Objective: Describe the demographics and clinical characteristics of patients within the University of Utah Healthcare system with stiff person syndrome (SPS), stiff limb syndrome, and progressive encephalomyelitis with rigidity and myoclonus (PERM).

Background: The clinical phenotypes associated with glutamic acid decarboxylase (GAD65), amphiphysin, and glycine receptor (GlyR)-associated neurologic autoimmunity have expanded from the original descriptions of SPS as an autoimmune disease that typically presents with symptoms of proximal limb and axial rigidity and spasms. Treatment with benzodiazepines is first-line, with immunotherapy also playing an important role.

Design/Methods:
We performed a retrospective review of patients within the University of Utah Healthcare system meeting criteria 1 and 2 or 2 and 3, with lists cross referenced against one another:
1. Positive GAD65 (>100 IU/mL) or amphiphysin serum or cerebrospinal fluid testing at Associated Regional and University Pathologists, Inc. (ARUP Laboratories), or positive GlyR testing (performed on a research-basis) at Mayo Medical Laboratories.
2. At least one visit with a University of Utah clinician in the Department of Neurology.
3. At least one ICD-9-CM or ICD-10-CM code for stiff person syndrome (333.91 or G25.82, respectively), encephalomyelitis (323.9 or G04.90, respectively); or ICD-10-CM code for GAD65 seropositivity (R76.0).

Results: We identified patients meeting the above criteria and characterized them as either classic SPS, stiff limb variant, or PERM variant, or if they had another recognized phenotype other than SPS spectrum, we described the associated phenotype. Each case was classified by relevant demographics and clinical data including age at symptom onset, age at diagnosis, spasm location, associated neurological symptoms, associated autoimmunity and malignancy, and response to symptomatic therapy and immunotherapy.

Conclusions: This is a comprehensive characterization of GAD65, GlyR, and amphiphysin-associated neurologic autoimmunity within a single regional academic medical center. These conditions may not be as rare as previously reported, and outcomes are variable, with a tendency toward progressive disability.