

Management of Keratoconus in an individual with Downs syndrome- A case study in India

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Authors have no Financial Interest



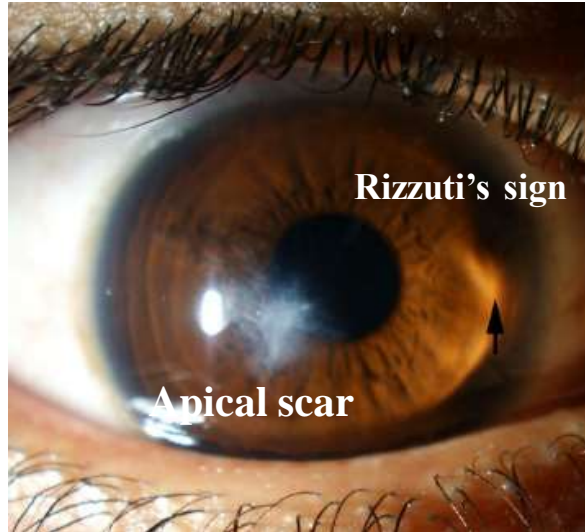
Case study



- ✓ 18 year old female
- ✓ Phenotype: Downs syndrome
- ✓ Genotype: Trisomy 21(non-disjunction)
- ✓ C/o watering in the left eye for the more than a week with bluish discoloration of the cornea

On examination

RE Advanced KC



LE large Acute Hydrops



Visual acuity not recordable in Both eyes

Treatment plan...

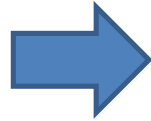
✓ Diagnosis:

Tomography not reliable

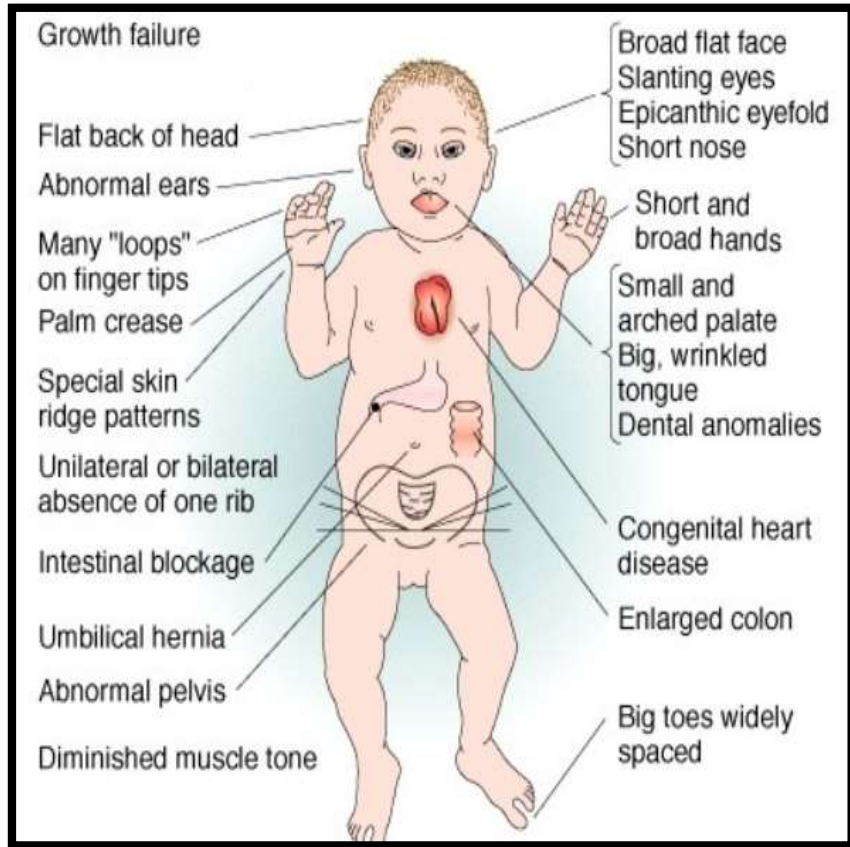
RE Advanced disease: Scleral Contact lens- parents not affordable, no insurance

