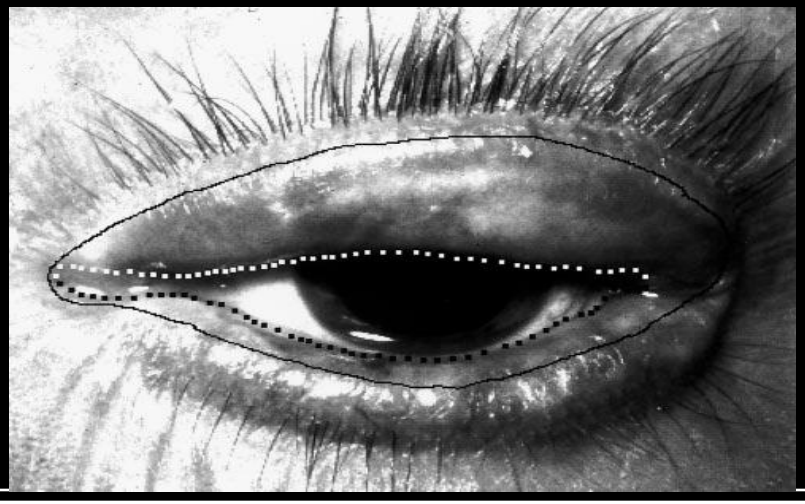
Eyelid Abnormalities in Lamellar Ichthyoses

Supervised by:

Dr.adel ALsuhaibani



Introduction

The ichthyoses are a heterogeneous group of inherited skin disorders characterized by abnormal keratinization or cornification.

Keratinization is the differentiation process that the epidermal cell layer undergoes to form the stratum Corneum.

Cont.

Depending on the association of dermatologic changes with other organ abnormalities, the ichthyoses can be differentiated as isolated or nonsyndromic versus associated or syndromic.

Among the nonsyndromic forms of ichthyosis, the term “lamellar” has been used to describe a broad group of conditions with different clinical presentations.

Cont.

Clinical, biochemical, and histologic evidence exists separating the lamellar ichthyoses into at least two distinct diseases:

(1)classic lamellar ichthyosis (LI)

(2) congenital ichthyosiform erythroderma (CIE).

However,this distinction is controversial, because some authors consider the erythroderma to be just a variable phenotype and do not accept the separation between CIE and LI.

Cont.

The association between ichthyosis and cicatricial ectropion was first reported in 1834. Since then, this eyelid malposition has been the most commonly reported eye abnormality of the LI.

Methods

- They examined 10 patients with LI. Eight patients had classic LI and two had CIE.
- No patient had previous eye or eyelid surgery.
- All patients were using ophthalmic tear lubricants when they were examined.
- Patients were asked to close their eyes gently, and any gap in the palpebral fissure after eyelid closure was diagnosed as cicatricial lagophthalmos.
- The upper and the lower eyelid margins were carefully evaluated with special attention to the presence (yes) or absence (no) of ectropion and eyelash abnormalities.
- Biomicroscopy of the cornea and conjunctiva was performed on all patients.

