

Ocular Complications of MPS Disorders

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11th International Symposium on MPS
& Related Disorders
Adelaide, June 2010

Ocular Complications

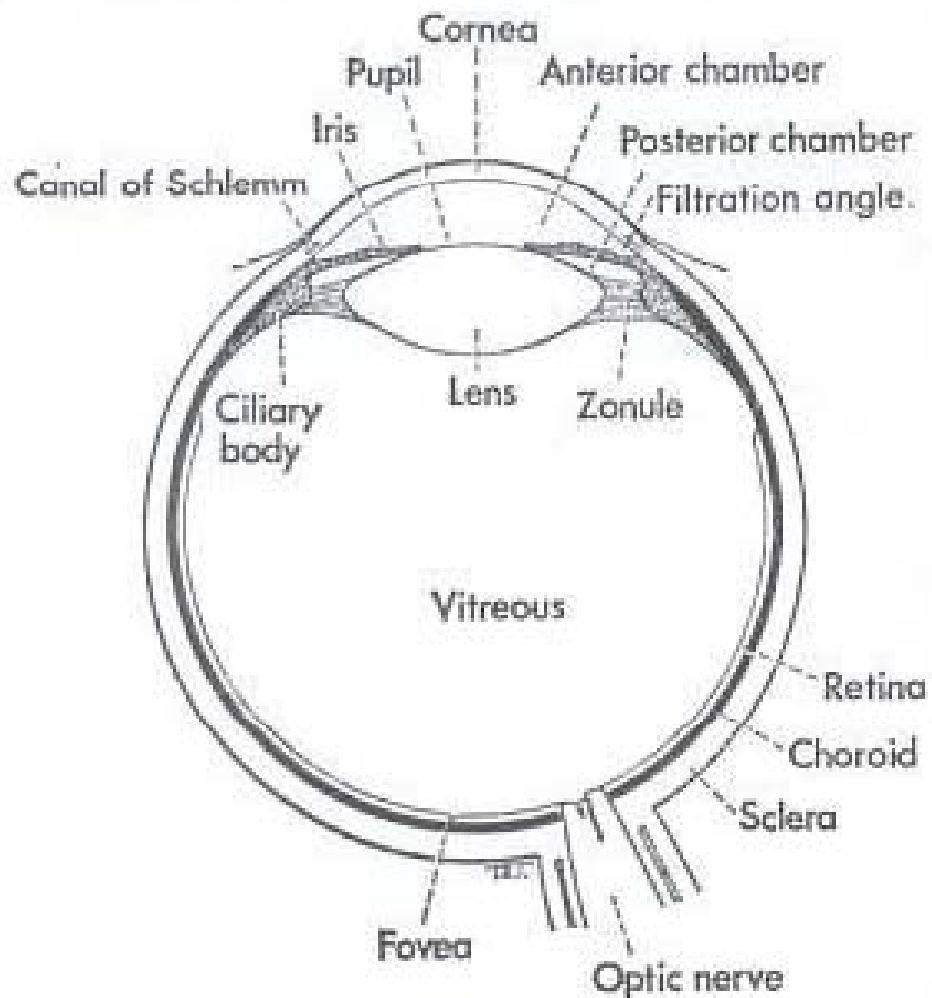
- Corneal clouding
- Glaucoma
- Retinopathy
- Optic neuropathy

