Visual development in the blepharophimosis syndrome

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Abstract

One hundred and one cases of the blepharophimosis syndrome presenting over a decade are reviewed with particular attention to the factors influencing their visual development. Three distinct clinical patterns emerge – severe bilateral ptosis, moderate bilateral ptosis, and asymmetric ptosis – and their differing incidence of amblyopia and strabismus is discussed. The risk of amblyopia is much higher than previously believed (56-4% in our series) and preventive management is discussed.

The blepharophimosis syndrome is characterised by a narrowed horizontal palpebral aperture, ptosis, telecanthus, and epicanthus inversus. We have reviewed 101 cases presenting over 10 years with a minimum two-year follow-up to assess whether the often severe degree of ptosis and the epicanthic folds predispose to the development of amblyopia and strabismus.

Materials and methods

One hundred and one patients who presented between 1978 and 1988 for an oculoplastic opinion on their blepharophimosis syndrome were reviewed. The minimum follow-up time was two years. The ocular examination included measurement of the horizontal and vertical palpebral aperture, levator function, telecanthus, severity of epicanthic folds, refraction, and results of orthoptic examination including the presence of amblyopia, palpebral movements and alignment, state of binocular vision by the usual methods including the 4 dioptrre and 15 dioptrre prism, and stereo tests.

All the patients received a full ophthalmic and orthoptic assessment at each visit. The frequency of the visits depended on the age of the patients and the results of the orthoptic examination.

All patients of preschool age had cycloplegic refraction with spectacle correction if necessary. Visual acuity was assessed according to age and co-operation.

Any child found to have amblyopia possibly attributable to the ptosis was treated as a surgical emergency with a brow suspension procedure. This was accompanied by occlusion and spectacle correction, if appropriate, as with any case of amblyopia.

Results

All 101 patients were diagnosed as suffering from the blepharophimosis syndrome at or around birth. Forty four were female and 57 were male. Thirty one had a family history of the condition, with a 5:1 predominance on the father's side.

Twenty seven cases presented for oculoplastic opinion by the age of 18 months and a further 25 cases before the age of 5 years. Therefore over half our cases presented before starting school.

Three distinct patterns of ptosis emerged. The patients were therefore grouped for analysis into those with severe bilateral ptosis, those with moderate bilateral ptosis, and those with asymmetric ptosis. Each group was then assessed in terms of amblyopia, refractive error, and telecanthus.

Thirty four patients had severe bilateral ptosis with palpebral apertures of less than 4 mm (Fig 1). Over half these patients (19 cases) had bilateral amblyopia, as defined by a visual acuity of 6/12 or less, which could not be improved with spectacle correction. Of these 19 cases 10 also had strabismus, of whom three had no refractive error, three had equal refractive error, and four were anisometropic. Among these 10 cases there were five patients with telecanthus greater than 35 mm (Fig 2).
Forty patients had moderate bilateral ptosis with palpebral apertures of more than 4 mm (Fig 3). Even in this group 17 were amblyopic and all of these had squints. Ten were anisometropic and seven either had no refractive error (three patients). The remaining 23 patients had normal visual acuity in both eyes and no squint (Fig 4).

Twenty one patients had asymmetric ptosis where the pupil of one eye was intermittently covered during waking hours according to the examiner, the parent, or an independent witness (Fig 5). All were amblyopic in that eye but none had squints (Fig 4).

Twenty one patients had asymmetric ptosis where the pupil of one eye was intermittently covered during waking hours according to the examiner, the parent, or an independent witness (Fig 5). All were amblyopic in that eye but none had squints. A further six patients had asymmetric ptoses but the pupillary axis was clear. These were not amblyopic and had no significant refractive error (Fig 6).

The type of strabismus in approximately three-quarters of cases was convergent, some with an associated vertical element, usually a 'V' phenomenon.

Seventy two cases had moderate telecanthus (29–34 mm) and 29 were severe (35+ mm). Asymmetric telecanthus (2 mm or more) was noted in only three cases, of which only one was amblyopic with a convergent squint. This patient had an asymmetry of 7 mm with dense amblyopia on the more affected side, though the eyelid was not covering the pupil, and there was no significant refractive error.

The strabismic and amblyopic patients underwent regular refraction and patching followed by squint surgery as necessary. Of the 57 amblyopes (two lines or more), 13 failed to improve. Of these patients five presented as adults and already had established amblyopia; four presented between the ages of 6 and 9 years and did not cooperate with patching (three had bilaterally moderate ptosis with hypermetropic astigmatism and one was educationally subnormal with severe bilateral ptosis.). Three failed to improve with occlusion therapy and developed non-central fixation, and one was lost to follow-up for over 18 months but reappeared with an established squint. He had a bilateral moderate ptosis with normal refraction.

Surgical intervention was carried out either to prevent amblyopia or for cosmesis. In cases of bilateral severe ptosis, bilateral brow suspension was performed either with stored fascia lata or Mersilene, or with autogenous fascia lata depending on the length of the patient's thigh. Asymmetric ptosis with pupil cover was treated with brow suspension if the levator function was less than 5 mm and with levator resection if better.

