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EDNF 2008 Conference

“Take Charge”

Your Eyes and Ehlers-Danlos Syndrome

A Doctor Who Understands!

- Classic EDS
- POTS
- Autonomic Dysfunction: (POTS, syncope, digestive problems, tachycardia, BP problems, anxiety, difficulty controlling body temperature, lack of sweating, difficulty controlling blood sugar levels, hormone fluctuations, tremor, extreme fatigue)
- “MASS” Phenotype: mitral valve prolapse, aortic root at upper end of normal, striae, skeletal abnormalities
- Possible overlapping phenotype with Marfans
- Bifid uvula – any overlapping phenotype with Loyaes-Dietz Syndrome?

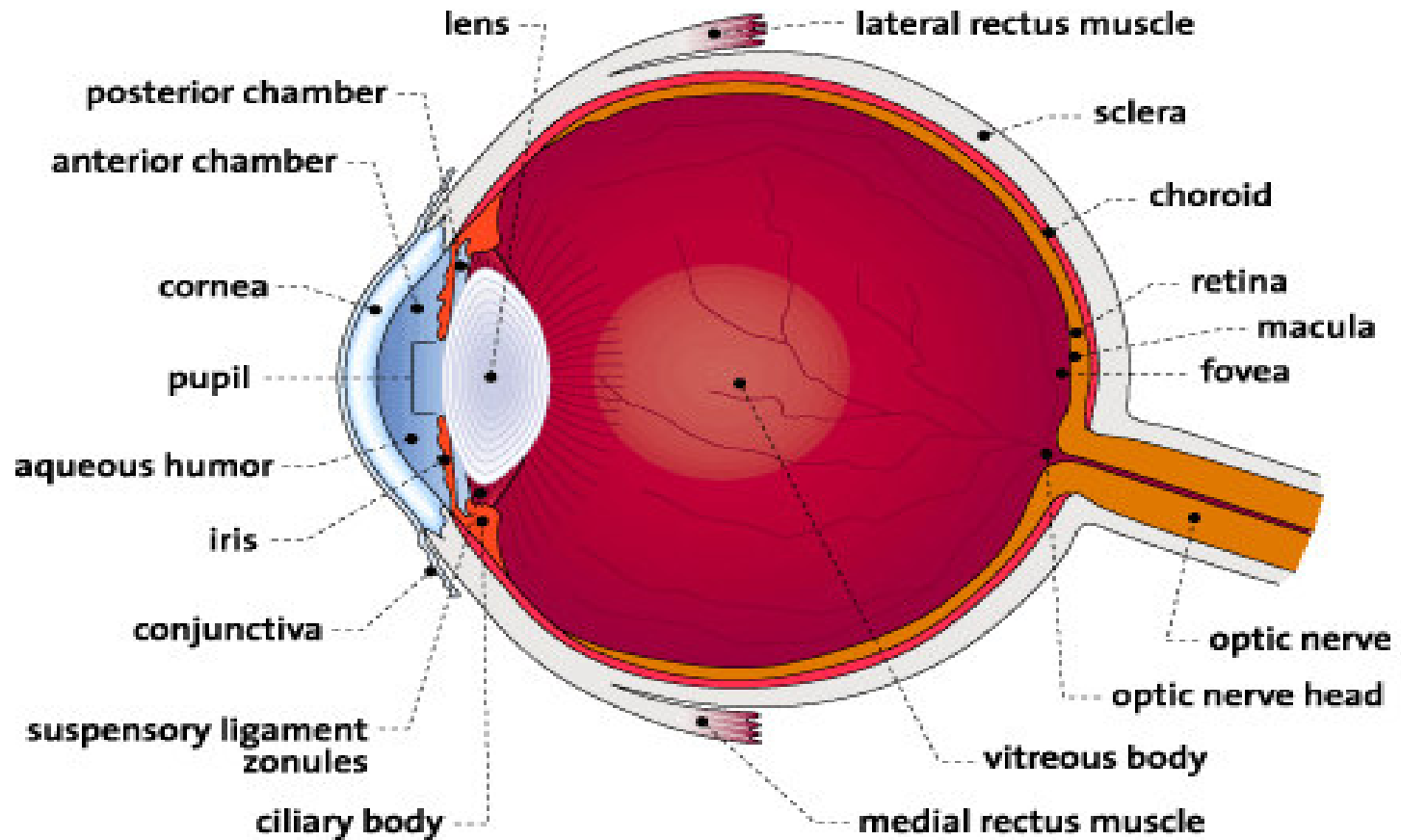
What We'll Discuss Today

- Collagen and your eyes
- Common eye problems with EDS
- Signs/symptoms/treatments: what to watch for, what is urgent, what is not
- Studies: completed
- Studies that need to be done
- How eye doctors can help locate patients who are suffering, but not yet diagnosed.

Doctor as Patient

- Every patient is different, but I will share with you some of the things that have helped me, and have increased my functionality.
- Medications
- Mechanical devices
- Check with your doctor before trying any new medications, even OTC's

Eye Anatomy



There is an amazing amount of collagen in the eye (80% of ocular structures), but relatively, a surprising lack of vision threatening, EDS related effects.

27 different genes are responsible for making the collagen in the structures of the eye.

Type VI- Kyphoscoliosis Type
Lack of Lysyl Hydroxylase
< 60 cases worldwide

Ocular Findings Possible with EDS

- Epicanthal folds
- High myopia
- Keratoconus
- Blue sclera
- Lens subluxation
- Angiod Streaks
- Cataracts
- Dry eyes
- Glaucoma

•More Ocular Findings

- Photophobia
- Retinal Detachments
- Strabismus
- Macular Degeneration
- Posterior Staphyloma
- Carotid-cavernous sinus fistulas

Ocular symptoms that patients complain of that MAY or may not be related to their EDS:

- Blurred vision that comes and goes; difficulty in accommodation
- Seeing double – out of one eye, or with both eyes open.
- Light sensitivity
- Complete, or almost complete, loss of vision in one eye that lasts a few minutes; migraine auras
- Dry eyes
- Tunnel vision
- Floaters