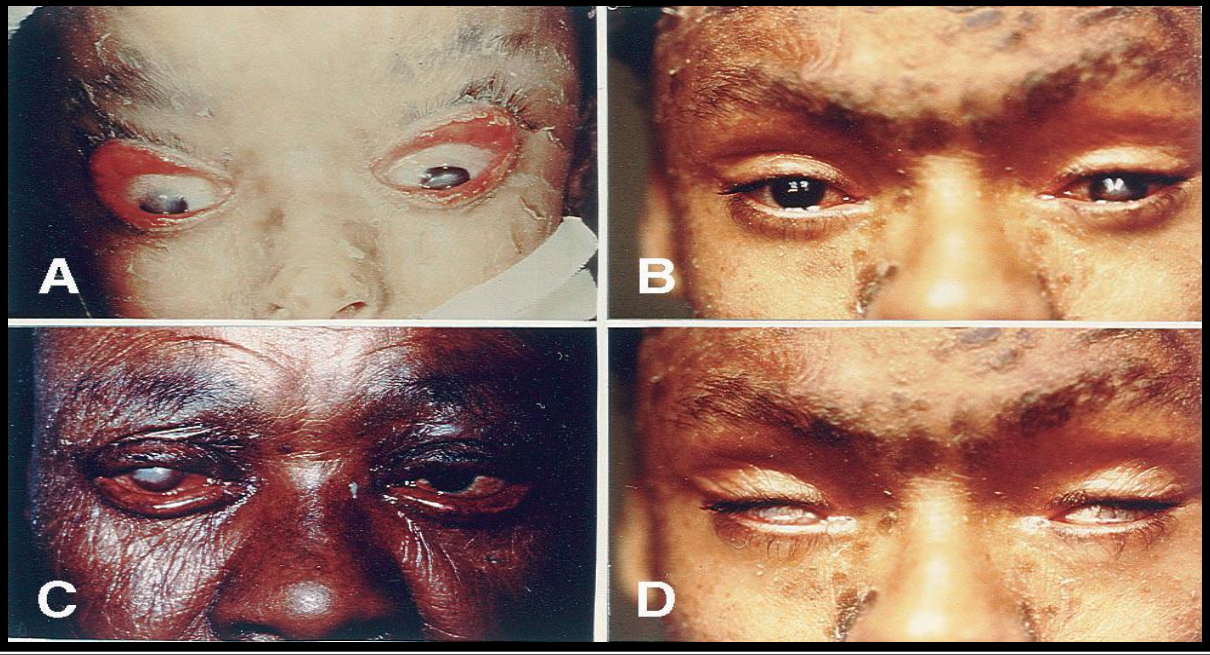
Results

cicatricial lagophthalmos was diagnosed in all patients.

Of the eight patients with LI, three had a history of exposure keratopathy, which led to corneal ulcer, perforation, and severe corneal scarring with loss of useful vision.

