GAD65 and Glycine Receptor-Associated Neurologic Autoimmunity and Stiff-Person Syndrome within the University of Utah Health Care System

Jonathan R. Galli MD, Amanda L. Piquet MD1,2, Jacob Kresser, Julia Klein APRN1, Judith Warner MD1,4, Kathleen Digre1,4, Lisa K. Peterson PhD5,6, Anne E. Tebo PhD5,6, Thomas R. Haven PhD6, M. Mateo Paz Soldan MD PhD3,1, John Rose MD1, John Greenlee MD1,7, Stacey L. Clandy MD PhD3,7

1. University of Utah, Department of Neurology, Salt Lake City, UT. 2. University of Colorado, Department of Neurology, Aurora, CO. 3. University of Utah, Department of Internal Medicine and Bioinformatics, Salt Lake City, UT. 4. University of Utah, Department of Pathology, Salt Lake City, UT. 5. Associated Regional and University Pathologists (ARUP) Laboratories, Salt Lake City, UT. 6. George E. Walensky Veterans Affairs Medical Center, Salt Lake City, UT.

Objective
Describe epidemiological characteristics, antibody status, and treatment outcomes of stiff person syndrome patients within University of Utah Health.

Background
Stiff person syndrome (SPS) is an autoimmune disease that classically causes co-activation of agonist and antagonist muscle groups leading to severe muscle rigidity and spasm.1 GAD65 is the most common associated neuronal autoantibody: patients may also have associated glycine receptor (GlyR) or amphiphysin autoantibodies.2

Epidemiological Data

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Use</th>
<th>Benefit</th>
</tr>
</thead>
<tbody>
<tr>
<td>Classic Stiff Person</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>SPS &amp;</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>SIP Lesion Syndrome &amp;</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>PERM</td>
<td>2</td>
<td>1</td>
</tr>
</tbody>
</table>

Antibody Status

![Antibody Status Chart]

Discussion and Conclusions
We identified 31 patients with SPS within the University of Utah healthcare system. Patients were predominantly female (78%) with the most common phenotype being classic SPS or SPS with concomitant cerebellar symptoms or epilepsy. Our cohort had co-existing autoimmune disease (63%) and malignancy (13%). GAD65 was the most common associated antibody; GlyR antibody testing should be considered in patients with SPS phenotype with negative or low GAD65 titers.

Limitations: Due to the electronic search method that was utilized, an accurate incidence could not be calculated. Treatment outcomes need more objective measures. The 2 patients who underwent bone marrow transplant were too early in their course to present additional data.

References