A Retrospective Study of Adult Patients with Parry Romberg Syndrome

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Background

- Parry Romberg Syndrome (PRS), also known as progressive hemifacial atrophy, is a rare variant of morphea.
- Estimated incidence of 1,700,000
  - Predominantly children
- Etiology and pathophysiology are unknown:
  - Likely autoimmune, though theories also suggest contributing factors including trauma, infection, and genetics
- Characterized by atrophy of skin and subcutaneous tissue, including muscle, bone, and cartilage, classically in a unilateral hemifacial distribution
  - Potential neurologic, ophthalmologic, and oral/dental involvement
- Diagnosis is typically clinical, although supportive imaging and/or histology are often obtained
- Early diagnosis and treatment are crucial to prevent functional impairment and tissue damage

Objectives

- Retrospective case series of 10 adult patients with PRS
- Goals: review the diagnosis, clinical course, and management of PRS

Methods

- Queried MGH Research Patient Database (excludes pediatric patients) using the search words “Parry Romberg Syndrome,” “linear scleroderma,” “PRS,” and “progressive hemifacial atrophy” for patients seen between 2000 and 2021
  - Excluded patients with only linear morphea en coup de sabre (ECDs)
- Demographics, clinical presentation, and relevant imaging, biopsy results, treatment and follow-up were extracted from chart review

Results

<table>
<thead>
<tr>
<th>Pt #</th>
<th>Sex</th>
<th>Race/Ethnicity</th>
<th>Age of Onset (years)</th>
<th>Years from Onset to Diagnosis</th>
<th>Method of Diagnosis</th>
<th>Imaging results</th>
<th>Additional Involvement</th>
<th>Overlapping Linear Morphea</th>
<th>Treatments</th>
<th>Disease course</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>F</td>
<td>NA/Hispanic</td>
<td>30</td>
<td>20</td>
<td>Biopsy/MR</td>
<td>MRI, positive</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
<td>MTX, IVIG, MMF</td>
</tr>
<tr>
<td>2</td>
<td>F</td>
<td>White/Not Hispanic</td>
<td>26</td>
<td>NA</td>
<td>Biopsy/MR</td>
<td>MRI, positive</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
<td>MTX, IVIG, MMF</td>
</tr>
<tr>
<td>3</td>
<td>F</td>
<td>White/Not Hispanic</td>
<td>14</td>
<td>3</td>
<td>NA/MR</td>
<td>MRI, positive</td>
<td>Yes</td>
<td>No</td>
<td>Stable</td>
<td>MTX, IVIG, MMF</td>
</tr>
<tr>
<td>4</td>
<td>F</td>
<td>NA/Hispanic</td>
<td>49</td>
<td>16</td>
<td>NA/Performe[d at NIH]</td>
<td>MRI, positive</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
<td>MTX, IVIG, MMF</td>
</tr>
<tr>
<td>5</td>
<td>F</td>
<td>White/Not Hispanic</td>
<td>36</td>
<td>NA</td>
<td>Biopsy/CT and MR</td>
<td>MRI, positive</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
<td>BB/Telement injections</td>
</tr>
<tr>
<td>6</td>
<td>M</td>
<td>White/Not Hispanic</td>
<td>23</td>
<td>NA</td>
<td>Clinical/MR</td>
<td>MRI, negative</td>
<td>No</td>
<td>No</td>
<td>Fat grafting</td>
<td>Stable</td>
</tr>
<tr>
<td>7</td>
<td>F</td>
<td>Asian/White/Not Hispanic</td>
<td>20</td>
<td>5</td>
<td>Imaging/CT</td>
<td>MRI, positive</td>
<td>No</td>
<td>No</td>
<td>Fat grafting</td>
<td>Stable</td>
</tr>
<tr>
<td>8</td>
<td>F</td>
<td>Other/Not Hispanic</td>
<td>15</td>
<td>NA</td>
<td>Clinical/MR</td>
<td>MRI, positive</td>
<td>Yes</td>
<td>No</td>
<td>Glabellar muscle transfer, MTX, prednisone, colchicine, penicillin, topical vitamin D analog</td>
<td>Stable for years then progressive (with stress and missed medication)</td>
</tr>
<tr>
<td>9</td>
<td>F</td>
<td>White/Not Hispanic</td>
<td>17</td>
<td>NA</td>
<td>Biopsy/Craniofa[cial imaging]</td>
<td>MRI, positive</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
<td>MTX, MMF</td>
</tr>
<tr>
<td>10</td>
<td>F</td>
<td>White/Not Hispanic</td>
<td>43</td>
<td>19</td>
<td>Clinical</td>
<td>MRI, positive</td>
<td>No</td>
<td>No</td>
<td>Prednisone</td>
<td>Stable for 20 years then progressive</td>
</tr>
</tbody>
</table>

- Most patients were female (90%), white (70%), and not Hispanic (80%)
- Average age of onset was 27.3 years
- Significant delay between disease onset and diagnosis: average 12.4 years
- 4 patients had supportive histopathology findings and 7 had positive imaging findings of atrophy (5 MRI, 2 CT, and 1 craniofacial imaging)
- 60% of patients had overlapping linear morphea ECDS
- 3 patients had associated neurologic symptoms (cognitive impairment, seizures, and memory loss)
- 50% of patients reported headaches
- 3 patients had dental involvement and 4 had coexisting ophthalmologic disease (glaucoma, enopthalmos, exotropia, and amblyopia)
- Treatment modalities:
  - Immunosuppressive therapies: primarily methotrexate and prednisone
  - Surgical/cosmetic: primarily fat transfers
- 9 patients had available long-term follow-up, 8 of whom noted improved or stable disease with treatment
  - Of these 8, 5 noted some degree of disease flare during treatment (2 during pregnancy and 3 with missed medication)
- Average available follow-up was 5.9 years

Limitations

- Small cohort size, retrospective single center approach, and relatively short follow-up time available

Conclusions

- In our small cohort, similar to other studies, almost all patients were female and white, with overlapping ECDs, arguing for these conditions existing on a spectrum
- Likely significantly more associated ocular and dental co-existing conditions compared to those reported in the pediatric population
- There is a significant delay in diagnosis of adult patients
- Further studies are needed to understand this rare condition in the adult population

References