An atypical presentation of progressive supranuclear palsy. Patient presents with gaze palsy very early after onset of disease and with supravergence restricted more than infravergence. Diagnosis and management are described.

I. Case History

a. 61 year old African American male presents for eye exam.

b. Chief complaint is blurred vision and binocular horizontal diplopia on left, right, and down gaze as well as when attempting to read with glasses. Symptoms have been increasing in frequency and worsening since he first noticed about 8 months ago.

c. Ocular history is remarkable for trauma OS with metal foreign body 6 years ago.

d. Medical history is remarkable for hyperlipidemia, hypertension, peripheral vascular disease, colonic and rectal polyps, cervical diskectomy with myocardial infarction during surgery, and erectile dysfunction. The patient was diagnosed with probable progressive supranuclear palsy (PSP) with classic Richardson syndrome phenotype during the course of follow-up.

e. The patient presents on hydrochlorothiazide, lisinopril, metoprolol, nifedipine, simvastatin, trazodone, vardenafil, tamsulosin, sildenafil, loratadine, oxybutynin, ranitidine, fish oil and a multivitamin. Carbidopa/levodopa was added during the course of follow-up.

II. Pertinent Findings

a. Clinical Findings

i. Entering VA with correction: OD 20/50+, OS 20/40+2

ii. Pinhole VA: OD 20/40+2, OS NI

iii. BVA: OD 20/25-3, OS 20/25+

iv. Pupils: (+) direct and consensual response without APD or light-near dissociation

v. Extraocular motilities: 0% of normal superior gaze, 60% of normal inferior gaze, and 30% of normal left and right gaze. No diplopia. Normal motilities with doll's test. Slow saccades noted.

vi. Cover test: orthophoria distance and near without correction

vii. Incomplete blink, (-) Bell's phenomena OD, OS
Anterior segment examination reveals inspissated meibomian glands, a mucous strand inferiorly OS and poor tear film OD, OS. Otherwise normal.

Intraocular pressure: 08 mmHg OD, 09 mmHg OS by Goldmann applanation tonometry.

b. Physical findings
   i. The patient has symptoms of postural difficulties and gait instability.
   ii. Patient states he has fallen 10 or more times in the last year, mostly backwards but occasionally forward.

c. Laboratory studies: none

d. Radiology Studies
   i. Recent MRI revealed mild frontal lobe atrophy, midbrain atrophy, periventricular diffuse hyperintensities which are likely microvascular changes, and no mass lesions.

III. Differential diagnosis
   a. Progressive supranuclear palsy (PSP)
   b. Chronic progressive external opthalmoplegia (CPEO)
   c. Dorsal midbrain syndrome
   d. Parkinson’s
   e. Multiple system atrophy (MSA)
   f. Corticobasal degeneration (CBD)
   g. Dry eye syndrome

IV. Diagnosis and discussion
   a. The patient was diagnosed with probable PSP with classic Richardson syndrome phenotype.
      i. Diagnostic criteria requires that a patient with suspected PSP be older than 40 years of age, have severe postural instability and falls within the first 12 months of disease, and have vertical supranuclear gaze palsy or slowed vertical saccades. Of importance to note, slowed vertical saccades may be apparent initially but a vertical gaze palsy usually doesn’t develop for many years after disease onset.
ii. The patient fits all of the above; additionally, his MRI is consistent with PSP, showing a classic “hummingbird” or “penguin” sign due to prominent midbrain atrophy.

iii. The disease is rare, estimated at only 6-7 cases per 100,000.

iv. PSP is defined pathologically by the accumulation of tau proteins and neuropil threads in specific areas of the brain.

v. Richardson syndrome is the classic form of PSP and is characterized by early onset postural instability and falls.

b. Unique features

i. The patient’s vertical supranuclear gaze palsy developed very early, only months after onset of the disease. Typically, this occurs many years after onset.

ii. Gaze is significantly more restricted superiorly rather than inferiorly. Patients with PSP usually have more trouble looking down than up.

iii. The patient occasionally falls forwards when patients with PSP usually fall backwards.

iv. The patient’s visual acuity is moderately reduced secondary to dry eyes and not directly to PSP.

V. Treatment and management

a. The patient was prescribed separate reading only and distance only glasses, and educated on keeping objects of interest in primary gaze of focus. Instructed patient to start artificial tears 4x/day OD, OS as well as before watching TV or reading.

b. The patient returned 1 month later denying diplopia but still complaining of blur at distance and near.

i. Anterior segment evaluation revealed inspissated meibomian glands, an instant tear break-up time, 1+ punctate epithelial erosions greater inferiorly, and mucous debris inferiorly OD, OS.

ii. Patient was educated on dry eyes being likely cause of blurred vision. Discontinued artificial tears and started Celluvisc every 2 hours and preservative free lubricating ointment at bedtime OD, OS. Patient to return in 3-4 weeks or sooner if needed.

1. Dry eye syndrome is very common in PSP secondary to a significantly reduced blink rate, increased meibomian gland
disease, and decreased corneal sensitivity. The patient is typically asymptomatic although corneal surface disease is apparent.

c. Bibliography


VI. Conclusion

a. PSP is a rare disease but has debilitating ocular sequelae. Education on gaze awareness, as well as postural changes, can be very beneficial.

b. Dry eye is common in PSP and can cause blurred vision. Because patients are often asymptomatic, strict education and aggressive treatment is required.

c. This patient was unique in that his vertical gaze palsy develop very early and his gaze was restricted more on supravergence than on infravergence. Additionally, the patient reported sometimes falling forwards when patients with PSP usually fall backwards.