LE Acute Hydrops: Advanced stage- needed immediate surgical intervention

Final result

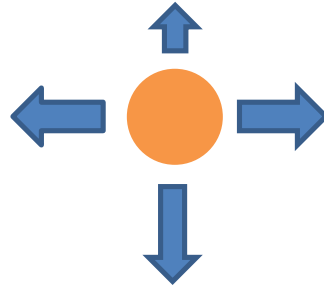
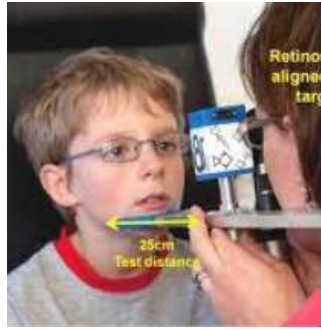


Discussion



OCULAR FINDINGS IN DOWN'S SYNDROME

Oblique fissures
Epicanthal folds
Refractive errors
External hypertelorism
Blepharoconjunctivitis
Brushfield spots
Thinned peripheral iris
Cataracts (become more apparent after age 8)
Decreased visual acuity
Narrowed interpupillary distance
Strabismus
Nystagmus
Keratoconus
Ectropion



KERATOCONUS



Accommodation and Refractive Error in Children with Down Syndrome: Cross-Sectional and Longitudinal Studies

Mary Clegg,^{1,2} J. Margaret Woodhouse,¹ Valerie H. Pakeman,¹ Kathryn J. Saunders,^{1,3} Helen L. Gunter,^{1,4} Margaret Parker,¹ William I. Fraser,³ and Prema Sastry⁵

Keratoconus

- Bilateral, Ectatic corneal disorder
- ***Structural changes:***
 - ✓ Corneal thinning
 - ✓ Biomechanical weakness
 - ✓ Corneal Protrusion
- ***Functional defects:***
 - ✓ Irregular astigmatism
 - ✓ Distorted vision
 - ✓ Deteriorating vision

- Begins in early puberty
- Manifests in early teenage years
- Progression during adulthood
- Associated with Asthma, Eczema, Several systemic diseases
- Affects 1 in 2000 general population
- Diagnosed only by an ophthalmologist through various investigative modalities



KC Incidence

➤ **Incidence of KC in DS – 0% to 71% (Norway)**
 ([JAMA Netw Open.](#) 2021 Mar; 4(3): e210814.)

Author	Location	Age	Sample Size	Prevalence In 100000	Source
Hof Stetter	Indianapolis, USA	001-79	13395	500	Population
Tanabe	Muroran, Japan	010-60	2601	9	Hospital
Jonas	Maharashtra, India	>29	4667	2300	Population
Millodot	Jerusalem, Israel	18-54	981	2340	Population
Ljubic	Skope, Macedonia	-	2254		Population
Ziaei <i>et al.</i>	Yazd, Iran	16-35	536		
Hashemi H	Mashed, Iran	22-29	1079		
Santigo	France	18-22	670		
Xu <i>et al.</i>	Beijing, China	50-93	3166		
Hashemi <i>et al.</i>	Tehran	20-34	1073		
Godefrooij DA	Netherlands	10-40	4million		
Mohammed Othman	Egypt	21-36	2116	170	

The incidence of KC in the general population is relatively low and variable, between 4 of 1000 and 6 of 1000, with the current incidence is 1 of 2000 per year.

The incidence of keratoconus in patients with DS may be between 10 and 300 times more frequent than in individuals without DS