Corneal clouding

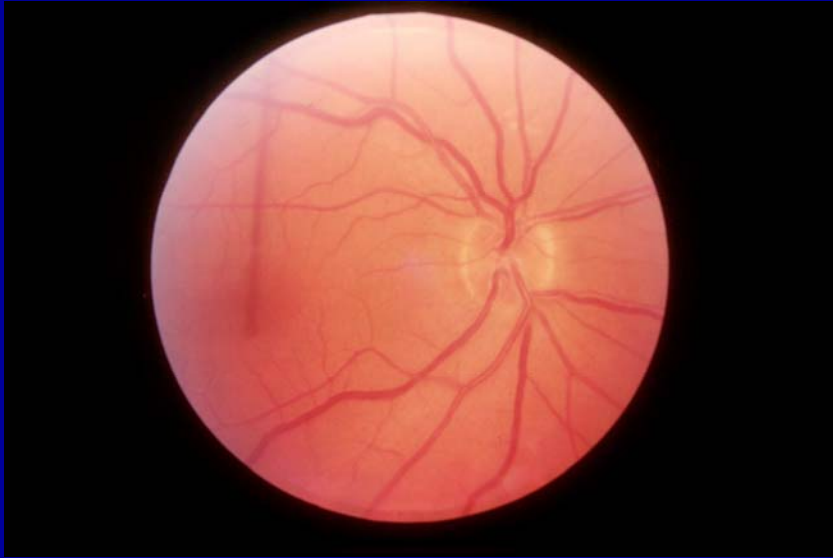
- Most severe in IH, IHS and VI



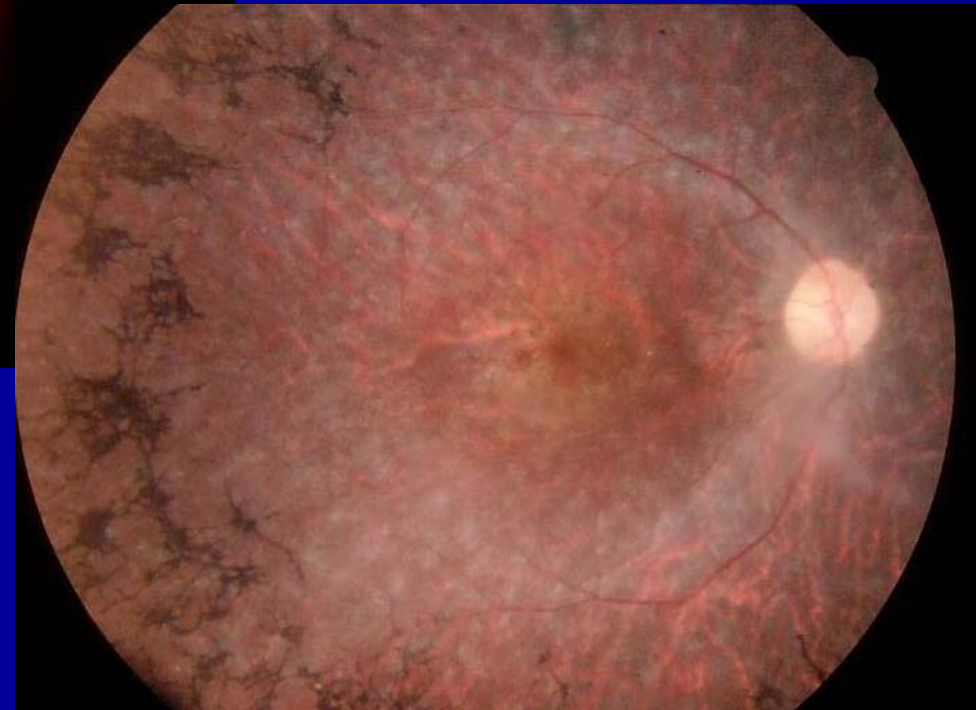
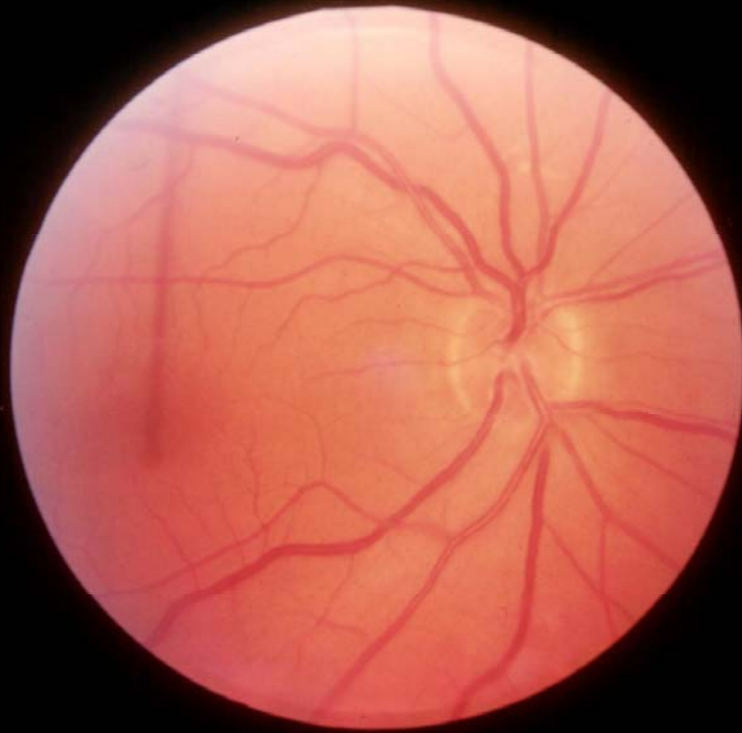
Glaucoma