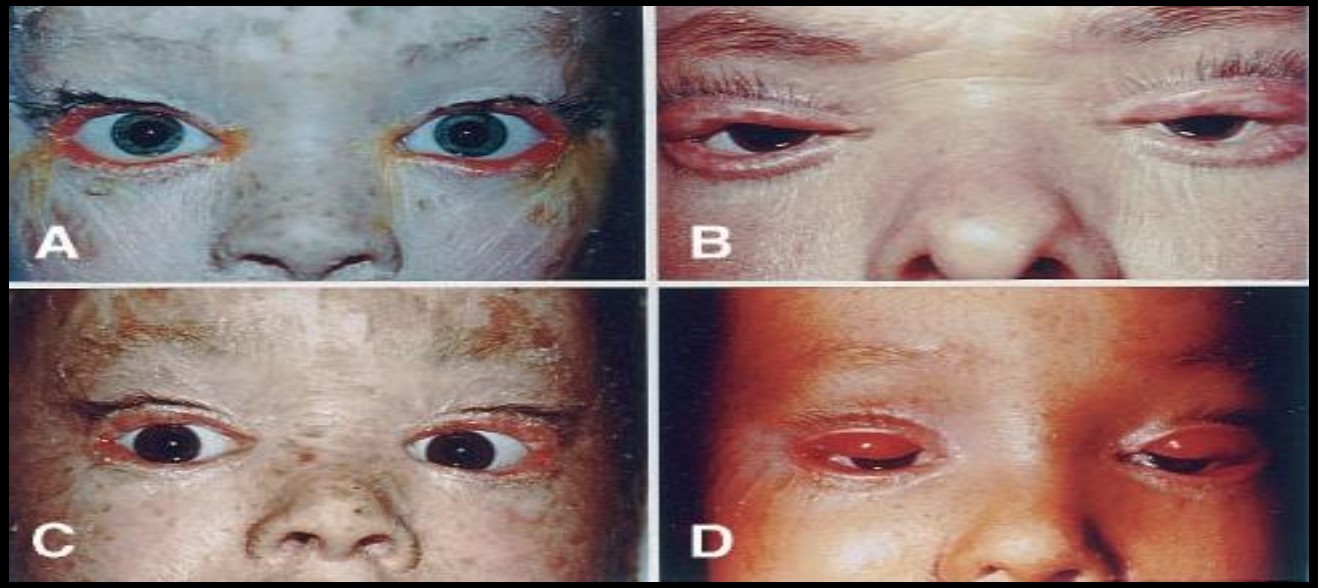
Cont.result

Of these, one had upper eyelid ectropion, one had lower ectropion only, and the last showed no sign of either upper or lower ectropion.



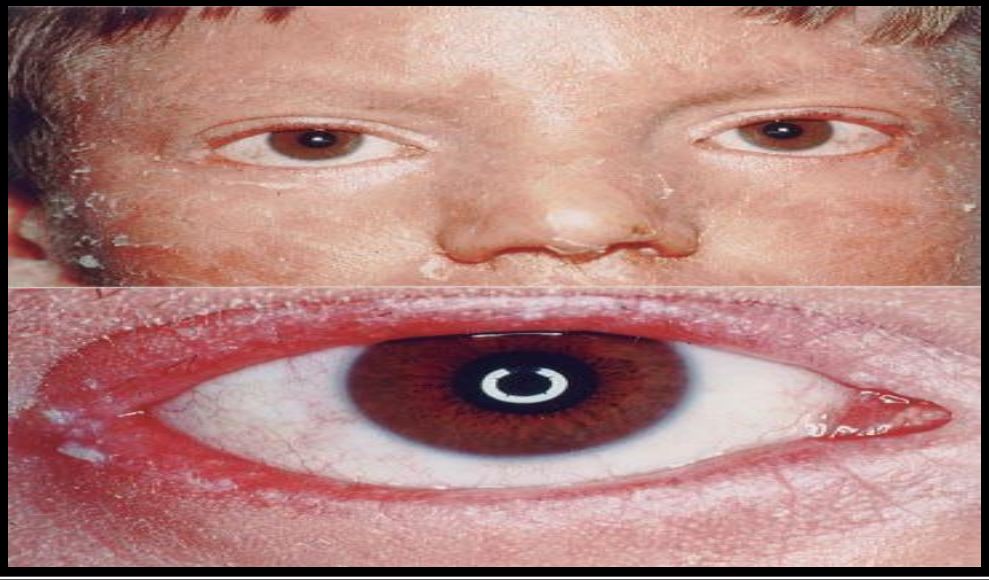
Cont.result

Of the remaining five patients, four had marked upper and lower eyelid ectropion, and one had isolated lagophthalmos. For all these five patients, the eyes were calm without any sign of exposure keratitis.