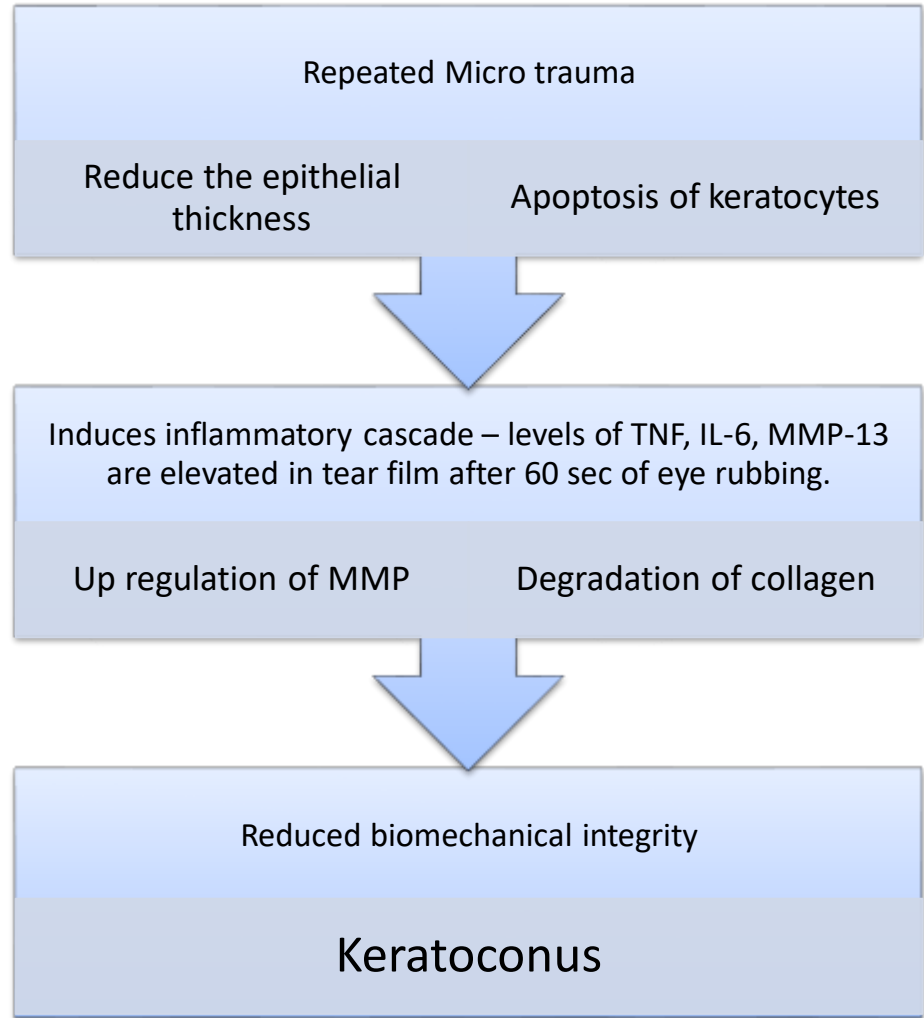
Etiopathogenesis:

EYE rubbing

- Major risk factor for Keratoconus
- 50% have association
- Vigorous and long duration of eye rubbing (10 sec – 180 sec) compared to <5 seconds is associated with KC vs non-KC

Other physiological conditions:

- Pregnancy- hormonal changes oestrogen modulates the corneal biomechanics
- Patients with PCOD, on hormonal therapy for infertility



