The management of orbital cysts associated with congenital microphthalmos and anophthalmos

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Aims: To study the management of the orbital cysts present in a group of patients with anophthalmos and microphthalmos.

Methods: A retrospective study of 34 patients (40 orbits) treated for orbital cyst associated with microphthalmos and anophthalmos.

Results: The two largest treatment groups comprised 17 orbits (42.5%) where the cyst was removed surgically and 17 orbits (42.5%) where the cyst was retained and conformers were used. The remaining cases comprised two orbits (5%) where the cyst was aspirated initially; two orbits (5%) with large cysts which will need to be excised after further orbital growth; one orbit (2.5%) in which a silicone expander was used initially, and one orbit (2.5%) in which a mildly microphthalmic eye had some vision and was monitored but required no surgery.

Conclusion: In this study 33 out of 34 patients had a good cosmetic result which illustrates that the orbital cyst in microphthalmos or anophthalmos performs a useful role in socket expansion and that the majority of patients with this condition can expect a good cosmetic outcome.

Congenital microphthalmos and anophthalmos are rare conditions with prevalence rates of 1.4–3.5 per 10 000 births for microphthalmos and 0.3–0.6 per 10 000 births for anophthalmos. In one study looking at both anophthalmos and microphthalmos the combined prevalence rate was 1.0 per 10 000 births. In one large study of 29 patients with anophthalmos and 48 with microphthalmos, two patients had bilateral microphthalmos and cyst and one had unilateral microphthalmos and cyst. There is no agreed management of the cyst which may accompany these conditions.

In this retrospective analysis of 34 patients treated for an orbital cyst associated with microphthalmos and anophthalmos we describe the management strategies in our hospital.

METHODS

All patients with either microphthalmos and cyst or anophthalmos and cyst seen at Moorfields Eye Hospital between 1970 and 1998 were included in this study.

The patients were examined by the same surgeon and a consultant paediatrician in a specially designated anophthalmic clinic. Patients underwent imaging, most commonly computed tomograph (CT) or magnetic resonance imaging (MRI).

Once a cyst had been diagnosed, the aim of the subsequent management was to keep the cyst for as long as necessary to ensure adequate socket size. If the socket became too large the cyst was removed immediately. If the socket was growing satisfactorily with the cyst, it was retained until the optimum socket size was reached. If the cyst was not causing problems and a satisfactory artificial eye could be fitted, the cyst was retained. If an artificial eye could not be fitted without removing the cyst, the cyst was removed once an adequate socket size had been achieved. Lid surgery was only carried out at an early stage if a conformer or artificial eye could not be retained. Elective lid surgery was otherwise left until after puberty and the patients were discharged in their mid-teens, when orbital growth was complete, if no other orbital or lid surgery was planned. They continued to have annual prosthetic appointments.

RESULTS

Thirty four patients were treated for orbital cyst associated with anophthalmos or microphthalmos (Table 1). In six of these cases the cysts were bilateral (three cases of anophthalmos and three cases of microphthalmos) giving a total of 40 orbital cysts. Fifteen patients were male and 19 were female. The mean age at presentation was 23 months (range 1–204 months). Of the unilateral orbital cysts 17 were on the left side and 11 were on the right side. Seven patients also had significant systemic abnormalities (Table 1).

The cases fell into six treatment groups: (A) the cyst was aspirated but fluid reaccumulation led to surgical excision; (B) the cyst was excised surgically; (C) conformer therapy only; (D) large orbital cyst preventing conformer use—future cyst excision planned; (E) mildly microphthalmic eye with some vision—no conformer used; (F) in this patient a silicone orbital expander was used initially followed by conformer treatment. The two largest treatment groups were 15 patients (17 orbits) requiring surgical excision of an orbital cyst (group B) and 17 patients treated with conformers only (group C). The orbital cysts were excised at a mean age of 41 months (range 6–128 months). In two cases the orbital cysts were initially aspirated, but owing to reaccumulation of fluid, the cysts required surgical excision. A large orbital cyst was present in two cases, which prevented the use of a conformer. These children are due to have cyst excision when they are older.
Table 1 Analysis of the 34 patients in this study and the presentation and management of their orbital cysts

<table>
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<tr>
<th>Number</th>
<th>Age at presentation (months)</th>
<th>Sex</th>
<th>Diagnosis</th>
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<th>Systemic abnormalities</th>
<th>Ocular abnormalities</th>
<th>Primary treatment</th>
<th>Age at time of primary treatment (months)</th>
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<th>Follow up (months)</th>
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UAC = unilateral anophthalmos with cyst; UMC = unilateral microphthalmos with cyst; BMBC = bilateral microphthalmos with bilateral cysts; MAC = microphthalmos (no cyst) and anophthalmos with cyst; BABC = bilateral anophthalmos with bilateral cysts; BMUC = bilateral microphthalmos with unilateral cyst; BAUC = bilateral anophthalmos with unilateral cyst; A = cyst aspirated but required surgical excision later; B = cyst excised surgically; C = conformer therapy only; D = large cyst preventing conformer fitting - for future excision; E = mildly microphthalmic eye with vision - no conformer used; F = silicone expander used initially, cyst later excised surgically; MM LLF = mucous membrane graft left lower fornix; MM LUF = mucous membrane graft left upper fornix.
A good cosmetic result was achieved in 33 out of 34 patients (see Fig 1). This was based on a subjective assessment by patient, parents, paediatrician and surgeon. One patient was judged to have a fair cosmetic result. No patients had a poor cosmetic result.