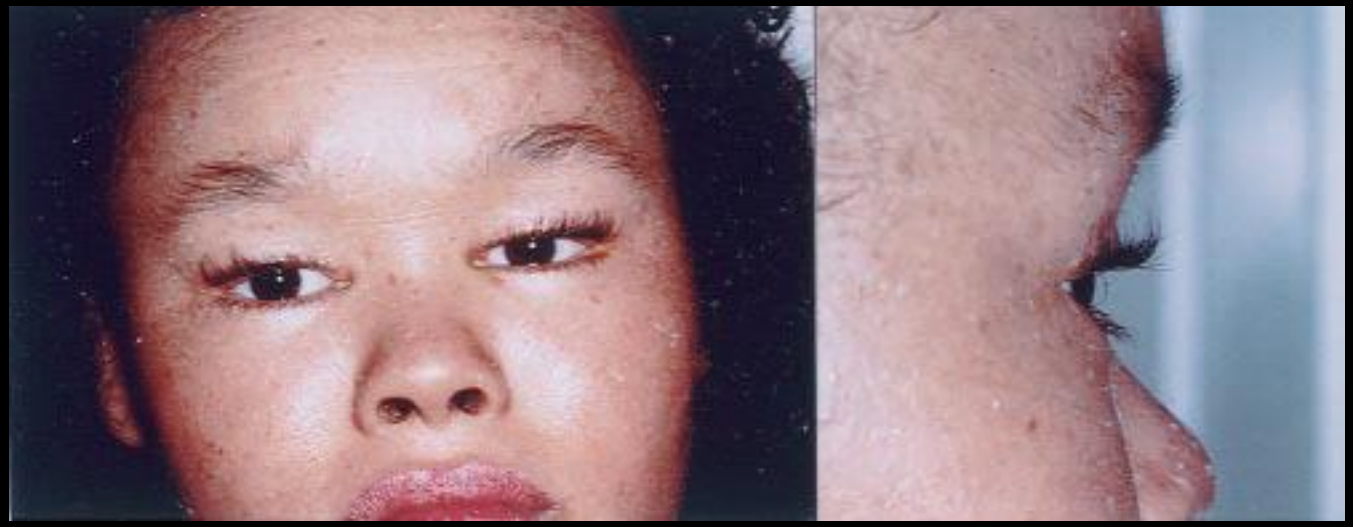
Cont. result

Both CIE patients also showed moderate degrees of lagophthalmos.Of these two patients, one had a small amount of ectropion, blepharitis, and eyelash madarosis of the four eyelids.



Cont.result

The other showed no blepharitis or madarosis, but the upper eyelashes were clearly retracted upwards.



Discussion

- LIs are rare. Both major forms (LI and CIE) are usually transmitted as an autosomal recessive pattern, with an estimated incidence of 1:250,000 to 300,000.

- LI has been linked to different gene mutations. Initially, LI was linked to a specific gene located on chromosome 14q11 that controls transglutaminase 1 expression.

Cont.discussion

However, it soon became evident that some patients with LI had normal transglutaminase activity, and another locus for LI was reported on chromosome 2q33-35.

These studies indicate that LI is genetically heterogeneous, and it is still not possible to find specific genotype-phenotype relationships.

Cont.discussion

LI and CIE are evident at birth. Affected neonates are called“collodion babies,” because they are embedded in an inelastic membrane that dries soon after birth and fissures with a high risk of dehydration and infection. Within 2weeks the collodion membrane spontaneously sheds and the skin develops platelike scales that cover the entire body.

The final clinical picture is quite variable. LI is characterized by the development of large, dark skin scales that cover the entire body, with its typical lifelong disfiguring appearance. In CIE, the skin is red with scales that are white and fine.

Cont.discussion

- In both forms, the most common eyelid abnormality reported is ectropion, which is assumed to be the cause of corneal exposure and ulceration.

Cont.discussion

Usually, when the dynamic properties of the upper eyelid are normal, the role of the lower eyelid in protecting the cornea is not critical.

It is a common experience to find patients who had their lower eyelids removed for tumor resection and lived for years without any sign of corneal decompensation.

In the ichthyoses the situation is distinct,because the upper eyelid movements are never normal. There is always a certain degree of restriction on the upper eyelid (cicatricial lagophthalmos), which can be extreme in the most severe forms of LI.

Cont.discussion

- For these patients, the presence of ectropion of both eyelids increases dramatically the area of the palpebral fissure and certainly augments the rate of tear film evaporation.

- the absence of the Bell's phenomenon is a clear indication that the risk of corneal scarring is high. This combination of factors explains the corneal disease of patients. However, it might seem reasonable to suppose that severe cicatricial lagophthamos induced tear film instability and corneal scarring.

Cont.discussion

In the two CIE patients, eyelid eversion was less marked,

and both patients displayed a significant amount of

upper eyelid movement.

For one patient, the main indication of margin eversion was the eyelash position. Eyelashes were clearly retracted upwards, a finding that is opposed to the lash ptosis found in some conditions in which the pretarsal anterior lamella is loose, such as in floppy eyelid syndrome and leprosy.

The other patient had a mild ectropion and severe blepharitis that provoked madarosis in all eyelids.