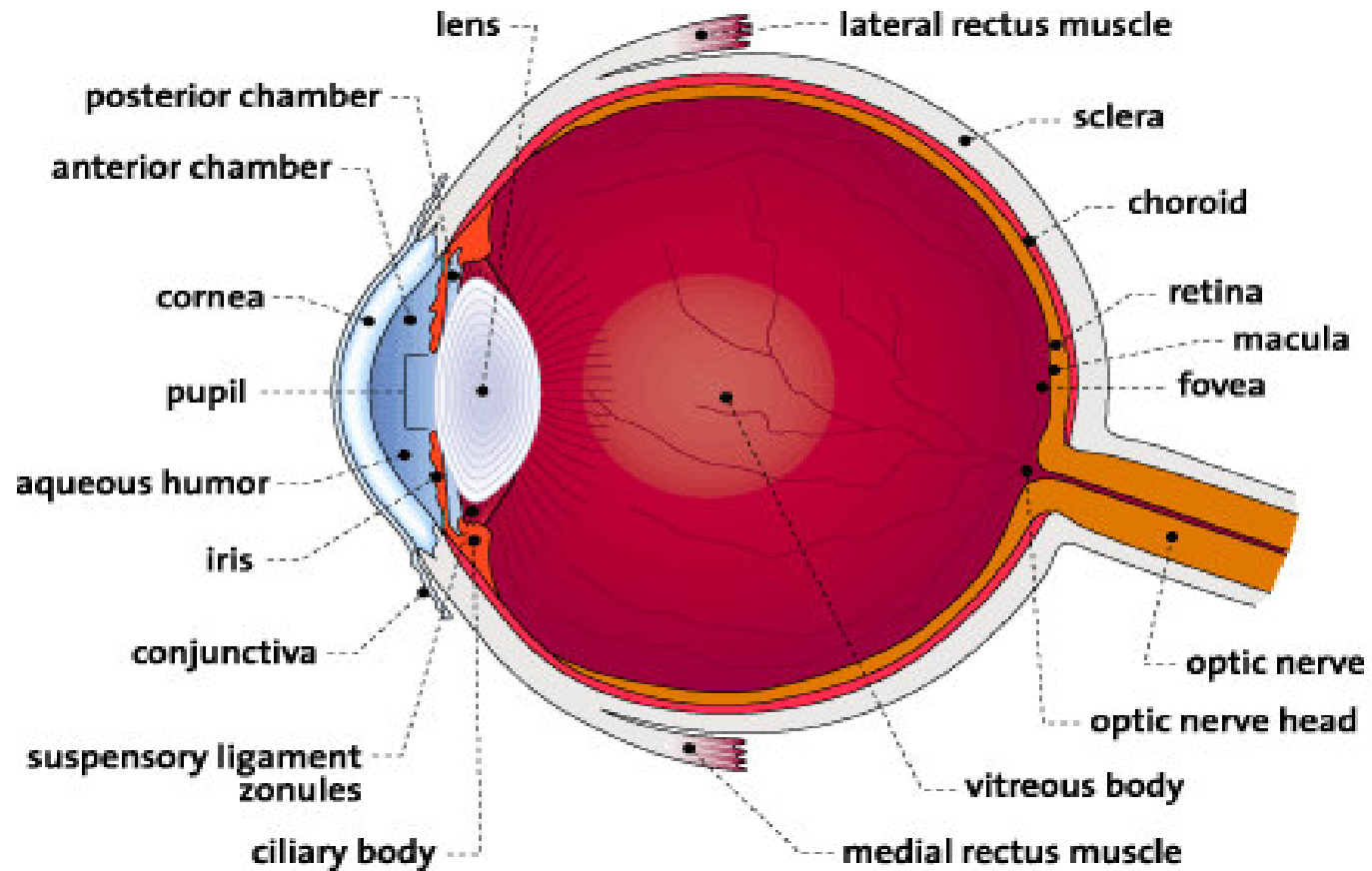
Eye Examinations

- Annually, with no symptoms
- Immediately if you notice: double vision, flashes of light (with or without “floaters”), pain, redness or discharge, a curtain coming up over your vision
- Frontal headache, and you “hear” your pulse in your temple
- Sudden change in vision
- Educate your doctor

Fluctuating Blurred Vision

- We need to be careful not to assume our symptoms are always due to our EDS and are non-actionable.
- #1 reason for fluctuating blurred vision: diabetes
- Every structure of the eye between the cornea and the ocular area of the brain could be the culprit.

