Blepharochalasis with multiple system involvement

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SUMMARY A rare case of bilateral blepharochalasis of the upper eyelids is presented in a 10-year-old boy with several systemic abnormalities—unilateral agenesis of the left kidney, multiple skeletal anomalies of the vertebral column, and congenital heart disease with a left to right shunt. Such widespread congenital defects in association with blepharochalasis have not been described before. Blepharochalasis may represent a part of a more generalised disorder.

Blepharochalasis is a rare condition, typically affecting the upper lids bilaterally, characterised by atrophy and relaxation of the lid tissues, following intermittent lid oedema.¹ The nomenclature of blepharochalasis was coined by Fuchs in 1896. After recurrent episodes of painless oedema the eyelid skin becomes baggy, wrinkled, sometimes papery thin, and laced with an increased number of tortuous blood vessels.

The condition usually occurs in young people, especially at puberty,^{2,3} and may occasionally be transmitted hereditarily as an autosomal dominant characteristic, both sexes being equally affected.⁴ It should be differentiated from dermochalasis, which usually occurs in older people without previous attacks of oedema.^{1,5}

Most reported cases of blepharochalasis have been limited to the eyelids, and no generalised abnormality has been recorded except in the Laffer-Ascher syndrome.^{1,6} We report here an unusual case with associated renal, skeletal, and cardiac abnormalities. To the best of our knowledge such congenital anomalies have not been described before in association with blepharochalasis.

Case report

A 10-year-old boy presented with a bilateral acquired ptosis following recurrent attacks of eyelid oedema over the past three years, each attack lasting between one and three days. He had a history of fever with burning micturition four years ago, for which he had been investigated elsewhere. There was no known history of any allergy and no significant family history.

On examination he had a mild bilateral ptosis, with

the thin wrinkled baggy upper eyelid skin and tortuous vessels typical of blepharochalasis (Figs. 1, 2, and 3). The vertical palpebral apertures were 8 mm in the primary position in both eyes, and the levator function was good (13 mm) on both sides. The unaided visual acuity was 6/6 J1 both eyes. No other ocular abnormality was detected.

On general physical examination the patient was found to be well built and nourished (body weight 35 kg), slightly obese, with a puffy face. There was no swelling of the lips, no thyroid enlargement, and no evidence of oedema elsewhere in the body.

On cardiovascular examination the first heart sound was heard normally, and the second heart sound was widely split but mobile. An ejection systolic murmur (grade 2/6) was detected, best heard at the lower sternal border, with no click or snap audible. Otherwise, the cardiovascular examination was within normal limits. The other systemic examinations were unremarkable.

Investigations done previously and, after admission to hospital, by us showed normal blood count, urine analysis, blood urea, and serum proteins. The karyotype was normal.

Chest x-rays revealed multiple congenital anomalies of the upper dorsal vertebrae, such as spina bifida, hemivertebra, and scoliosis to the right (Fig. 4).

The size of the heart was normal, even on barium swallow examination, but mild pulmonary plethora was present (Fig. 4). Electrocardiography results were within normal limits. Taken in conjunction with the clinical findings, these were indicative of congenital heart disease with a left to right shunt, with a possibility of partial anomalous pulmonary venous connection. The patient is awaiting cardiac catheterisation for determination of the exact site of the shunt.

Fig. 1 Mild bilateral ptosis (in primary position) with fold of skin overhanging the upper lid margins.

Fig. 2 Thin wrinkled baggy upper eyelid skin (on looking down) laced with an increased number of tortuous blood vessels.

Fig. 3 Good levator action evident (on looking up).

Abdominal x-rays disclosed the presence of an extra lumbar vertebra, with features of congenital fusion of two vertebrae in the lumbar region (Fig. 5).

Intravenous pyelography revealed a non-functioning left kidney, with compensatory hypertrophy of

the right kidney (Fig. 6). Serial scanning renograms with ^{131}I -hippuran confirmed the absence of the left kidney (Figs. 7, 8, 9). The urinary bladder was normal (Fig. 9).