**DISCUSSION**

Cysts associated with microphthalmos and anophthalmos may present in a characteristic fashion. However, as they represent two points on the spectrum of colobomatous eye disorders, clinical distinction may be difficult and discernible only upon postmortem examination. Anophthalmos with cyst often presents at birth as a bluish orbital mass which pushes the upper lid forward. The cyst, which varies in size, represents the primary optic vesicle and histologically it may be solid or cystic and a maldeveloped lens may be present. Anophthalmos with cyst is usually associated with a normal eye in the contralateral orbit, although there have been cases associated with microphthalmos and cyst in the fellow orbit, such as cases 3, 18, and 25 in this study.

Duke Elder described three categories of cyst associated with microphthalmos: a relatively normal eye with a small cyst, not apparent clinically; an obvious cyst associated with a grossly deformed eye; a large cyst which has pushed the globe backwards so that it is not visible clinically. The cyst usually presents as a bluish bulge in the lower eyelid and is firm on palpation. If the cyst is visible through the conjunctiva it transilluminates readily. There have been a few reports where the cyst has presented as a bulge at the upper eyelid. Most cases are diagnosed at birth although late presentation is possible.

Both microphthalmos and anophthalmos can be associated with systemic abnormalities. We found systemic abnormalities associated with microphthalmos more probably no more common in cases with bilateral cysts compared to cases with unilateral cysts.

The diagnosis of orbital cyst associated with microphthalmos and anophthalmos can be made clinically in some cases, such as a moderately microphthalmic eye associated with a large orbital cyst pressing on the lower lid. However, in many cases some doubt often remains as to the nature of the orbital lesion and whether there is a microphthalmic eye or ocular remnant present hidden behind a large cyst. In these cases additional imaging has been valuable. Orbital ultrasound (both A and B modes) have been used to identify orbital cysts. More recently both CT and MRI scanning have proved invaluable for diagnosis.

Following the diagnosis of orbital cyst associated with anophthalmos and microphthalmos a review by a paediatrician is mandatory to search for any other associated systemic abnormalities. The classification proposed by Warburg is useful for identifying syndromes associated with microphthalmos and cyst.

Genetic disorders responsible for microphthalmos and cyst include autosomal dominant, autosomal recessive, and X linked syndromes. Once the condition has been categorised, the family should be given genetic counselling.

Consideration should next be given to the visual potential in cases of microphthalmos. Elder found that microphthalmic eyes with a corneal diameter of 5 mm or less at birth were associated with a visual acuity of no perception of light in 81% cases. If doubt persists as to the level of vision then electrodagnostic tests should be considered. Eyes with some residual vision can be treated with clear conformers which do not obscure the visual axis.

Once the visual potential of the microphthalmic eye has been assessed, further treatment depends upon the age of the patient at presentation and the volume of the orbital contents (microphthalmic eye plus orbital cyst or just orbital cyst in cases of anophthalmia).

Patients can be categorised into two groups by age at presentation. These are birth to 5 years and 5 years or older. Within these groups the orbital volume is assessed clinically using the Duke-Elder classification: (A) a relatively normal eye with a small cyst; (B) an obvious cyst associated with a grossly deformed eye; (C) a large cyst which has pushed the globe backward so that it is not visible clinically.

It has been noted already that patients with poor orbital volume can achieve very good cosmetic results if treated with conformers as early as possible. Several clinical and experimental studies have shown that enucleation in childhood compromises orbital growth. The earlier the enucleation, the greater the reduction in orbital bone growth. Although orbital bone growth is complete, no orbital implant is used. This has resulted in the recommendation that elective enucleation should be delayed until orbital bone growth is complete.

In cases of microphthalmos and anophthalmos with a noticeable cyst, parents are often keen to have the cyst removed to improve cosmesis. It is then necessary to discuss with the parents the timing of the cyst removal. The relevant considerations are that the orbit is not fully grown until puberty and that any small or medium sized cyst (with an associated ocular remnant if present) will help to stimulate orbital expansion, much more effectively than an artificial implant. However, an equally important consideration is that of cosmesis, which becomes more relevant as the child starts school. A reasonable compromise is to consider orbital cyst removal around the age of 5 years. Although not fully developed, the orbital growth for both males and females at this stage is about 90% of that of an adult. Thus, some orbital expansion may be sacrificed for cosmesis as the child begins schooling. In cases of very large orbital cysts, prolapsing through the palpebral fissure, they may cause bony expansion as well as being cosmetically unacceptable to parents. For this reason they are often excised at an early age.