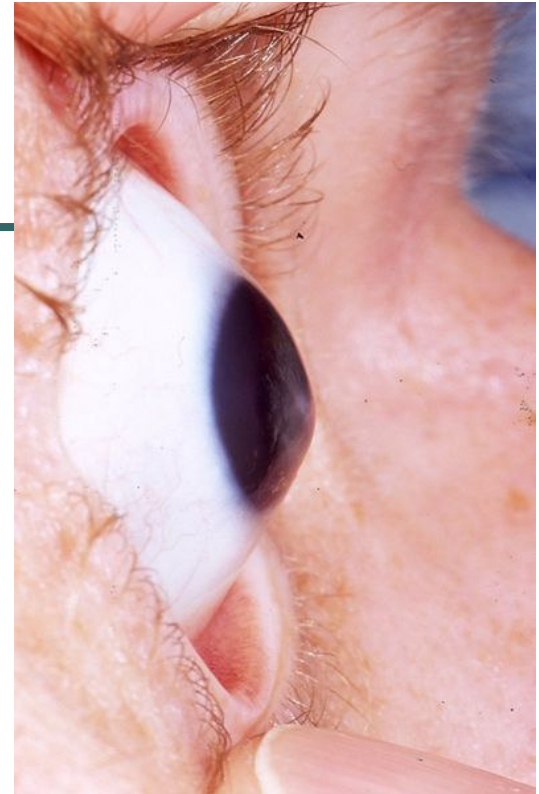
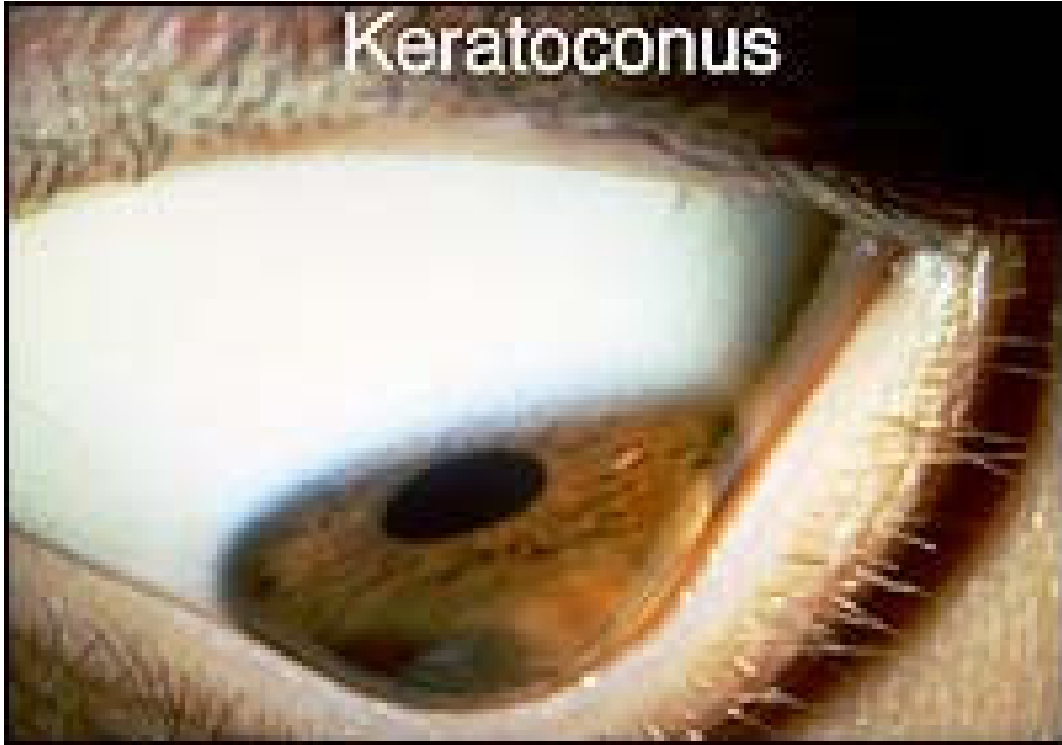
Eye Anatomy



Cornea

- Made of 25% of type VI collagen and MMP-2 (matrix metalloproteinase-2)
- Keratoconus
- Symptoms
- Ocular Topography
- Doctors should perform topography on all patients with unexplained blurred vision
- If topography indicates keratoconus, doctors should do a quick Beighton scale, understanding that hypermobility is more common in the metacarpo-phalngeal and wrist joints with keratoconic patients.
- Treatment
- EDS patients are NOT candidates for LASIK