Existing evidence of genetic association between DS and KC

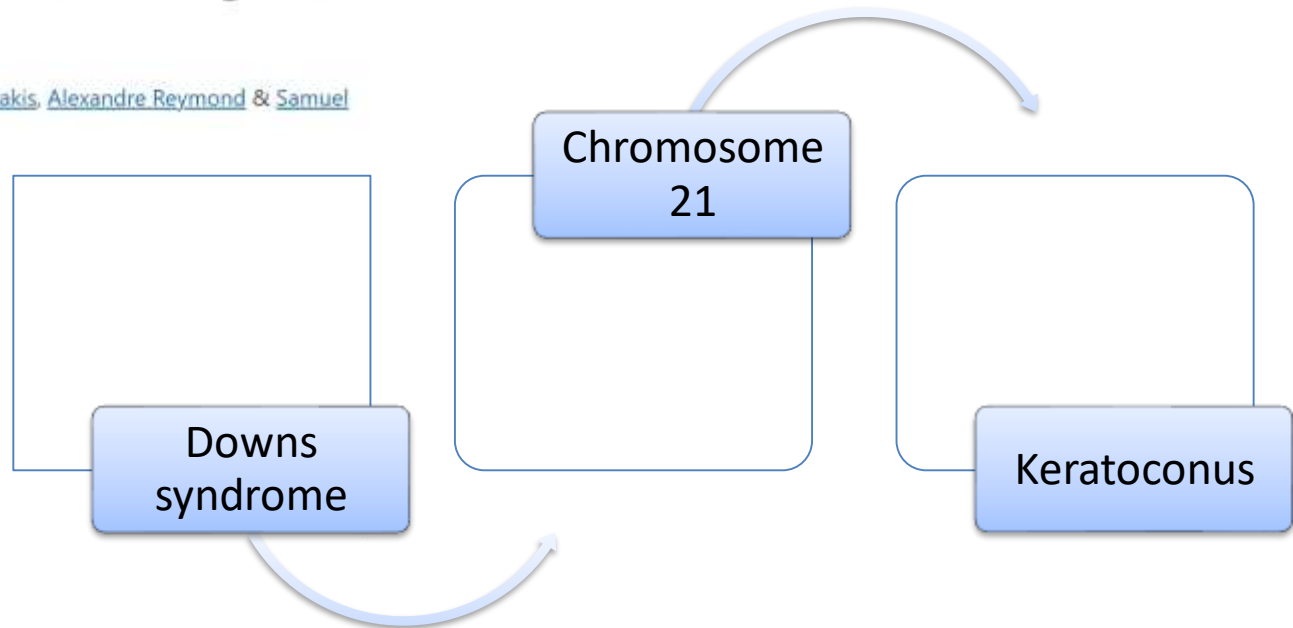
Published: 01 October 2004

Chromosome 21 and Down syndrome: from genomics to pathophysiology

[Stylianou E. Antonarakis](#) , [Robert Lyle](#), [Emmanouil T. Dermitzakis](#), [Alexandre Reymond](#) & [Samuel](#)

[Deutsch](#)

[Nature Reviews Genetics](#) **5**, 725-738 (2004) | [Cite this article](#)



Is there a genetic association between KC and DS?????

The structure of the optical components of the DS eye when compared with the general population,

- Thinner, steeper corneae, thinner crystalline lenses, reduced accommodative function, and variations in tear film composition.
- An increased prevalence of high astigmatism, keratoconus, and cataract.
- These variations in the optical and refractive components of the DS eye may influence optical quality and integrity and help to explain the poor visual performance found in this unique group.

> [Ophthalmic Physiol Opt.](#) 2000 Mar;20(2):79-89.

The prevalence of ocular defects and the provision of eye care in adults with learning disabilities living in the community

J M Woodhouse ¹, C Griffiths, A Gedling

> [Br J Ophthalmol.](#) 1998 Jul;82(7):793-6. doi: 10.1136/bjo.82.7.793.

Emmetropisation, axial length, and corneal topography in teenagers with Down's syndrome

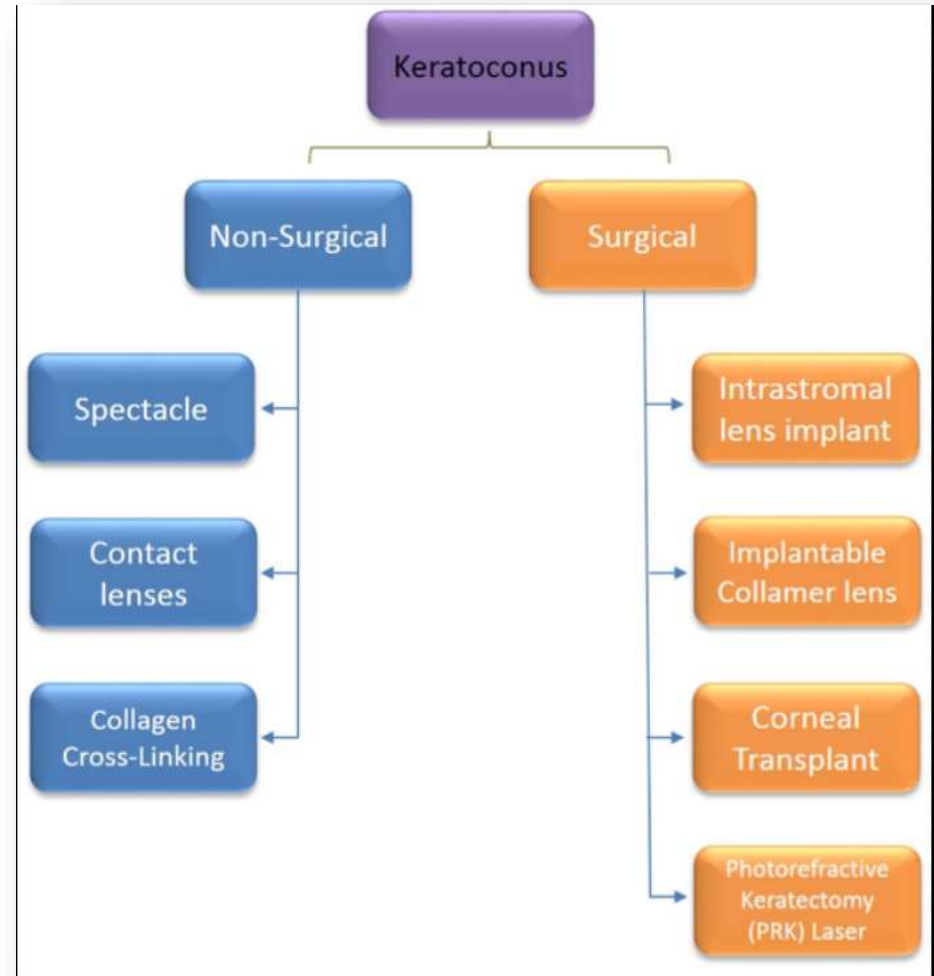
S J Doyle ¹, J Bullock, C Gray, A Spencer, C Cunningham