Glaucoma

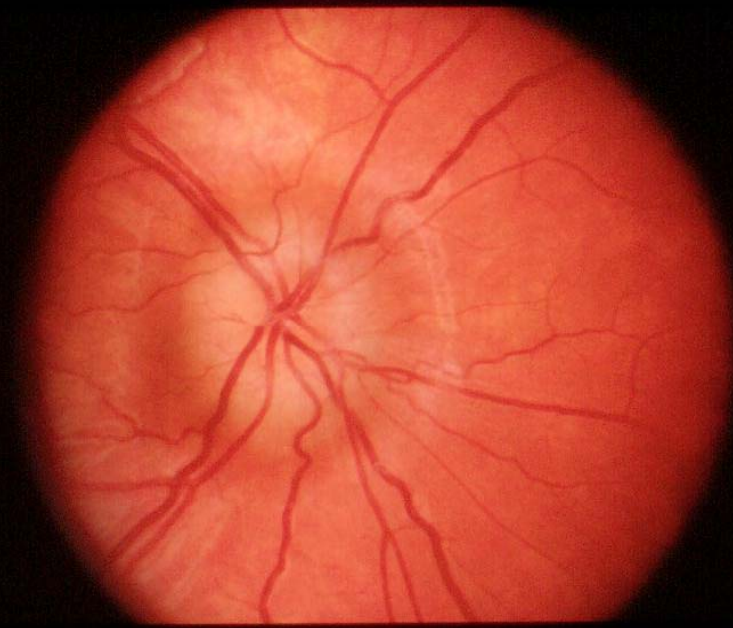


Retinopathy



Ocular Complications

- Optic Neuropathy



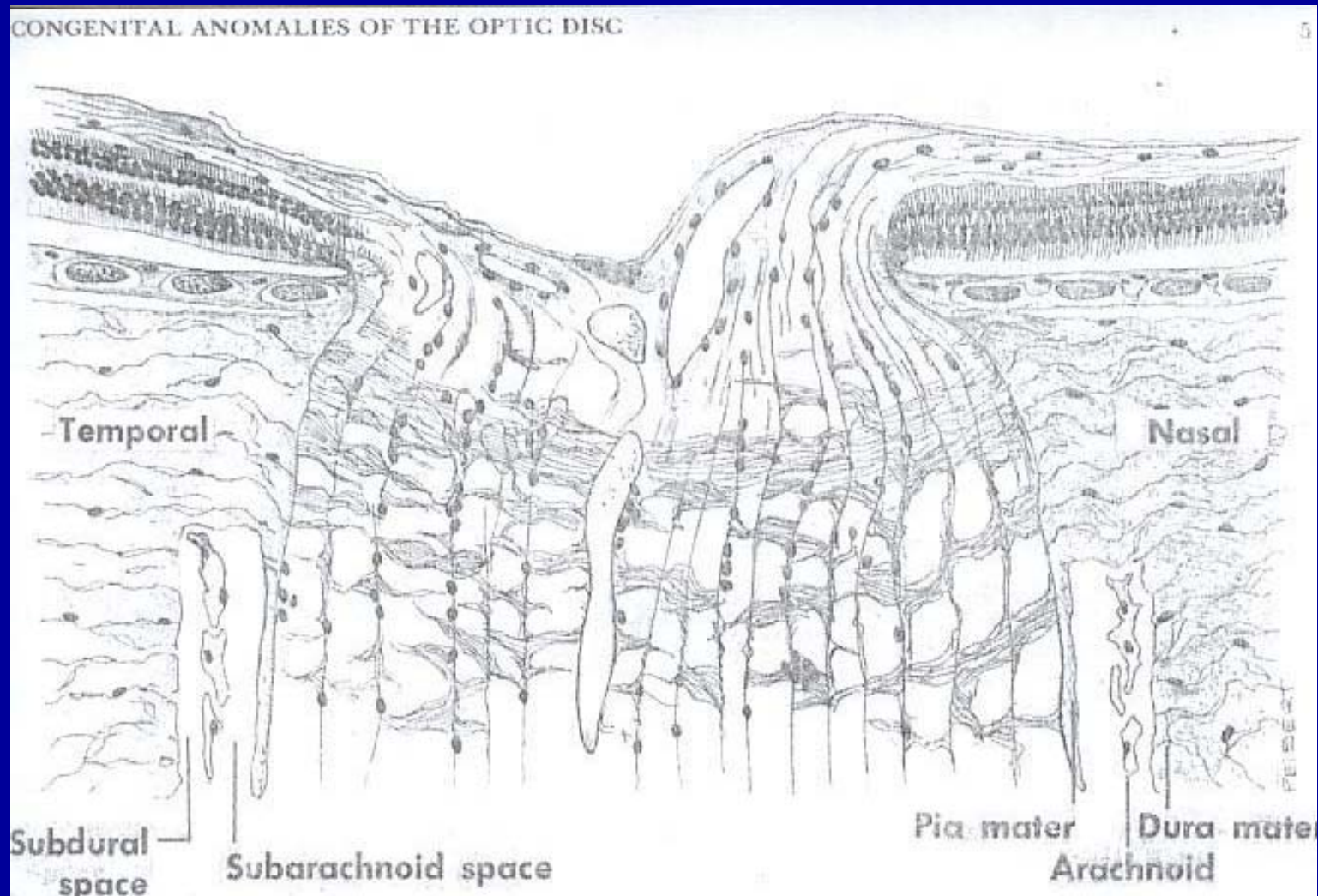
Optic Neuropathy- Causes

- Glaucoma
- Retinopathy

- Deposition within optic nerve fibres
- Increased intracranial pressure

- Deposition within sclera

Optic Neuropathy



Retrospective study

- Children's Hospital, Westmead, 1997-2009 (Dr Azura Ramlee)
- Financially supported by Genzyme Australasia
- 45pts:
 - only 29 eye reviews

Retrospective Study

- MPS I – 11
- MPS II – 11
- MPS III – 18
- MPS IV – 1
- MPS VI – 4

Retrospective study- Results of 29pts

- 3 > 6/12, most had ↓VA 6/15 to 6/60 (4 blind)
- Corneal clouding- 13 (all MPS I & VI, 1 MPS IV)
- Glaucoma- 1 (MPS VI)
- Retinopathy- 4 (MPS I & II) ?MPS III (most not seen)
- Optic disc changes- 6 (MPS I,II & VI)
- Long sightedness and astigmatism were common refractive errors