Keratoconus



KERATOCONUS

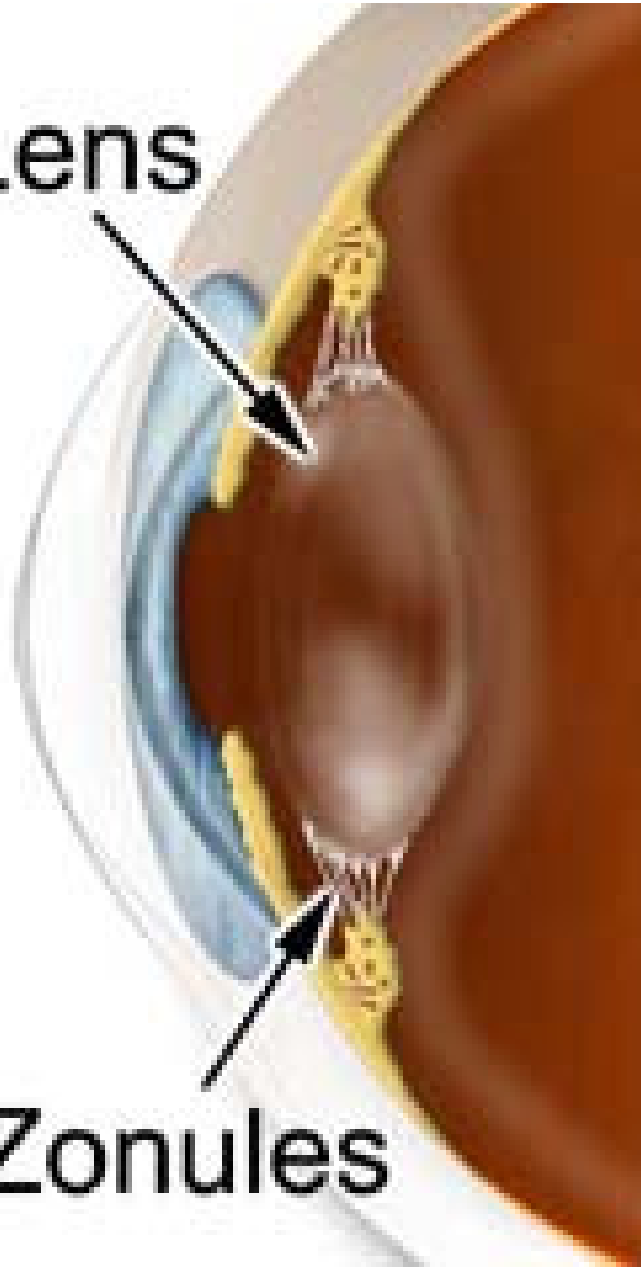
Current Studies

• “**Corneal topography in Ehlers-Danlos syndrome**”: 72 eyes of patients with EDS (genetically typed) had slitlamp biomicroscopy, retinoscopy and corneal topography to diagnose keratoconus. **Result**: no keratoconus was found. **Conclusion**: it appears that keratoconus in known population of patients with EDS remains rare. (1998)

• “**Orbscan mapping in Ehlers-Danlos Syndrome**”: A candidate for refractive surgery presented with classic (type I) EDS. Clinical examination revealed blue sclera, limbus-to-limbus corneal thinning, myopia, and astigmatism. Orbscan (B&L) pachymetry mapping provided a striking demonstration of the limbus-to-limbus thinning... Despite the theoretical biomechanical weakness from the thin cornea and defective collagen, regular surface topography was maintained without the development of keratoconus. Although all types of EDS remain a contraindication to laser refractive surgery, Orbscan mapping provides a valuable insight into corneal shape and thickness in this condition. (2004)

Lens

Zonules



More Studies

- **“*Joint Hypermobility in keratoconus*”**: This study of joint hypermobility among patients with and without keratoconus indicated that there was no statistically significant difference between the groups, when examining hypermobility of the joints in the trunk or knees. A difference, however was found for the metacarpo-phalyngeal and wrist joints. This supports the theory that keratoconus is a localized manifestation of a mild connective tissue disorder. Also, it can be stated that patients with keratoconus are five times more likely to show hypermobility of the metacarpo-phalyngeal and wrist joints. (1990)

Intraocular Lens

- Cataract
- Subluxation – weak zonules
- Difficulty in accommodation

Vitreous

- Made with collagen fibers
- Becomes more liquid over time
- Can cause retinal detachment, floaters
- EDS-ers are believed to experience more floaters
- Flashes of light, curtain over your vision - emergency

Retina

- Neural tissue
- Thin sclera, weak or stretchy sclera can cause myopia
- Look for blue sclera
- More prone to retinal holes, detachments, staphylomas, degeneration of the peripheral retina.
- Dilation of the eyes recommended

Diplopia

- See your eye doctor right away if the double vision is of sudden onset, especially if vertical.
- Notice: Is it out of one eye only?
Is it vertical or horizontal or both?
Only with near or distant objects?
Any other symptoms (lid droop?
pupil look bigger than other eye?)

