OCULAR MANIFESTATIONS OF EPIDERMOlYSIS BULLOSA

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Eye involvement in inherited Epidermolysis Bullosa was first reported in 1904 - Case report

**Why does it affect the Eye?**
- The conjunctiva, cornea and eyelids are derived from *surface ectoderm*, like skin.
- Biochemical and *ultrastructural similarities* like plasma membrane and the epithelial-connective tissue basement membrane zone.

*Light and Transmission electron microscopy:* **
- Multiple duplications of basement membrane
- Redundant anchoring fibrils

*Pernet G. Involvement of the eyes in a case of epidermolysis bullosa. Ophthalmoscope 1904;2:308-9.*

Risk of ocular manifestations among various subtypes of EB*  

1. EB Simplex  12%  
2. Junctional EB  40%  
3. Dominant Dystrophic EB  4%  
4. Recessive Dystrophic EB  51%  

Most common ocular finding as per NEBR**: corneal vesiculation, blistering and erosions  

Frequency of ocular manifestations usually mirrors relative severity of skin disease  


## Various documented ocular manifestations in EB

<table>
<thead>
<tr>
<th>Affected Structure</th>
<th>Disorder</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Lids</strong></td>
<td>Blisters, Blepharitis, recurrent hordeolum, ankyloblepharon, Ectropion</td>
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<tr>
<td><strong>Conjunctiva</strong></td>
<td>blisters, congestion, chemosis, subconjunctival hemorrhage, symblepharon, pseudopterygium, conjunctival shrinkage</td>
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<tr>
<td><strong>Cornea</strong></td>
<td>blisters, recurrent erosions, ulcerations, opacities, scarring, pannus, limbal broadening, vascularisation, punctate/ exposure keratitis</td>
</tr>
<tr>
<td><strong>Adnexa</strong></td>
<td>loss of eyelashes, lacrimal duct obstruction</td>
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</tbody>
</table>

• 3280 patients
• 16 years of longitudinal follow up
• 8 variables – Corneal erosions/blistering, corneal scarring, symblepharon, blepharitis, ectropion, lacrimal duct obstruction, impaired vision, blindness.

• STUDY PURPOSE:
  - To determine the frequency of ocular manifestations in EB
  - To define estimated cumulative risk of developing non scarring and scarring corneal manifestations within each major EB subtype

NEBR: Eye involvement in EB

- Ectropion: Primarily found in JEB-H
- Lacrimal duct obstruction: Primarily in Inversa subtype of RDEB

Outcomes

- Cumulative risk of corneal scarring is lower than blistering and recurrent erosions
- Life table analyses showed that corneal blistering may arise in EB patients as early as 1 year of age, risk being 18.6% in RDEB and 27.1% in JEB.
- Increased frequency of impaired vision and blindness: RDEB-HS
Management

- Multidisciplinary approach
- Regular short admissions to a specialized centre to minimize disability and help the child achieve his full potential.
- Initial full ophthalmic examination in all EB patients. Then follow up as per need/annually.
- Extra vigilance in screening patients with Recessive Dystrophic and Junctional type of EB.
Management: Spectacles

• Large padded spectacle arms & sponge padding of pressure points of frames to avoid friction blisters.
• Silicon frames: Miraflex
• Alternate methods of amblyopia therapy (other than patching) like penalization using eyedrops
**DRY EYES**

- Artificial tear drops (preservative free)
- Lubricating eye ointments/gels (preservative free)
- Ultrasonic humidifiers

- Preservative free ocular lubricants - as frequent as one hourly.
- Hold eyelids gently to avoid blistering.

RECURRENT CORNEAL EROSIONS

- Soft contact lenses
- Antibiotic ointment
- Amniotic membrane transplant - facilitates re-epithelialization

CORNEAL INFILTRATES

- Corneal scraping- Gram stain and KOH mount
- Frequent instillation of preservative free antibiotic drops and ointment
- Frequent follow up.
- May need corneal transplant if don't respond to medical management.
LIMBAL STEM CELL DEFICIENCY

- Partial or Total
- Slowly progressive conjunctivalization of cornea
- May need excision and limbal stem cell transplant with AMT
PANNUS/ CONJUNCTIVALIZATION

- Excision with amniotic membrane transplant
- Limbal stem cell transplant

Amniotic membrane used as dressing in burn wounds

Advantages
- readily available in sufficient quantities
- simple preparation and sterilization
- no allergic reactions, no immunological problems
- 15% reduction of water loss

- Reduce infection, pain, accelerate wound healing
  - Reinforce basal cellular adhesion
  - Anti inflammatory and modulated stroll scarring
  - Rapid adherence and improved angiogenesis
  - improves graft take up (faster)
CORNEAL SCARS

- Lamellar/penetrating keratoplasty - guarded prognosis in view of poor ocular surface status

Cicatricial ectropion and severe symblepharon sequelae can cause exposure keratitis or lagophthalmos

- Maintain on lubricants for as long as possible
- Tarsorrhaphy/ectropion correction surgery - when ocular discomfort and visual compromise becomes intolerable.
- Symblepharon: Release with conformer placement & steroid ointment use.

Sound general health and nutrition of the child must be ensured before surgery.
Keratoprosthesis

Front Part

Corneal Graft

Back Plate

Locking Ring

Auro Kpro
ADVICE TO PARENTS: LOOK FOR · Pain
· Redness
· Watering/Discharge
· Photophobia (inability to open eyes in bright light)
· Discoloration/loss of transparency of the cornea
· Lagophthalmos & exposure (during waking hours or while asleep)
· Diminution of vision
NARAYANA NETRALAYA

your faith shall heal you...

Thank you