Retrospective Study

- MPS 1-2pts BMT- stable (6.5yrs)
1pt ERT- progressive corneal clouding
- MPS VI- 4/4 pts ERT-stable (6.5yrs)

Prospective Study of Ocular Complications of MPS Disorders

- Review of all MPS children on data base
CHW- 5 yr study (Ethics approval)
- Financially supported Genzyme
Australasia
- Other pts in Australia as well (Adelaide)

MPS Prospective Study- Aims

- To determine the rate and severity of ocular complications in each type
- To set up clinical guidelines as to the frequency of eye assessments and which are the best tests

MPS Prospective Study- Aims

- To detect and prevent vision loss from a treatable cause
- To assess if BMT/ERT slows down, arrests or reverses eye disease

MPS Prospective Study- Challenges

- Corneal clouding- how to assess progression?
- Corneal thickness- pachymeter



MPS Prospective Study- Challenges

- Glaucoma- how to monitor?
- Measuring IOP- Icare tonometer



- Optic nerve and retinal changes (limited by corneal clouding)
- Visual fields-requires co-operation

MPS Prospective Study- Challenges

- Retinopathy- symptoms?
- May need ERG- requires significant co-operation



MPS Prospective Study- Challenges

- Optic Neuropathy- likely to be multifactorial
- Ocular ultrasound- requires co-operation
- VEP's
- May need MRI scan



MPS and the EYE

- Rare conditions
- Difficult to interpret results with low numbers
- Planned multicentre trial with Jane Ashworth (UK), Gail Summers (USA), Kristina Tear Fahnehjeln (Sweden) and Susanne Pitz (Germany)
- Meeting in Stockholm Sept 2010 (Biomarin)

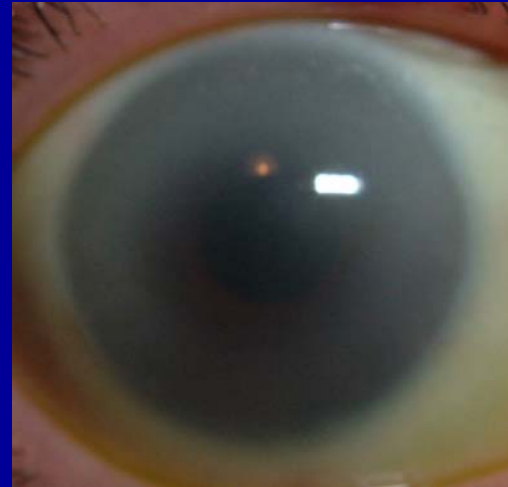
MPS and the EYE

- Prospective Study in Australia- colleagues:
- Azura Ramlee- KL Malaysia (Recent Paediatric Ophthalmology Fellow CHW)
- Katie Scanlon, Head Orthoptic Dept CHW
- Sue Silviera, JAFF Research Fellow, RIDBC, Renwick Centre, Uni Newcastle
- Deepa Taranath, Consultant Ophthalmologist, WCH, FMC Adelaide