Causes

- Structural: lens subluxation, staphyloma, weakness of eye muscles
- Neurological:
 - III. Nerve (oculomotor palsy) or IV. Nerve (trochlear nerve palsy)
 - If III. Nerve, and pupil is spared, it is ischemic. If pupil is involved, is usually due to compression.
- Basic rule: sudden onset, see your eye doctor.

Loss of Vision

- Complete or incomplete, one eye or both, lasting a few minutes.
- Sudden onset: see your doctor.
- Want to be sure it is not a blocked artery (or vein). CRA, CRV, branch artery or branch vein occlusion. Can cause stroke.
- Migraine episode: scintillating scotoma, may or may not get a headache, is vascular.
- My experience with this



Branch Artery Occlusion

Dry Eyes

- Normal tears that cover the corneal surface comprise three layers:
- **The lipid**, or oil layer, which is the outer layer of the tear film and helps prevent the lacrimal layer beneath it from evaporating or overflowing the lower eyelid
- **The lacrimal**, or watery layer, which is the middle layer and contains salts, proteins, and an enzyme called lysozyme that actually protects and nourishes the eye
- **The mucoid**, or mucus layer, which is the bottom layer of tears. It contains cells called goblet cells that cause the tears to adhere to the eye.
- All three layers of tears, in proper balance are necessary for proper lubrication.

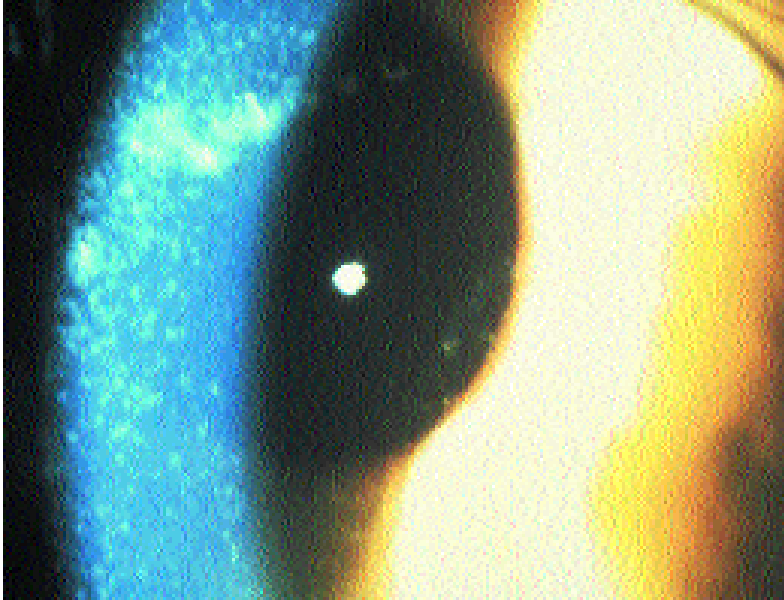
Dry Eye Evaluation

- Helps the doctor determine the cause, and thus the treatment, for the dryness.
- Is there too much, or too little of one or more layers of your tears?
- We will examine the structure of your tears. Fluorescein, for example, will tell us your “tear break-up time”.
- Fluorescein, together with other dyes (lysamine green, rose bengal) will indicate the extent of cell dryness and damage. All painless.
- We can also measure the amount of “watery tears” formed over 5 minutes (Called a “Schirmer Test”).

Treatment

- Depends upon the cause, but could include:
- Non-preserved moisture drops or jells.
- Restasis
- Topical steroid drops
- Punctal Occlusion
- Fish oil capsules, Omega 3
- Mucinex with guaifenesin if the mucous layer is out of balance
- Staying hydrated
- Changing make-up or make-up removers
- Contact lens wearers may need a different type of contact lens.
- Avoidance: ceiling fans, car vents, prolonged computer use, some medications (Benadryl, Sudafed, Bromfed, for example).

