Figure 2 (A) Anterior segment OCT image (at low resolution; coronal section across the dashed line in Figure 1) of left eye showing posterior corneal wound (CW), nasal iris tissue defect with peripheral anterior synechiae (PS) and abnormal angle (A). (B) Anterior segment OCT image at high resolution showing posterior corneal wound edge (CW), abnormal angle (A), zonular dehiscence (ZD), lens subluxation (LS) and vitreous in pupillary area (V).

peripheral anterior synechiae in the adjacent angle (fig 2A). In addition, ASOCT showed zonular dehiscence with vitreous prolapse and mild lens subluxation localised to the injury site (fig 2B). At 7-weeks follow-up (dexamethasone stopped 3 weeks earlier), his uncorrected vision improved to 6/6 with an IOP of 40 mmHg. Oral acetazolamide and topical antiglaucoma medications were prescribed to lower IOP. Residual vitreous in anterior chamber was now visible clinically using conventional methods.

Comment

ASOCT is a high-resolution, non-contact method of anterior segment imaging that uses 1310-nm light.¹ It shows the anatomical relationships of cornea, iridocorneal angle and other anterior segment structures in real time.² It enables measurement of anterior segment structures such as corneal thickness, anterior chamber depth and anterior chamber angle. In addition to preoperative and post-operative assessment, ASOCT can be used intraoperatively which can help in the intraoperative diagnosis and subsequent surgical decision making.³

In this case, ASOCT showed angle damage, zonular dehiscence and residual vitreous prolapse, and suggested residual vitreous in anterior chamber as the cause of raised IOP. Due to the presence of corneal oedema, these findings could not be determined during the early postoperative period using conventional examination methods such as slit-lamp examination and gonioscopy. Although high-frequency ultrasound biomicroscopy could have revealed such clinical information, it would be unsuitable in a case with perforating eye injury, due to the immersion method required.

Anterior segment OCT is a novel imaging modality, which is valuable in the evaluation of eye injuries and especially useful in the management of these cases where a non-invasive method of anterior segment assessment is required.

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Figure 1 Fundus photograph of the right eye showing exudative retinal detachment involving the macula.

eye revealed severe submacular lipid exudation with an inferior exudative retinal detachment involving the macula. The left eye had a waxy disc, some midperipheral subretinal flecks, a characteristic macular sheen and no visible subretinal exudation. Fundus fluorescein angiogram showed massive capillary dropout and mid-peripheral neovascularisation in the right eye. The left eye only showed hyperfluorescence from subretinal flecks. There was no angiographic evidence of retinal ischemia in this eye. Based on this, a clinical diagnosis of exudative retinopathy in the right eye was made. Management was difficult in view of the poor visual acuity in the right eye with such a dramatic exudative retinal detachment. After discussing treatment options with the parents, they opted for retinal cryotherapy to prevent the development of a painful, rubeotic eye.

The patient was the second child of first cousin parents. She had a cousin and two siblings similarly affected. They had a similar phenotype with cardiac failure in infancy, early deafness, retinal dystrophy and diabetes mellitus. Molecular analysis confirmed linkage to the Alström locus on chromosome 2p13. Mutation analysis identified a 1444 nucleotide deletion (IVS8 + 895 del 1444) encompassing the whole of exon 9 of the *ALMS1* gene. This is a novel mutation for Alström syndrome, identified in this single family and has recently been reported by our institution.⁵

Comment

Alström syndrome is a rare autosomal recessive disorder caused by mutations in the novel gene *ALMS1*² which has been shown to be widely expressed and localised to centrosomes and the base of cilia.^{3,4}

A review of ocular features in Alström Syndrome reported poor visual acuity in the order of 20/200 or worse by 10 years.⁶ Unlike other retinal dystrophies, this condition can result in vision as poor as perception of light by the end of the second decade. However exudative retinopathy has not been documented in any of the above 22 cases in this series.

Nonetheless, exudative retinopathy has been demonstrated uncommonly in longstanding retinitis pigmentosa. Mutations in the crumbs homologue 1 (*CRB1*) gene are strongly associated with the development of Coats-like exudative vasculopathy in these patients.⁷ This entity differs from classic Coats disease in terms of relatively higher incidence of bilateral involvement in exudative retinopathy, equal gender incidence, mode of inheritance and location of retinal pathology.¹ We are reporting a unique mutation associated with

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Exudative retinopathy in a girl with alström syndrome due to a novel mutation

Coats disease is an uncommon telangiectatic retinal exudation that is most often seen in males. Similar changes are an uncommon but well known complication of retinitis pigmentosa, occurring in up to 3.6% of the affected individuals.¹ This association has not been reported with Alström syndrome. Alström syndrome is a rare autosomal recessive disorder caused by mutations in a novel gene of unknown function, *ALMS1*,² widely expressed in centrosomes and the base of cilia.^{3,4} We report an Asian girl with bilateral congenital cone-rod dystrophy due to Alström Syndrome who developed subretinal exudation resembling Coats disease.