Discussion
Blepharophimosis was first reported in 1841 by von Ammon. The syndrome was subsequently described, and various methods of treatment to improve the cosmetic appearance have been advocated. The reports, however, made no reference to the presence or absence of amblyopia or strabismus. Although Allen mentions these in discussing eyelid surgery, their incidence and...
management are not described. We have therefore examined those cases of blepharophimosis presenting to us for oculoplastic opinion, with a view to determining the incidence of amblyopia and strabismus and the factors that may contribute to their cause.

Thirty one of 101 cases (30-7%) had a family history of blepharophimosis with a 5:1 predominance of inheritance through the father. This corroborates the finding of relative infertility of women with this condition.

Amblyopia was found in a total of 56-4% of cases (57 of 101). 18-8% (19 of 101) had bilateral amblyopia with best corrected visual acuity of 6/12 or less and severe bilateral ptosis. 57-9% of these (11 of 19) had either equal or anisometropic refractive error and the remaining 42-1% had no error. 16-8% (17 of 101) were strabismic amblyopes with moderate bilateral ptosis. 20-8% (21 of 101) had asymmetric ptosis and unilateral amblyopia. This emphasises the risk of development of amblyopia due to ptosis.

Strabismus was found in 26-7% of cases (27 of 101). Of these 51-9% (14 of 27) were anisometropic, 22-2% (6 of 27) had equal refractive error, and the remaining 25-9% (7 of 27) had none, showing that refractive error is only partially responsible for squinting in these patients.

Moderate telecanthus of less than 35 mm was found in 71-3% of cases (72 of 101) and the remainder (29 of 101) had severe telecanthus of 35 mm or more. Asymmetry of the telecanthus of more than 2 mm was unusual (3 of 101). One case had an asymmetry of 7 mm with dense amblyopia and a convergent squint on the affected side but no refractive error and a clear pupillary axis. In the group with severe bilateral ptotic amblyopia (19 of 101) five had severe telecanthus with convergent squint but no refractive error. It is not possible to say whether telecanthus as such contributed to the squint and amblyopia, as these patients also had severe bilateral ptosis. However, we do not consider that the correction of telecanthus is indicated to prevent amblyopia or squat. Indeed telecanthus greater than 35 mm was found in 48-3% (14 of 29) of cases with no squint or amblyopia.

The indications for early ptosis surgery included bisection of the pupillary axis (with or without abnormal head posture), particularly if unilateral or if the child failed to make the effort to lift the lid; the development of intermittent strabismus; objection to occlusion treatment; or if tests show suppression to base out 4 and 15 dioptre prisms. However, these tests failed to identify those at risk of amblyopia in the severe bilateral ptosis group.

The most surprising finding was the high incidence of amblyopia and strabismus in this group. It had been presumed in the past that, since the ptosis was bilateral and approximately equal, these patients were not at risk of amblyopia. The second group with a more moderate degree of bilateral ptosis had a lower incidence of amblyopia and squint (42-5%–17 of 40); but despite the higher incidence of anisometropia in those who developed a squint (58-8%–10 of 17) there was still a significant number of patients (41-2%–7 of 17) who were amblyopic with a squint and no anisometropia. The 21 patients with asymmetric ptosis in whom the pupil of one eye was intermittently covered were all amblyopic, but none had squints. These findings clearly indicate the importance of the ptosis and lid level in amblyopia associated with the blepharophimosis syndrome. We consider therefore that ptosis surgery should be carried out more readily and at an earlier stage than has been previously advocated.

The unoperated cases presenting in adulthood with bilateral ptosis were bilaterally amblyopic, whereas those children who underwent surgery early did better. Of our paediatric cases 74% (20 of 27) underwent surgery prior to reaching their fourth birthday but only three developed strabismus (only one with anisotropia), and two had amblyopia without strabismus. This emphasises the preventive advantage of appropriate early surgery.

We did not find the degree of epicanthic folds significant in relation to squint and amblyopia, but it is difficult to judge results objectively as there is no standard method of measuring them. This is in contrast to telecanthus, which lends itself well to accurate measurements, but the extent did not appear relevant to squint or amblyopia unless the telecanthus was asymmetrical. The moderate cases of epicanthus were treated with a Y-to-V plasticy, whereas the more severe cases were lengthened with a double Z plasticy. At the same time the telecanthus was corrected with a medial canthal tendon shortening or transpalpebral wiring as appropriate.

The correction of the epicanthic folds is probably cosmetic. We found only one case of severe asymmetric telecanthus with epicanthic folds in which there was no other obvious cause for the amblyopia, as there was no ptosis and no refractive error.

CONCLUSIONS

In our series of 101 cases of blepharophimosis syndrome the risk of amblyopia for whatever reason, single or combined, was more than half. The risk was highest if the ptosis was asymmetrical with pupillary axis embarrassment but was also present if the ptosis was bilateral. The effect of severe or asymmetric epicanthic folds seems to have little bearing on the normal development of binocular single vision. The innate desire to develop and maintain binocular single vision is strong but relatively short-lived. In cases of blepharophimosis syndrome we advocate careful regular orthoptic follow-up with early ptosis surgery where necessary.

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