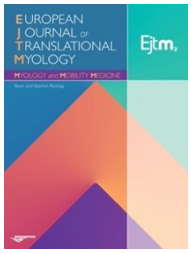
European Journal of Translational Myology



pISSN: 2037-7452 eISSN: 2037-7460

https://www.pagepressjournals.org/index.php/bam/index

Publisher's Disclaimer. E-publishing ahead of print is increasingly important for the rapid dissemination of science. The ***Early Access*** service lets users access peer-reviewed articles well before print / regular issue publication, significantly reducing the time it takes for critical findings to reach the research community.

These articles are searchable and citable by their DOI (Digital Object Identifier).

The **European Journal of Translational Myology** is, therefore, e-publishing PDF files of an early version of manuscripts that undergone a regular peer review and have been accepted for publication, but have not been through the typesetting, pagination and proofreading processes, which may lead to differences between this version and the final one.

The final version of the manuscript will then appear on a regular issue of the journal.

E-publishing of this PDF file has been approved by the authors.

Eur J Transl Myol 2025 [Online ahead of print]

To cite this Article:

Regazzo G, Piccione F, Marenghi M, et al. **Parry-Romberg syndrome associated with ataxia: description of a patient improved after neurorehabilitation.** *Eur J Transl Myol* doi: 10.4081/ejtm.2025.14103

©The Author(s), 2025

Licensee PAGEPress, Italy

Note: The publisher is not responsible for the content or functionality of any supporting information supplied by the authors. Any queries should be directed to the corresponding author for the article.

All claims expressed in this article are solely those of the authors and do not necessarily represent those of their affiliated organizations, or those of the publisher, the editors and the reviewers. Any product that may be evaluated in this article or claim that may be made by its manufacturer is not guaranteed or endorsed by the publisher.

Submitted: 29 June 2025

Accepted: 19 September 2025

Early access: 25 November 2025

Parry-Romberg syndrome associated with ataxia: description of a patient improved after neurorehabilitation

Gianluca Regazzo,[1,2] Francesco Piccione,[1] Marco Marenghi,[1,2] Paola Contessa,[1] Stefano Masiero[1,2]

[1]Department of Neuroscience, Rehabilitation Unit, University of Padua, Padua, Italy; [2]Physical Medicine and Rehabilitation School, Department of Neuroscience, University of Padua, Italy

E-publishing of this typescript was supported by the Armando & Carmela Mioni-Carraro Foundation for Translational Myology, Galleria Duomo 5, 35141 Padua, Italy, and PAGEpress Publications, via A. Cavagna Sangiuliani 5, 27100 Pavia, Italy

Abstract

Parry-Romberg Syndrome (PRS) is a rare condition that can affect the musculoskeletal system, resulting in progressive motor disabilities. The existing literature provides limited evidence regarding physical therapy interventions for PRS. We present a case of a 69-year-old woman diagnosed with PRS with associated ataxia treated with a neurorehabilitative program aimed at improving motor function and balance. We administered various functional scales (SPPB, Tampa Scale of Kinesiophobia, Berg Balance Scale, and Tinetti Scale). Additionally, a standardized gait analysis was conducted in our Gait and Motion Laboratory. Following neurorehabilitative therapy, the patient exhibited significant positive changes in various motor outcomes, as demonstrated by improvements in scores on standardized tests (SPBB, Berg, TSK and Tinetti scales when compared pre- and post-treatment), as well as enhancements in biomechanical parameters and quality of life (FIM, SF-12). This case report suggests that neurorehabilitative therapy can be beneficial in improving motor function and balance in PRS patient, as well as other rare condition associated with disabilities.

Key words: Parry-Romberg Syndrome, neurorehabilitation, ataxia, gait analysis.