Case report

A twelve year old girl of ethnic Pakistani origin presented with marked deterioration of vision in her right eye over the previous year. She had attended our institution with cone-rod retinal dystrophy (ERG demonstrated absent photopic response & markedly reduced rod response), infantile-onset obesity, cardiomyopathy, sensorineural deafness, diabetes mellitus with hyperinsulinaemia, hepatosplenomegaly and recurrent urinary tract infections for several years. There was no history of polydactyly. On this basis, a clinical diagnosis of Alström syndrome was made.

On examination, there was no demonstrable vision in the right eye and only bare perception of light in the left. Slit-lamp examination revealed quiet anterior chambers without evidence of rubeosis in either eye. A right exotropia on penlight examination with manifest horizontal pendular nystagmus was noted. Fundal examination (fig 1) of the right

a novel phenotype in our patient in the Alström locus. As this is a single case, we cannot be certain that the retinal exudation is definitely caused by this mutation.

Our patient however had unilateral exudative retinopathy. This may be due to an asymmetry of the retinal dystrophy between the eyes. The natural history of this retinopathy is unknown and the beneficial results of treatment varying from photocoagulation,⁸ cryotherapy⁹ and pars plana vitrectomy¹⁰ are difficult to quantify.

It is possible that in time, the other eye may become affected, probably as the retinal dystrophic changes progress.

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Inverse relationship between age and severity and sequelae of acute corneal hydrops associated with keratoconus

Acute hydrops occurs in approximately 3% of eyes with keratoconus.¹ The incidence increases dramatically in eyes with associated vernal keratoconjunctivitis.^{2–3} Neovascularisation rarely develops after resolution of acute hydrops although it has been reported with large area of involvement with hydrops associated with either close proximity to the limbal vascular arcades⁴ or intrastromal clefts.⁵ The association between patient age and area of involvement of hydrops, with the subsequent risk of development of neovascularisation, has not been adequately addressed in a prospective clinical trial.

In Saudi Arabia, patients present with severe keratoconus at a much younger age than in Western populations and have a higher incidence of associated atopic eye disease.^{2–3} The average age at the time of penetrating keratoplasty in our patient population is 19 years, with nearly one quarter of all cases performed in children 15 years of age or younger.³ Approximately one fifth of cases have severe vernal keratoconjunctivitis or seasonal allergic conjunctivitis, 30% have a history of hydrops

prior to corneal transplantation,³ and one quarter of the cases have penetrating keratoplasty at 5 years of age or younger.³ In this study we evaluate the influence of early age on the severity and sequelae of acute hydrops.

Methods

After obtaining approval of the Institutional Review Board of the King Khaled Eye Specialist Hospital, all patients with keratoconus who presented to the emergency room with acute hydrops between February 1, 2004 and July 31, 2004 were enrolled in a prospective, observational clinical trial. A comprehensive ocular examination was performed at the time of presentation and after resolution of the hydrops. The location of acute hydrops was identified and measured using slit lamp examination of the hydrops and its relation to the distance from the limbus. Also, the presence or absence of vascularisation was noted at the time of hydrops. All patients were treated with a tapering regimen of topical fluoromethalone 0.1% and lubrication.

Results

Thirteen patients (eight males, five females) were enrolled in the study (table 1). The median age was 20 years (range, 11–36). Complete resolution of hydrops occurred after a mean of 10.2 weeks (range, 6–13). Seasonal or vernal keratoconjunctivitis was present in nine (69.2%) eyes. There was an inverse correlation between age and area of involvement of corneal hydrops and the likelihood of developing neovascularisation after resolution (table 1, fig 1). Based on the Fischer exact test, patients 16 years of age or younger (5 out of 13) were significantly more likely to have an area of involvement of more than 90 mm² (p=0.03), to have involvement less than 1.0 mm of the limbal vascular arcades (p=0.02), and to develop neovascularisation after resolution of the hydrops (p=0.03) than older patients. Proximity of the hydrops of less than 1.0 mm from the vascular arcades was significantly correlated with the risk of development of neovascularisation (p=0.03). Although atopic disease was present in all 3 eyes that developed neovascularisation, there was no significant correlation between the presence of atopic disease and the likelihood of development of neovascularisation.

Table 1 Case Summaries

Case	Age, gender	Hydrops size Max., Min. (mm)	Area (mm ²)	Limbal Vascular arcade proximity (mm)	Duration of hydrops resolution (weeks)	Vascularisation after resolution	VKC/SAC
1	11, F	10, 9	90	0.2	13	Yes	Yes
2	12, M	8, 8	64	1.0	11	No	Yes
3	13, M	10, 9	90	0.2	12	No	Yes
4	14, F	10, 10	100	0.1	12	Yes	Yes
5	16, M	8, 8	64	0.1	12	Yes	Yes
6	19, F	6, 5	30	1.5	8	No	Yes
7	20, M	7, 7	49	1.5	9	No	No
8	22, M	9, 7	63	0.2	9	No	Yes
9	23, M	7, 7	49	1.7	6	No	No
10	24, M	8, 8	64	1.0	11	No	Yes
11	31, F	5, 5	25	2.0	10	No	No
12	33, F	7, 6	42	1.5	9	No	Yes
13	36, M	6, 4	24	1.3	11	No	No

VKC, vernal keratoconjunctivitis; SAC, seasonal allergic conjunctivitis; Max, maximum; Min, minimum