Punctal Occlusion



Disclaimers and Caveats

- This next section discusses some of the devices and medications that have helped me regain some functionality. Of course, all of us are different to some degree, and you should check with your physician before trying any of the medications discussed – even the ones that are “over the counter”.
- Some of the medications that help my form of dysautonomia, especially the vasoconstrictors, can raise blood pressure, and be quite dangerous to those of us with weak blood vessels.
- This can be a starting point, however, for you and your doctors to discuss some forms of therapy that you may not have yet considered.

What Has Helped Me

- Ruby – my pink exercise ball – for sitting and exercising!
- Hard cervical collar in the car and doing things around the house.
- Soft cervical collar for sleep
- Clavical, shoulder brace at night
(helped the systemic symptoms!)
- Small towel under my neck and head, instead of pillow
- Sleeping on my back
- C Pap?
- Braces as needed: finger splints, wrist splints, elbow braces, knee braces, ankle splints

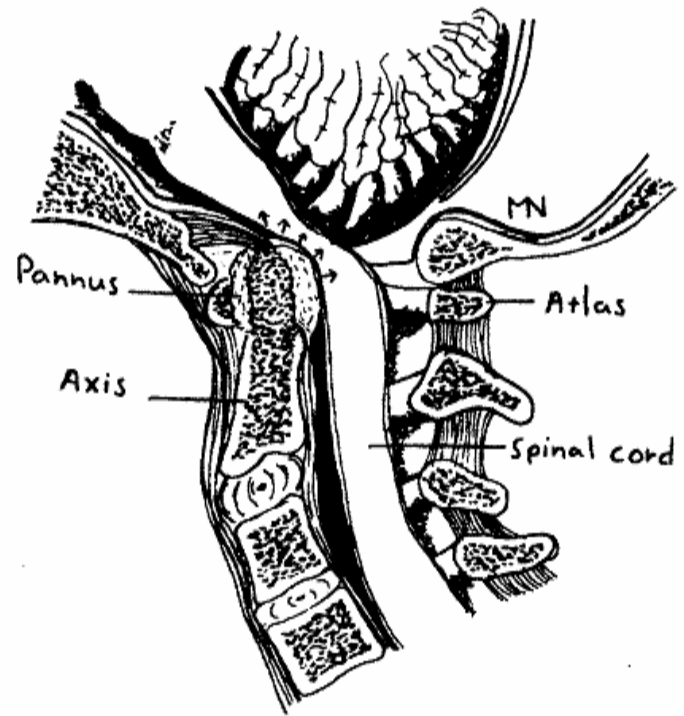
More Helpers

- Abdominal binder!
- Adjust reading and work to eye level
- Wearing cervical collar while exercising
- Neck massages
- Paraspinal exercises
- Arch supports in shoes
- Tempur-Pedic mattress, satin sheets and jammies
- A rubber ball to pop hip back in place
- A glass of alcohol!
- Cold, caffeinated beverage in the afternoon

Why do We Have all of These Weird Neurological Symptoms?

- Cranio-cervical instability causes pannus formation around the odontoid bone, crowding the brain stem, sometimes accompanied by Chiari (often only visible with a vertical MRI).
- Look for “**blepharoclonus**”: tremors of the eye muscles during gentle closure of the eyes.
- Study: “***Blepharoclonus and Arnold-Chiari malformation***”: Study examined 4 patients, two of whom had EDS.
Conclusion: blepharoclonus is an underdiagnosed neuro-ophthalmological sign of ACM (and I would propose, crowded brain stem area in general). (2001)
- I would also propose the mislocation of the odontoid periodically (causing episodic and dramatic regression).

MRI



Health Through Pharmacy!