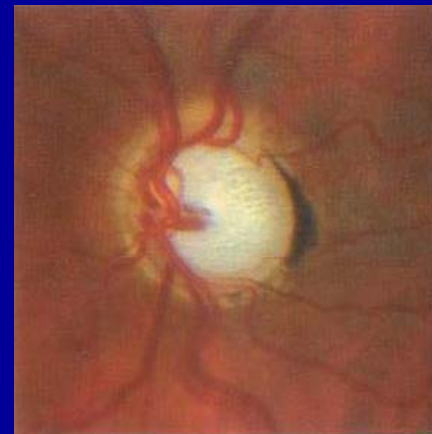


Ocular Complications

- Corneal clouding



- Glaucoma



Glaucoma- Assessment

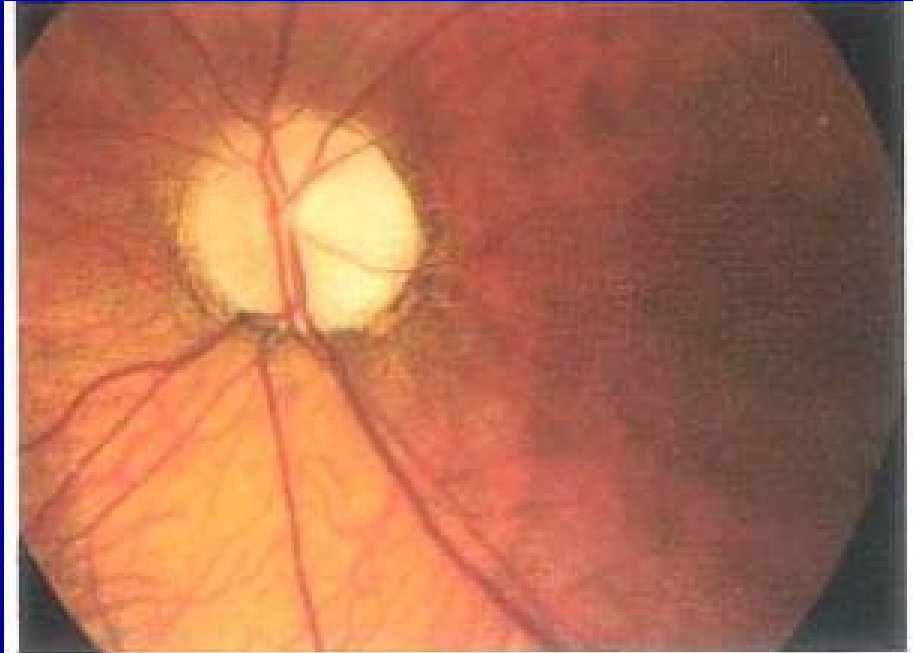
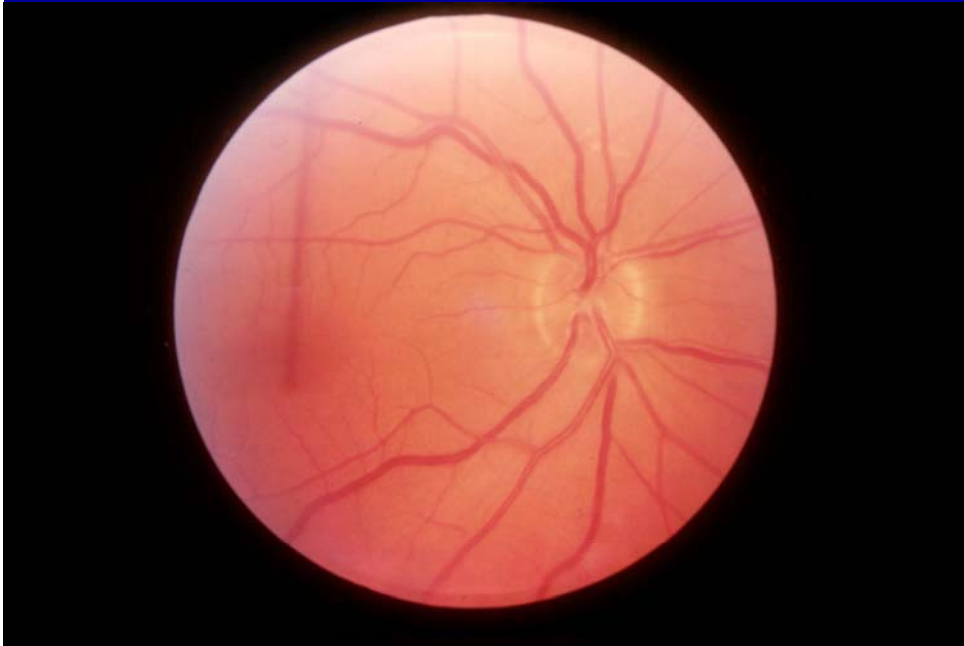
- Buphthalmos
- Measuring IOP
- Optic nerve changes
- Visual fields