Conclusion

- this study shows that the need for surgical treatment in LIs must be individualized. Patients with small fissures without upper or lower ectropion and normal Bell's phenomenon are good candidates for medical management only.
- The rationale for surgical treatment is to reduce the palpebral fissure area by correcting significant amounts of upper and lower eyelid ectropion. If the fissure area is not increased, surgery may still be considered, depending on the relation between the amount of lagophthalmos and the magnitude of the Bell's phenomenon.

Conclusion

- In any case, it will always be necessary to lengthen the anterior lamella with free skin grafts. Because of the widespread involvement of the skin, the grafts will contract, and lagophthalmos is likely to recur.

- Because the eyelid abnormalities in LI and CIE are lifelong, careful serial ophthalmic examination for corneal exposure is warranted.

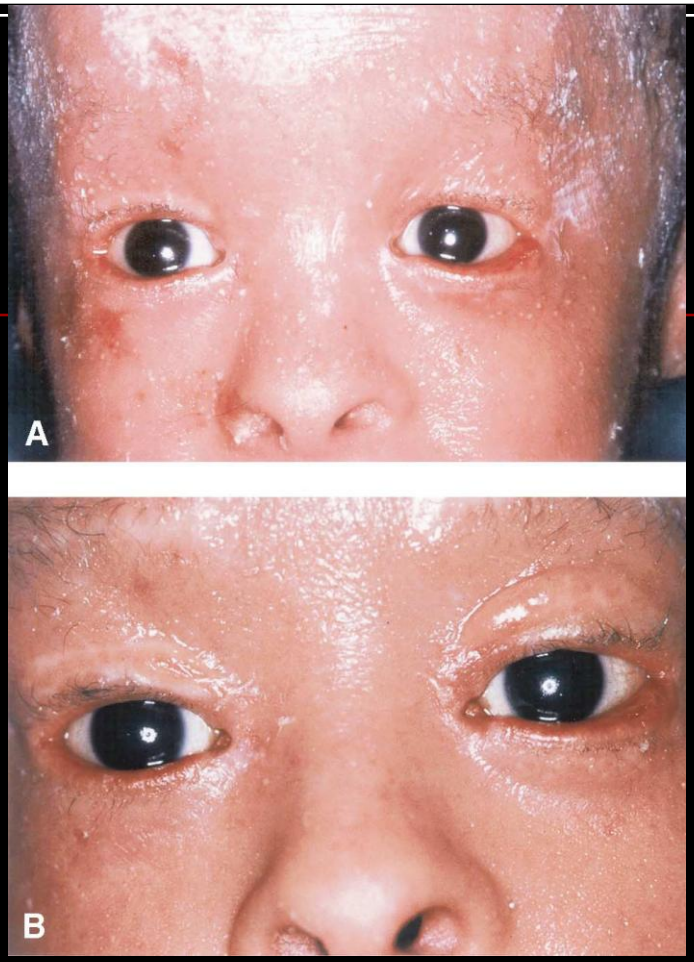
Transforming Mucous Membrane Grafts into Skin Grafts

The use of mucous membrane graft in ichthyosis is really very interesting, because it is not always easy to obtain skin in these patients. This problem has already been recognized,and some authors have even suggested that.

It was said that “mucous membrane grafts do not appear to desquamate or contract as much as skin grafts.

Transforming Mucous Membrane Grafts into Skin Grafts

Although extension of the anterior lamella is frequently useful, they generally prefer oral mucous membrane to skin for grafts of 1.5 cm or less of vertical height, avoiding a slow-healing donor site in these patients, because the oral mucosa is unaffected by the disease. Once implanted into the eyelid, mucous membrane grafts quickly keratinize and do not seem to desquamate or contract as much as true skin grafts.



An 8-year-old male with congenital ichthyosis. **A,** Two year postoperative for lower eyelid mucous membrane grafts with some mild recurrence of ectropion, left greater than right. **B,** Two months later after upper eyelid mucous membrane grafts

Thank you