

[Neurology](#). 2000 Nov 28;55(10):1531-5.

## **The clinical spectrum of anti-GAD antibody-positive patients with stiff-person syndrome.**

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### **Abstract**

**OBJECTIVE:** To evaluate the clinical spectrum of anti-GAD-positive patients with stiff-person syndrome (SPS) and provide reproducible means of assessing stiffness.

**BACKGROUND:** SPS can be difficult to diagnose. Delineation of the clinical spectrum in a well defined population will increase diagnostic sensitivity.

**METHODS:** In 20 anti-GAD-positive patients with SPS (six men, 14 women), screened among 38 referred patients, the authors assessed symptoms and signs, degree of disability, associated conditions, and immunogenetic markers. Degree of bending, distribution of stiff areas, timed activities, and magnitude of heightened sensitivity were examined monthly for 4 months in five patients.

**RESULTS:** Average age at symptom onset was 41.2 years. Time to diagnosis was delayed from 1 to 18 years (mean 6.2). Stiffness with superimposed episodic spasms and co-contractions of the abdominal and thoracic paraspinal muscles were characteristic. All had stiff gait and palpable stiffness in the paraspinal muscles. Stiffness was asymmetric or prominent in one leg in 15 patients (stiff-leg syndrome) and involved facial muscles in 13. In one patient spasms lasted for days (status spasticus). Twelve patients needed a cane and seven a walker due to truncal stiffness and frequent falls (average three to four per month). Distribution of stiffness and degree of heightened sensitivity were two reproducible indices of stiffness and spasms. Autoimmune diseases or autoantibodies were noted in 80% and an association of with DRss(1) 0301 allele in 70%.

**CONCLUSIONS:** SPS is 1) frequently misdiagnosed due to multifaceted presentations and asymmetric signs, 2) disabling if untreated, and 3) associated with other autoimmune conditions.

PMID: 11094109 [PubMed - indexed for MEDLINE]