Introduction

Parry Romberg Syndrome (PRS) is an idiopathic condition characterized by the atrophy of subcutaneous and muscolo-skeletal tissues and structures.[1] Although the primary characteristic of PRS is the gradual degeneration of facial tissues, the syndrome has been associated with several systemic manifestations, including neurologic, cardiac, rheumatologic, infectious, and endocrine symptoms.[2]

The incidence of PRS ranges from 0.3 to 2.5 cases per 100,000 individuals per year, and seems to affect mostly males. The condition is characterized by a progressive course, and the onset typically occurs within the first two decades of life.[3,4]

The pathogenesis of PRS is heterogeneous, and is currently seen as a multifactorial condition, with multiple factors contributing to the development of the disease. Familial PRS has been reported, but no clear pattern of inheritance or specific genetic defect has been found.[5] Several etiopathogenetic theories have been proposed, including trauma, autoimmune responses, infections, vasculitis and malformations. Histopathological examinations of patient's brain have revealed lymphocytic vasculitis, gliosis, and sclerosis of the leptomeninges, suggesting an underlying inflammatory process.[6]

A variety of neurological symptoms have been reported in these patients, including movement disorders, cognitive impairments and behavioral changes.[7] These symptoms have been associated with several MRI findings, the most common of which are ipsilateral T2 hyperintensities, primarily observed in the white matter, gray matter, and corpus callosum. Additional findings include leptomeningeal enhancement, calcifications, cerebral atrophy, intracranial vascular malformations, focal infarctions in the corpus callosum, infarcts in the amygdaloid body and lesions in the meninges and basal ganglia. While neuroimaging findings in patients with PRS are more often ipsilateral, contralateral findings have also been documented.[8]

The treatment of PRS is multidisciplinary and involves plastic surgeons, ENT specialists, neurologists and often psychiatrists. Currently, there are several alternative available for the correction of deformities, like cosmetic surgeries utilizing autologous fat grafts, silicone or bovine collagen injections.[9] In addition to aesthetic improvement, medical therapies such as steroid therapy, methotrexate, azathioprine, and cyclophosphamide are also indicated.

As for the neuromotor symptoms, there is limited evidence in the literature regarding the effectiveness of neurorehabilitation programs for PRS patients with associated neurological disabilities. Studies have mostly focused on the treatment of the acute phase of the disease, and there is little knowledge about long-term management and rehabilitation of these patients.

In this case report, we present a personalized rehabilitation program designed to restore balance and walking function in a patient with a rare ataxic presentation, the third case documented in the literature to our knowledge. While a previous article demonstrated improvement in a patient with PRS and ataxia through the use of oral prednisolone, this is the first description to date of neurorehabilitative interventions for this specific population.[10]

Materials and Methods

The patient is a 69-year-old woman with a history of right unilateral facial atrophy since childhood that began in 2019 to experience episodes of postural instability, ataxia and a progressive reduction on muscle strenght in the left lower limb. In 2024, she was admitted to the neurology ward in Padua for further investigation due to the persistence of her symptoms. Genetic testing (SCA1, SCA2, SCA3, SCA6, SCA7, SCA8, SCA12, and SCA17) ruled out the presence of genetic spinocerebellar ataxia, and no MRI evidence of lumbar stenosis was found. However, there was an alteration in the PEM/PESS evaluation, with a significant change in central conduction time, and brain imaging revealed the presence of right pontine hypotrophy (Figure 1). The patient was discharged with a diagnosis of Parry-Romberg syndrome associated with ataxic symptoms and was referred to Neurorehabilitation Unit in Padova to improve her functional mobility and quality of life. There, she began a rehabilitation program focused on improvement of ataxia and gait retraining.

The clinical evaluation confirmed the presence of ataxic gait in the absence of additional cerebellar signs, associated with hypotrophy of the left lower limb, particularly pronounced in the anterior tibialis and gastrocnemius areas. Patient reported using assistive devices for ambulation in her daily life and exhibited significant kinesiophobia, a fear of movement caused by pain or risk of falling. Instrumental evaluation, including surface electromyography and a biomechanical assessment of walking patterns, was conducted in our Gait Analysis laboratory, and standardized scales were administered to evaluate motor function (Motricity Index, [MI]), balance (Short Physical Performance Battery [SPPB], Tampa Scale of Kinesiophobia [TSK], Berg Balance Scale, Tinetti Scale), and quality of life (Short form Health Survey-12 [SF-12], Functional Independence Measure [FIM]) (Figure 2).