Treatment


Early stage:
Collagen cross
linkage with
riboflavin

to

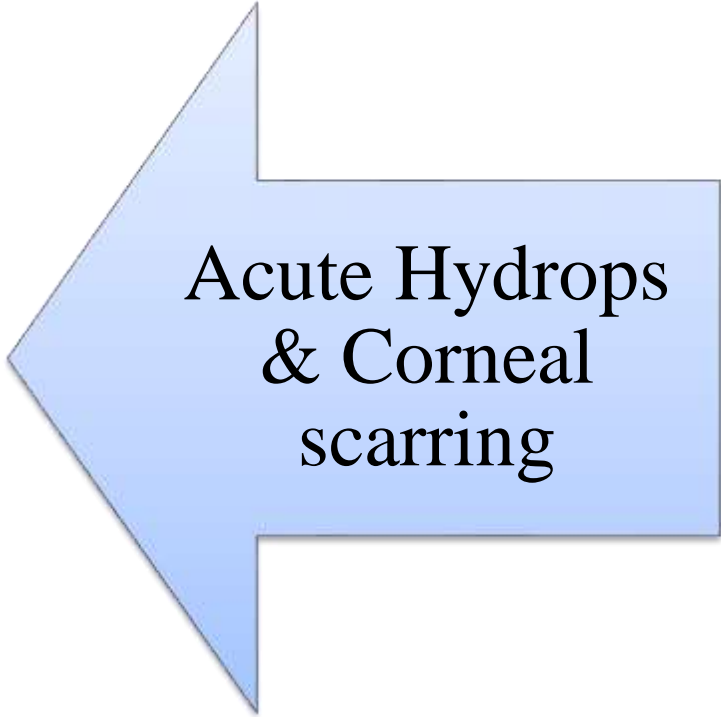
Stop the
progression of
KC



If left untreated?



Progressive
loss of vision
& Amblyopia



Acute Hydrops
& Corneal
scarring

Pathogenesis

Acute corneal hydrops (CH) is the development of marked corneal edema due to a tear in Descemet membrane (DM) followed by leakage of aqueous into stroma.