Surgical correction of both upper eyelids was

Fig. 4 Chest x-ray showing mild pulmonary plethora with multiple congenital anomalies of upper dorsal vertebrae (spina bifida, hemivertebra, and scoliosis to the right).

Fig. 5 Abdominal x-ray showing presence of an extra lumbar vertebra, with congenital fusion of two lumbar vertebrae.

Fig. 6 Intravenous pyelogram showing a non-functioning left kidney, with compensatory hypertrophy of right kidney.

undertaken in the same sitting under general anaesthesia. After the dimensions of excess tissue had been determined, the redundant horizontal skin spindles, wider temporally than nasally, were symmetrically mapped out on both upper lids, and carefully excised without disturbance of the deeper structures. The lower edge of the spindle was placed at the level of the lid fold of the upper lid. During surgery no unusual laxity of the orbital septum or fat herniation was observed. After haemostasis had been secured the skin edges were coapted, without any undermining, by interrupted 6-0 atraumatic silk sutures. Postoperatively the vertical palpebral aperture was 9 mm with a levator action of 15 mm on both sides. Histopathology showed a moderate infiltration of the dermis by mononuclear cells.

Discussion

This case illustrates many of the known characteristics of blepharochalasis. Features occasionally described, such as involvement of the lower eyelids,¹⁷ herniation of fat with weakening of the orbital septum, a pseudoepicanthic fold, and atrophy of the nasal fat pad,⁶ were not, however, observed in our case. There was no evidence of hereditary transmission.⁴

Three broad stages in the clinical course of blepharochalasis have been described: a stage of intumescence or oedema, followed by the second stage of atonic ptosis, and, with the further relaxation of the orbital septum, the third stage of ptosis adiposa or fat hernia.¹ Our case was possibly in the early second stage.

The excision of redundant skin in blepharochalasis leads to cosmetic improvement.^{1,5} The orbital septum did not have to be tackled in our case; in young patients resection of fat is rarely required.⁵ However, ptosis caused by a disinsertion of the levator aponeurosis and its surgical repair have been described.⁸ Since the attacks of oedema may continue postoperatively, surgery may be delayed until after a prolonged quiescent period in the natural course of the disease.⁷ A longer follow-up in our case may indicate the need for further surgery.

The aetiology of blepharochalasis is by and large unknown, though various hypotheses have been postulated from time to time. The episodes of oedema usually begin around puberty,³ which might suggest an endocrine imbalance; non-toxic enlargement of the thyroid occurs occasionally.⁴ Our patient showed no clinical evidence of dysthyroid status. The attacks of oedema often resemble angioneurotic

Fig. 7 Scanning renogram with ¹³¹I-hippuran immediately after injection: no activity detected on left side.

Fig. 8 Scanning renogram with ¹³¹I-hippuran 7 minutes after injection: no evidence of activity on left side.

Fig. 9 Scanning renogram with ¹³¹I-hippuran 13 minutes after injection: a normal urinary bladder scan.

oedema of the eyelids,⁹ which might be because of an allergic diathesis, but definite proof of this is not yet available.¹⁰ The recurrent attacks of upper lid oedema in this patient could possibly be of renal origin, although renal oedema characteristically involves the lower eyelids. Mononuclear infiltration of the dermis in our case is consistent with the known histopathological features of blepharochalasis,¹ and may be suggestive of a chronic low-grade inflammatory process.

Although most cases of blepharochalasis are limited to the eyelids, it could well be a part of a more generalised disorder, as is evident in this patient. He was found to have a wide variety of systemic abnormalities associated with blepharochalasis—unilateral agenesis of the left kidney, multiple skeletal anomalies of the vertebral column, and congenital heart disease with a left to right shunt. Although the triad of blepharochalasis, swelling of the mucous membrane of the lips (double lip), and non-toxic thyroid enlargement has been known since 1922 as the Laffer-Ascher syndrome,¹⁶ generalised congenital abnormalities as in our case have not been described before.

Developmental defects of the kidney are known to occur hereditarily or sporadically, and are frequently associated with anomalies in the other organ systems, as skeletal and cardiac in our case, and in the nervous,

genital, and gastrointestinal systems.¹¹ However, their association with blepharochalasis does not seem to have been reported previously.

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