The results indicated a strength deficit in the lower left limb (MI: 83/100), accompanied by a compromise in balance (SPPB: 2/12, Berg Balance Scale: 36/56, and Tinetti Scale: 18/28) and significant kinesiophobia, particularly during walking (TSK: 43/68). These findings were associated with a moderate reduction in indipendence in daily activities (FIM: 118/126) and quality of life (SF-12: PCS-12 [Physical Score] 28.5, MCS-12 [Mental Score] 41.2). Electromyography (EMG) revealed a reduction in muscle activity in the left leg, particularly in the gastrocnemius and anterior tibialis muscles, while kinematic analysis of gait demonstrated reduced speed, an increased support base and a diminished joint excursions at the hip, knee, and ankle. Finally, stabilometric and baropodometric assessments indicated anteroposterior oscillations and a medial progression of the center of pressure.

Based on initial assessment we proposed a treatment focused on ataxic symptoms and gait instability, consisting of 20 physical therapy sessions (three times per week) that included exercises for strengthening, proprioception, coordination, neuromuscular reeducation, and gait training, with the ultimate goal of enhancing functional mobility.

In the first phase of the treatment (sessions 1-3), the patient underwent strengthening exercises to enhance postural control and pelvic stability, as well as resistance exercises for the left lower limb, primarily targeting the anterior tibialis and gastrocnemius muscles. In the next sessions,[4-10] the Physical Therapist (PT) focused on proprioception, balance, and coordination. For proprioception training, the PT utilized exercises such as seated weight shifts (shifting weight side to side while seated to enhance postural control), gentle trunk rotations (gradually rotating the upper body to improve coordination), and standing on a proprioceptive foam pad. To enhance balance and postural control, static balance exercises were used, including tandem stance (heel-to-toe stance) and single-leg stance activities, gradually combined with dynamic balance training focused on weight transfers (deliberately shifting weight from side to side while standing), marching, and obstacle walking. For coordination and motor control, the PT used several exercises, including repetitions of finger-to-nose, heel-to-shin, and reciprocal arm and leg movements (marching and cycling movements in the air). In the final phase of the treatment (sessions 11-20), the patient underwent interventions aimed at improving motor control and functional mobility of the lower limbs. These included slow, controlled step walking emphasizing a heel-to-toe gait pattern, dual-task training (walking while simple counting or cognitive tasks), fall prevention strategies and gait training with and without aids (sit-to-stand transitions, side-stepping, backward walking, and practicing turns).

Results

At the conclusion of the treatment, the patient was able to walk short distances and climb stairs in both directions without the use of an assistive device. Clinical assessments indicated improvements in balance (SPPB: 6/12, Berg Scale: 45/56, Tinetti Scale: 22/28) and kinesiofobia (TSK: 34/68), along with positive effects on functional indipendence (FIM: 124/126) and quality of life (PCS-12: 31.8, MCS-12: 44) (Table 1).

Gait analysis following the treatment revealed an increase in gait speed and joint excursions, accompanied by a reduction in left knee recurvatum. Additionally, stabilometric and baropodometric assessments indicated a decrease in oscillations and a lateral shift in the center of pressure (CoP). Since the treatment primarily targeted balance and ataxia, only a minimal improvement in muscle activation was observed in EMG and no improvement was showed ine MI scale (MI: 83/100) (Table 2).