At Moorfields Eye Hospital, we follow a policy of aggressive socket expansion, with conformers, at first presentation of anophthalmos or severe microphthalmos without a sizeable cyst. Ideally, this should start as soon after birth as possible. At the first visit to the prosthodontist, it may be difficult to fit a rigid conformer owing to the very shallow conjunctival fornices. In these cases the use of a HEMMA expander is very helpful. This hydrophilic material swells within the socket as it absorbs moisture and gently stretches the fornices, which enables a larger conformer to be inserted after 24–48 hours. The availability of the HEMMA conformer has most recently been necessary to suture conformers into the socket.

With new patients (particularly those with poor orbital volume) follow up is on a weekly basis for the first few months as, with aggressive treatment with increasingly larger conformers, there should be a steady progression through the range of conformers. As the socket size increases, so the standard conformers fit less readily. At this stage a mould of the socket can be taken using Orthoprint, under general anaesthetic and a custom made prosthesis fitted.

When the decision to remove the orbital cyst has been made, the method of removal must be considered. The easiest and most effective way is to aspirate fluid from within the cyst cavity. In cases of microphthalmos and anophthalmos with a noticeable cyst, parents are often keen to have the cyst removed to improve cosmesis. It is then necessary to discuss with the parents the timing of the cyst removal. The relevant considerations are that the orbit is not fully grown until puberty and that any small or medium sized cyst (with an associated ocular remnant if present) will help to stimulate orbital expansion, much more effectively than an artificial implant. However, an equally important consideration is that of cosmesis, which becomes more relevant as the child starts school. A reasonable compromise is to consider orbital cyst removal around the age of 5 years. Although not fully developed, the orbital growth for both males and females at this stage is about 90% of that of an adult. Thus, some orbital expansion may be sacrificed for cosmesis as the child begins schooling. In cases of very large orbital cysts, prolapsing through the palpebral fissure, they may cause bony expansion as well as being cosmetically unacceptable to parents. For this reason they are often excised at an early age.

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Within the range of conformers used, the HEMMA conformer is the most versatile, being available in all sizes and being very comfortable to wear. The HEMMA conformer is inserted into the socket and is removed after 2–3 days. The conformer is then replaced with a larger conformer and this process is repeated until the desired size is achieved. Once the desired size is achieved, the HEMMA conformer is removed and a custom made prosthesis is fitted.

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The presence of an orbital cyst with anophthalmos or microphthalmos aids orbital expansion and thus most cases can be treated successfully with aggressive conformer therapy alone. In cases of anophthalmos or microphthalmos with cyst where the orbital volume is poor other orbital volume expansion techniques should be considered if conformer therapy is thought to be inadequate on its own.

Dermis fat grafts have been proposed, as they have the advantage of being autogenous and have an ability to enlarge as the child grows. Fat atrophy with this implant in paediatric patients is rare. Tissue expanders have also been proposed for enlargement in children with poor orbital volume. These comprise a silicone balloon with a reservoir. They are inserted and usually left for 6 months. Periodically, additional saline is injected into the reservoir to expand the balloon.

Craniofacial surgery is required infrequently in patients with microphthalmos or anophthalmos and cyst, as the presence of the cyst prevents severe micro-orbitism. However, a few patients have such contracted sockets that aggressive conformer therapy and orbital volume expansion do not result in satisfactory cosmesis. In these patients good results have been reported using three dimensional orbital bone expansion.

It has been suggested that any secondary surgery to the lids (such as medial and lateral canthoplasty, expansion of the palpebral fissures) or mucous membrane grafting to the socket are best left until maximal expansion has been achieved by conformer therapy. Early treatment may lead to cicatricial tissue formation which may ultimately lead to a poorer cosmetic result. Exceptions to this rule might be those cases where a lower lid coloboma is present which prevents the fitting of a stable conformer, such as in case 21 in this study.

In this study we have outlined the current treatment regimen used at Moorfields Eye Hospital which is based primarily on early and aggressive use of conformers. Grossly enlarged cysts (with propthiasis through the palpebral fissure) were excised soon after presentation and replaced with an orbital implant and ocular prosthesis. Large cysts (with no propthiasis from the palpebral fissure) were left to encourage orbital growth. Between the ages of 3–5 years they were excised soon after presentation and replaced with an enlarged cysts (with proptosis through the palpebral fissure) or mucous membrane grafting to the palpebral fissures or mucous membrane grafting to the palpebral fissures. In all cases regular follow up with frequent changes in conformer size, especially in the first weeks of life, maximised orbital growth and the subsequent cosmetic result. In this study 33 out of 34 patients had a good cosmetic result which illustrates that the orbital cyst in microphthalmos or anophthalmos performs a useful role in socket expansion and that the majority of patients with this condition can expect a good cosmetic outcome.

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