Intra- stromal inflammation- release of inflammatory mediators

Increased Proteolysis

Decreased protease inhibitors



Stromal lysis & Corneal thinning – Keratoconus progression

Stretching of peripheral DM –
tightly adhered

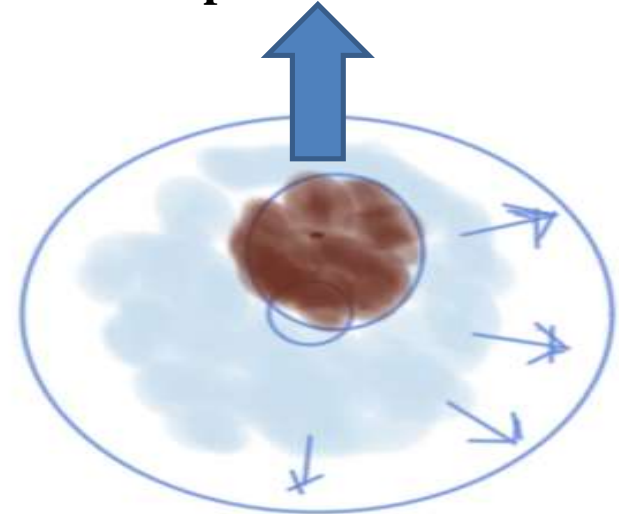
Circumferential stretching of DM



Membrane ruptures

HYDROPS

Rupture of DM



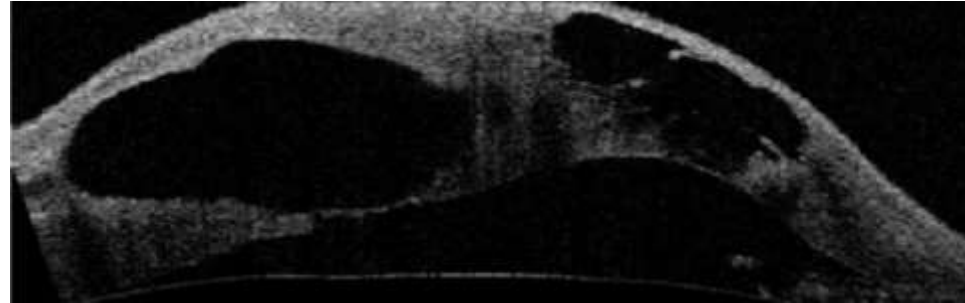
Surgical Management

- Grade 3 Hydrops
- Grade 1 & 2 non resolving hydrops with medical RX

Aim is to hasten resolution of corneal edema and leading to non vascularised scar, thereby increasing prognosis for keratoplasty.

Surgical modalities:

- Compression sutures
- Intracameral gas
- DALK
- Mini- DMEK



Acute corneal hydrops in keratoconus

Prafulla K Maharana, Namrata Sharma, and Rasik B Vajpayee

Indian J Ophthalmol. 2013 Aug; 61(8): 461–464.

Our patient...

RE advanced stage KC

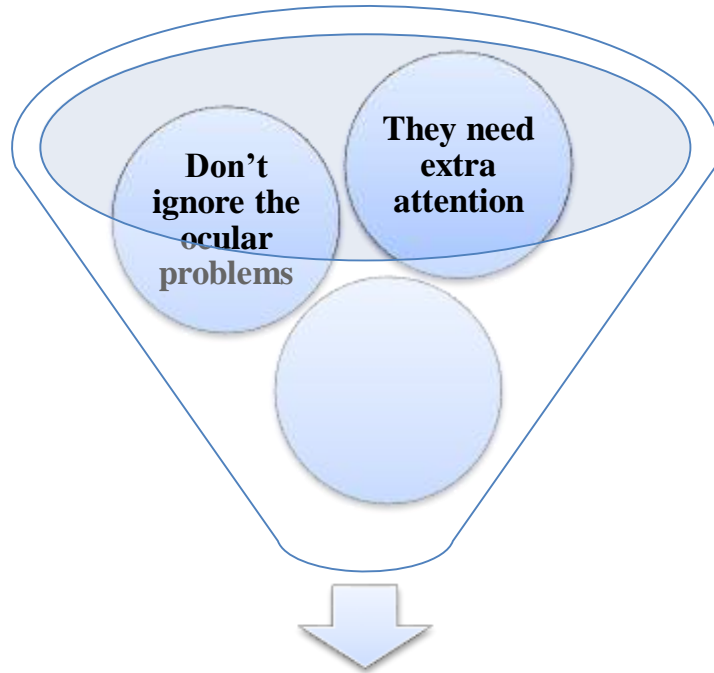


LE central corneal scarring



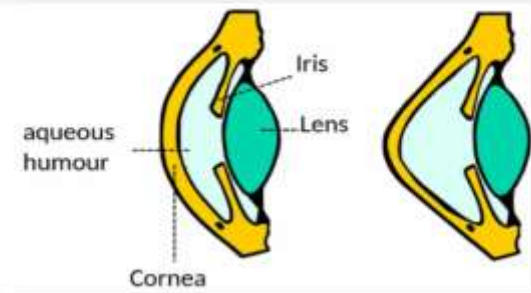
Early Diagnosis of KC through regular screening with Tomography is imperative for stopping the progression of the disease with CXL.

Downs syndrome



Thank you

Mail to dranitha@aravind.org for further information



**DOWN'S SYNDROME
&
KERATOCONUS**