Discussion

Parry-Romberg Syndrome (PRS) is an uncommon disorder characterized by the progressive atrophy of facial tissues, which may be associated with various systemic symptoms that can lead to functional limitations and disabilities. It typically manifests during the first or second decade of life; however, a few cases of late onset have been documented.[11]

About 15-20% of patients with PRS develop neurological symptoms, including headaches, trigeminal neuralgia, hemiparesis, cognitive deficits, nerve palsy, and, in severe cases, death due to brainstem involvement. Brain MRI abnormalities are observed in 60% to 74.6% of patients, with the most common findings being white matter disease, hemispheric atrophy, leptomeningeal enhancement, and microhemorrhages.[12] As there is no definitive cure for PRS, the therapeutic approach focuses on controlling disease progression, alleviating symptoms, and enhancing the patient's aesthetic appearance. Symptomatic treatment may include pain relievers and anticonvulsants to manage pain and seizures, corticosteroids to control the active phase of the disease and reconstructive and orthognathic surgery.[13]

Despite the chronic nature of the condition and the disabilities associated with PRS there is limited evidence regarding the role of rehabilitation. However, it can be speculated that rehabilitation may play a key role in preserving residual autonomies and developing adaptive compensations.

In our case report, we described a neurorehabilitative intervention for a patient with PRS associated with a rare neurological symptom, an unilateral ataxia. To our knowledge, only two cases of PRS

with ataxia have been previously reported, and this is the first case in which a rehabilitative intervention has been documented.

Ataxia, characterized by impaired motor coordination and balance, is a strong predictor of falls. Falls are a primary cause of morbidity and functional decline with several consequences on individuals health. The direct health impacts include a spectrum of physical injuries, from soft-tissue damage to severe hip fractures and traumatic brain injuries, which are linked to increased mortality. A critical psychological sequela is the "fear of falling," a condition that promotes activity avoidance, leading to physical deconditioning and social isolation, as reported by our patient. Cumulatively, these events often trigger a loss of independence and necessitate long-term institutional care, making it a critical target for rehabilitation.

We implemented a personalized therapeutic exercise program focused on the ataxic component, resulting in improvements in the patient's balance, kinesiophobia, and biomechanical parameters of walking, as assessed through gait analysis and surface electromyography. With the limitation of being a case report, our work shows that neurorehabilitation interventions can be effective in patient with PRS syndrome and should be offered to all patient with associated neuromotor involvement.

This result highlights the need to extend functional assessment and rehabilitative interventions to rare conditions with uncommon neurological symptoms. While there are several challenges in managing these conditions, such as variability in disease progression and late diagnoses, early monitoring of disabilities and the development of symptom-specific strategies can significantly enhance the quality of life for patients. The integration of new health and digital technologies, as wearable sensors and virtual reality, could help to improve the management of ataxia and fall risks by shifting from subjective, in-clinic assessments to objective, continuous monitoring and personalized rehabilitation.

Conclusions

In this study, we present the case of a woman experiencing a rare manifestation of Parry-Romberg syndrome, with a significant impact on her balance and walking abilities. We demonstrate that neurorehabilitation can enhance function in patients with rare diseases such as PRS, thanks to several treatment options that can be beneficial for these individuals, including physical therapy, occupational therapy, botulinum toxin injections, pain management, and cognitive training.

Further work is needed to systematically develop treatments aimed at improving the quality of life for patients with rare diseases. We hope that the scientific and political communities will make significant efforts to ensure rehabilitation for conditions that currently lack effective therapeutic and rehabilitative options.

Abbreviations

PRS (Parry-Romberg Syndrome), TSK (Tampa Scale of Kinesiophobia), SPPB (Short Physical Performance Battery), FIM (Functional Indipendence Measure), SF-12 (12-Item Short Form Survey), SCA (Spinocerebellar Ataxia), MI (Motricity Index), PT (Physical therapist)

Correspondence

Gianluca Regazzo, Department of Neuroscience, Rehabilitation Unit, University of Padua, Via Giustiniani, 2, 35128 Padova, Italia.
gianlucareg@gmail.com
0009-0004-6277-4199