Retinopathy

- Symptoms- night vision difficulties, loss of peripheral vision
- Signs- pigment changes in retina, attenuated vessels, macular changes, optic atrophy
- ERG to confirm

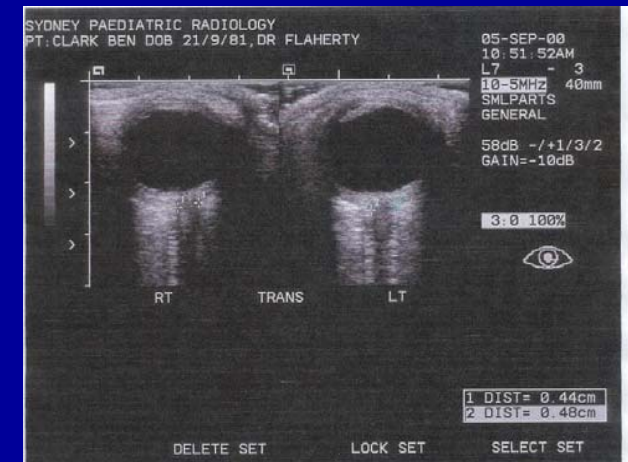
Optic Neuropathy



79 Primary optic atrophy. The disc is flat and has a well-defined margin; pallor is more pronounced on its temporal side.

MPS Prospective Study- challenges

- Optic Neuropathy:
- Fundal view limited by corneal clouding
- Likely to be multifactorial
- Ocular U/S- requires co-operation
- May need MRI scan

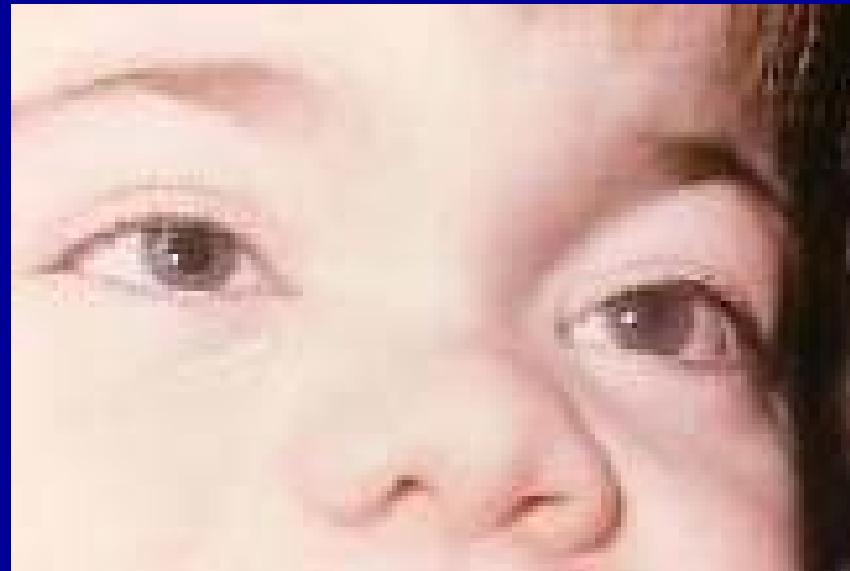


Retrospective study- Results of 29pts

- 3 > 6/12, most had ↓VA 6/15 to 6/60 (4 blind)
- Corneal clouding- 13 (all I & VI, 1 IV)
- Glaucoma- 1 (VI)
- Retinopathy- 4 (I, II) ?III (most not seen)
- Optic atrophy-6 (I,II,VI)
- HM/ astigmatism common refractive errors

MPS - treatment

- Correct refractive errors
- Treat glaucoma
- Treat \uparrow ICP
- Corneal grafting



MPS- New modalities Tx

- Bone marrow transplantation (BMT)
- Enzyme replacement therapy (ERT)
- Slow down, arrest or reverse eye changes?