- Improving blood flow to the brain helps with many symptoms, including “brain fog”:
- Sudafed, or almost any vasoconstrictor (caution!). Abdominal binder. Find your “brain position”.
- Cymbalta: balances some of the brain chemistry changes with hyperadrenergic POTS, some pain relief, some mood lifting. Usual maximum dose: 60 mg in a.m. Some doctors prescribe as much as 120 mg. q d for EDS.
- Magnesium supplements – in p.m.
- Fish oil: shown to help with pain, especially spinal pain. May help those of us with abnormal lipid profiles, also
- Provigil: Take in the morning (usually with Xanax to avoid the jitters). Helps with mental focus and fatigue.
- Have hormones checked frequently. Testosterone: be sure your doctor checks the “free test.”.

Pharmacy Continued

- Xanax: we are more prone to anxiety (?), but it is certainly a life saver for hyperadrenergic POTS!
- I have an anti-inflammatory every day (ibuprofen, but beware of IBS symptoms and eosinophilia).
- HGH? (are levels normal? HGH can improve bone density and muscle mass).
- Watch blood sugar levels – are we more prone to reactive hypoglycemia? Be especially careful with HGH and glucosamine! Consider avoiding refined carbs and have some protein with every meal (small meals 5 – 6 times per day).
- Tachycardia episodes at night relieved for me with Xanax and Lunesta in combo.

More Pharmaceuticals

- Retin-A for wound healing – especially if mild
- **Study:** “Tretinoin Pre-Treatment but not Direct Treatment Shows a Beneficial Effect on Wound Healing in Diabetic Mice”; (2004)
- **Study:** “Effectiveness of short-contact topical tretinoin in promoting wound healing in mice (1987). **Result:** wound healing was accelerated with short-contact topical application of tretinoin in mice.

Eye Doctor's Information

- I have Ehlers-Danlos Syndrome Type _____, which causes defective connective tissue in my collagen. Please be sure to check carefully for the following:
 - Epicanthal folds
 - High myopia
 - Keratoconus
 - Blue sclera
 - Lens subluxation
 - Angiod Streaks
 - Cataracts
 - Dry eyes
 - Glaucoma (with pachymetry for accuracy)

More Eye Doctor Information

- Photophobia
- Retinal Detachments, holes, tears
- Strabismus
- Macular Degeneration
- Posterior Staphyloma
- Carotid-cavernous sinus fistulas
- Accommodative difficulties
- Dry eyes
- Diplopia, monocular or binocular
- Large phorias which may be symptomatic
- Blepharoclonus

Recommended Testing/Information as Part of a Routine Ocular Exam for a Patient with EDS

- Complete slit lamp exam with TBUT. Understand that I may be more prone to corneal dystrophies, dry eyes.
- Dilated fundus exam; fundus photography
- Ocular topography to rule out early keratoconus
- Scanning Laser Ophthalmoscopy may be helpful – Retinal Thickness Analyzer, Stratus OCT, etc.
- Orbscan and/or pachymeter to check corneal thickness
- EDS patients are not good candidates for LASIK.
- Pupil testing (rule out APD), aperture measurements (check for ptosis), rule out blepharoclonus.
- ?? I may be more prone to recurrent corneal erosions ??
- I may be more prone to migraine episodes and/or aura without the migraine headache.
- I may be more prone to macular degeneration

So Many Questions!

- Viral-induced – improves over time. Why?
- Pressure of the brain (or CSF) on pituitary, brain stem. What about the hypothalamus? What about the reticular activating system? (When I leaned forward, my heart rate jumped up!). Change in hormones – due to affects of pituitary or hypothalamus? (mine go up and down).
- Pressure on cranial nerves?
- Ischemic or mechanical pressure or both?
- Studies indicate that checking EDS patients reveals few keratoconic patients – HOWEVER – anecdotal evidence reveals that the reverse may not be true and needs to be studied.
- Dramatic and immediate change in symptoms is possible – why?

Research

- Any relationship of EDS to recurrent corneal erosions?
- How can research be shared more readily in the medical community?
- How can we educate all medical professionals?
- More research into genotypes and phenotypes, and their overlap
- Better treatment options for cranio-cervical instability, pannus, etc.
- More research on how this affects our brain chemistry – Depression? Bipolar-like behavior? Hypomania? Our personalities?