Francesco Piccione

francesco.piccione@aopd.veneto.it

0000-0002-0173-903X

Marenghi Marco

marco.marenghi@studenti.unipd.it

0009-0007-9217-863X

Paola Contessa

paola.contessa@aopd.veneto.it

0000-0002-8645-6783

Stefano Masiero

stef.masiero@unipd.it

0000-0002-0361-4898

Contributions

FP, GR, study concept and design; GR, MM, PC, data acquisition; FP, GR, PC, data analysis and interpretation; GR, drafting of the manuscript; FP, SM, critical revision; PC, statistical analysis; SM, administrative, technical, and material support, supervision.

Conflict of interest

The authors have no conflict of interest to declare.

Ethics approval and consent to participate

No ethical committee approval was required for this case report by the Department, because this article does not contain any studies with human participants or animals. Informed consent was obtained from the patient included in this study.

Patient consent for publication

The patient gave her written consent to use her personal data for the publication of this case report and any accompanying images.

Availability of data and materials

All data underlying the findings are fully available.

References

1. Schultz KP, Dong E, Truong TA, Maricevich RS. Parry Romberg Syndrome. Clin Plast Surg 2019;46:231-7.
2. Sudarshan R, Annigeri RG. Parry Romberg syndrome. Wien Klin Wochenschr 2010;122:606.
3. Patel H, Thakkar C, Patel K. Parry-romberg syndrome: a rare entity. J Maxillofac Oral Surg 2010;9:247-50.
4. Van der Cruyssen F, Meeus J, Schoenaers J, et al. Parry-Romberg syndrome: a mini review. Acta Dermatovenerol Alp Pannonica Adriat 2020;29:193-9.
5. Yu BF, Dong LP, Dai CC, Wei J. Genetic variations in patient with Parry-Romberg syndrome. Sci Rep 2023;13:400.
6. Wong M, Phillips CD, Hagiwara M, Shatzkes DR. Parry Romberg Syndrome: 7 Cases and Literature Review. AJNR Am J Neuroradiol 2015;36:1355-61.
7. Kumar M, Singla R, Singh G, et al. Parry Romberg Syndrome: A Case Report and an Insight Into Etiology. Cureus 2023;15:e41465.
8. Budrewicz S, Koszewicz M, Koziorowska-Gawron E, et al. Parry-Romberg syndrome: clinical, electrophysiological and neuroimaging correlations. Neurol Sci 2012;33:423-7.
9. Parry Romberg syndrome: A long-term retrospective cohort study of 10 patients. Oral Maxillof Surg Cases 2018;4:73-83.
10. Lee YJ, Chung KY, Kang HC, et al. Parry-Romberg syndrome with ipsilateral hemipons involvement presenting as monoplegic ataxia. Korean J Pediatr 2015;58:354-7.
11. Sharma PK, Sekar A, Amir AP, Prabhu ALR. Parry-Romberg syndrome: A case report and literature review. Radiol Case Rep 2024;19:2230-8.
12. Aydın H, Yologlu Z, Sargın H, Metin MR. Parry-Romberg syndrome. Physical, clinical, and imaging features. Neurosciences (Riyadh) 2015;20:368-71.
13. de Vasconcelos Carvalho M, do Nascimento GJ, Andrade E, et al. Association of aesthetic and orthodontic treatment in Parry-Romberg syndrome. J Craniofac Surg 2010;21:436-9.

14. Lee YJ, Chung KY, Kang HC, et al. Parry-Romberg syndrome with ipsilateral hemipons involvement presenting as monoplegic ataxia. Korean J Pediatr 2015;58:354-7.

