**Poster 76**

**Ophthalmoplegia and Downbeat Nystagmus in Stiff-Person Syndrome**

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**Introduction:**
Stiff-person syndrome (SPS) is a poorly understood condition with generalized stiffness and rigidity thought to be centrally mediated. Diagnosis is supported by the presence of glutamic acid decarboxylase antibodies (GADab). Abnormal eye movements are infrequently reported in SPS and were attributed to co-existent myasthenia in early cases. Recent reports suggest GADab as a direct cause. GADab have also been reported with downbeat nystagmus (DBN) with or without ataxia in the absence of SPS.

**Methods:**
A 19 year old woman with SPS supported by GADab (501 nmol/L; normal <0.02) presented with increasing stiffness, tremors, binocular diplopia, and ptosis. Evolution of eye movements over several days is presented and discussed in the context of prior reported cases and the pathophysiologic basis by which GADab may directly impair ocular motility.

**Results:**
Exam revealed diffuse ophthalmoplegia (downgaze greater than upgaze and leftgaze greater than rightgaze) with motility reduced to < 30% of normal in all directions. Saccades were slow and were affected to the same extent as smooth pursuit. Deficits were slightly improved with vestibulo-ocular reflexes. Bilateral ptosis and generalized weakness, stiffness, and tremors were present. EMG with repetitive stimulation, myasthenia serologies, tensilon test, and chest CT were negative. Five days later, eye movements were stable with exception of interval development of DBN.

**Conclusion:**
Eye movements initially suggested a brainstem supranuclear gaze palsy, rather than a neuromuscular junction deficit. GADab are increasingly associated with DBN in the absence of SPS and evolution to DBN over several days in this patient provides support of direct GADab mediated injury. This case adds support to mounting evidence from 2 prior reports without DBN of direct GADab impairment of GABAergic transmission in supranuclear brainstem and cerebellar motility pathways.

**References:**

**Key Words:** stiff person syndrome, GAD antibodies, downbeat nystagmus, ophthalmoplegia, supranuclear

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