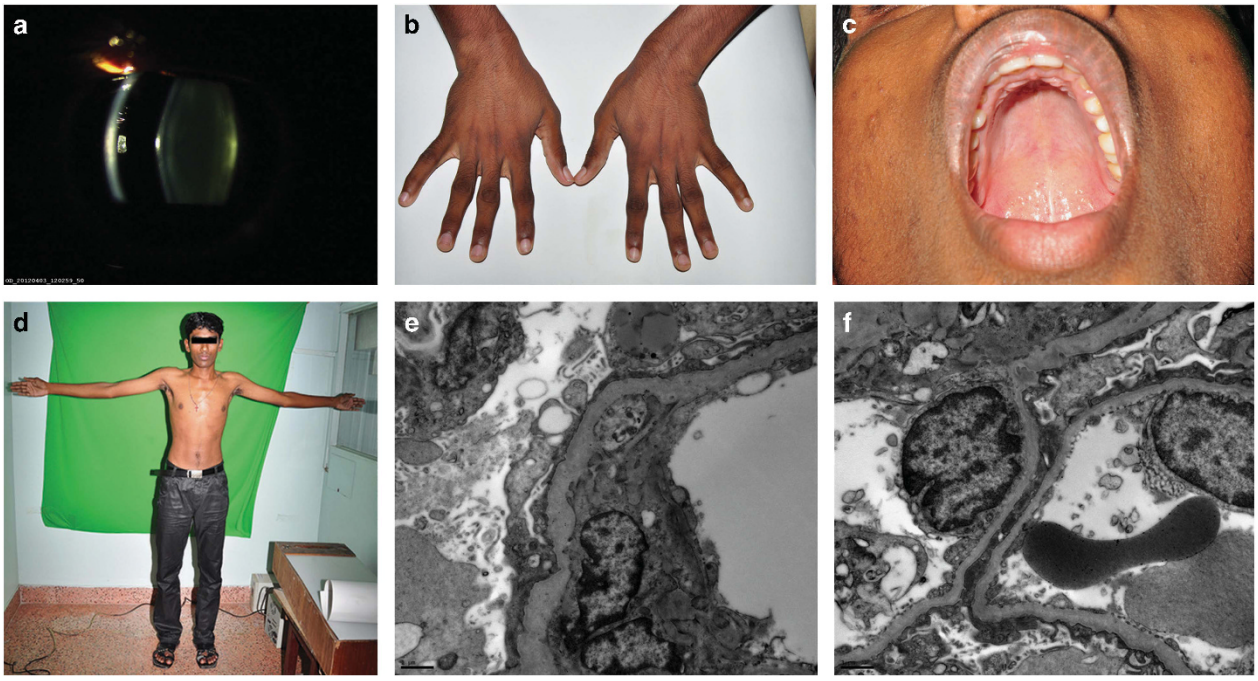

Figure 1 (a) Anterior and posterior lenticonus; (b) arachnodactyly; (c) high arched palate; (d) arm span >height. (e and f) Electron microscopic picture showing thickening and thinning of capillary loops suggestive of hereditary nephritis consistent with Alports (courtesy: Patrick D Walker, M.D, NephroPath, Little Rock, AR 72211, USA).

anterior and posterior lenticonus in the same eye is rare.[3] However, in our case series all the patients had bilateral simultaneous anterior and posterior lenticonus (Figure 1a).

Out of our five cases, three had cataract. Two underwent uneventful phacoemulsification with implantation of acrylic intraocular lens with good post-operative outcome. In all our cases, there was a considerable overlap of marfanoid features[4] (Table 2, Figure 1b–d). Reports of Marfan syndrome having lenticonus as an ocular feature are described.[5] However, no reports of Alport syndrome having phenotypic marfanoid features could be found. The overlap of marfanoid phenotype in Alport syndrome is apparently being reported for the first time.

Our case series highlights that the diagnosis of Alport syndrome is commonly done through ophthalmic portal. Safe and effective phacoemulsification with good visual outcomes is possible in cases with anterior and posterior lenticonus secondary to Alport syndrome. As there is no specific phenotypic description of Alport syndrome in the literature, less attention is paid to the physical features. In our case series we found marfanoid habitus in all cases. So, we recommend a thorough physical examination to look for subtle phenotypic features that can be associated with Alport syndrome. Although genetic analysis is warranted to establish the genotype–phenotype correlation, it was not pursued due to feasibility issues.

Conflict of interest

The authors declare no conflict of interest.

Disclaimer

No author has a financial or proprietary interest in any case report mentioned.

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