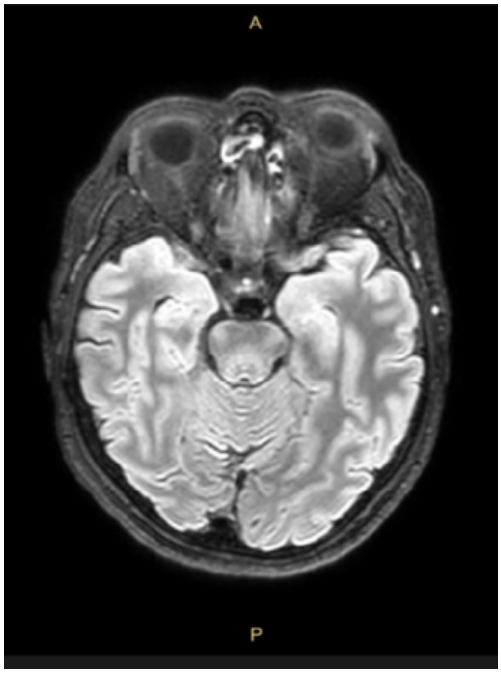


Figure 1. MRI of the patient, showing pontine hypotrophy compatible with the clinical findings of right hemifacial emiatrophia and left ataxia.

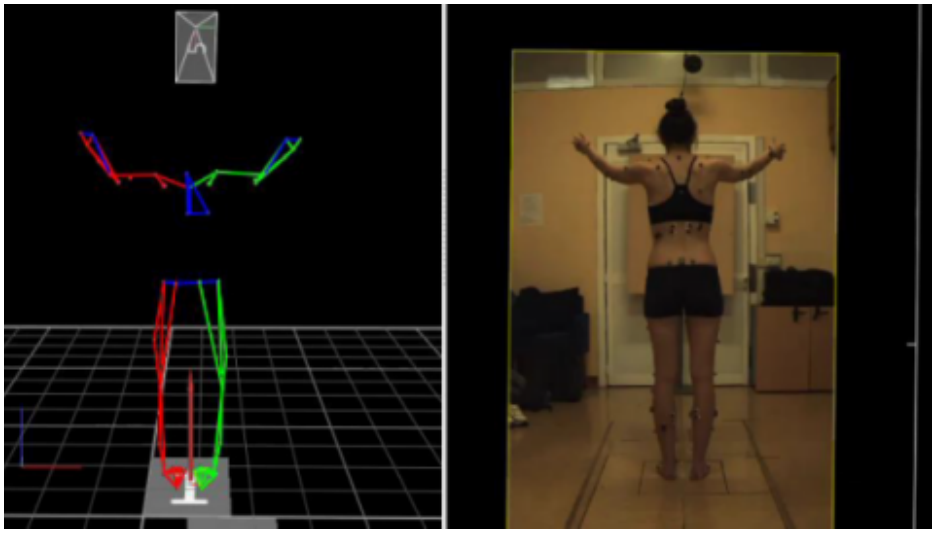


Figure 2. Gait Analysis of the patient in our Laboratory.

Table 1. Comparison of balance and motor function pre and post treatment (MI: Motricity Index; SPPB: Short Physical Performance Battery, TSK: Tampa Scale of Kinesophobia, SF-12: Short form Health Survey, PCS-12: Physical Score, MCS-12: Mental Score, FIM: Functional Indipendence Measure).

	Pre-treatment	**Post-treatment**
MI	83/100	83/100
Tinetti Scale	18/28	22/28
Berg Scale	36/56	45/56
SPPB	2/12	6/12
TSK	43/68	34/68
FIM	118/126	124/126
SF-12	PCS-12: 28.5, MCS-12: 41.2	PCS-12: 31.8, MCS-12: 44

Table 2. Improvement in Gait Analysis and sEMG post-treatment compared to pre-treatment evaluations.

Spatiotemporal parameters	Slight increase in walking speed and cadence was observed
Kinematics	At the knee level, a slight improvement on the right, with the presence, albeit minimal, of the load acceptance peak. On the left, improvement in the range of motion during the stance phase
Kinetics and	Increase in moments and powers
Stabilometry	Slight reduction in oscillations with eyes closed
Baropodometric examination	Decrease in pressures in the left hindfoot
sEMG	Improvement in muscle activation