

Patient Journeys are **info-graphical overviews** that visualize patients' needs in the care of their rare disease.

Because Patient Journeys are designed from the **patient's perspective**, they allow clinicians to **effectively address the needs** of rare disease patients.

Download this patient journey on our website.

Area	First symptoms	Diagnosis	Treatment	Monitoring
Disease	<ul style="list-style-type: none"> First symptoms are usually in the lower extremities, starting with stiffness and cramps, followed by weakness and spasticity. Progressive spastic paraparesis, with increasing stiffness and weakness, leading to difficulty walking and eventually to the need for a walking aid or wheelchair. Bladder and bowel dysfunction, often manifesting as urinary urgency, frequency, and incontinence, as well as constipation. Sexual dysfunction, including erectile dysfunction in men and decreased libido in both sexes. Weight loss and fatigue, which can significantly impact quality of life. 	<ul style="list-style-type: none"> Diagnosis is often delayed, as symptoms are non-specific and may be mistaken for other conditions like multiple sclerosis or vitamin B12 deficiency. Diagnosis is typically confirmed through a combination of clinical history, physical examination, and genetic testing. Genetic testing is crucial for identifying the specific HSP subtype, which guides prognosis and management. Some patients may also undergo MRI to rule out other neurological conditions. 	<ul style="list-style-type: none"> Treatment is primarily symptomatic and supportive, aiming to improve quality of life and manage complications. Physical therapy and occupational therapy are essential for maintaining mobility and independence. Medications like baclofen or tizanidine may be used to manage spasticity, while laxatives and catheters help with bowel and bladder issues. Assistive devices, such as walking aids or wheelchairs, are often necessary as the disease progresses. Psychological support and counseling are important for addressing the emotional and social challenges of living with a chronic condition. 	<ul style="list-style-type: none"> Monitoring involves regular clinical follow-ups to assess the progression of the disease and the effectiveness of treatments. Patients may also undergo periodic genetic testing to monitor for any changes in their genetic profile. Research and clinical trials are ongoing, offering hope for more targeted and effective treatments in the future.
Obstacles	<ul style="list-style-type: none"> Lack of awareness and knowledge about HSP among healthcare providers and the general public. Delayed diagnosis due to non-specific initial symptoms. Limited access to specialized care and genetic testing, particularly in rural areas. Financial barriers to accessing necessary treatments and assistive devices. Emotional and psychological challenges, including isolation and a sense of loss. 	<ul style="list-style-type: none"> Fragmented healthcare system with poor coordination between different specialists. Lack of standardized diagnostic criteria and procedures. Limited availability of genetic testing and counseling services. Insufficient research funding for HSP, leading to a lack of new treatments. 	<ul style="list-style-type: none"> Over-reliance on symptomatic treatment without addressing the underlying genetic cause. Lack of multidisciplinary care teams that integrate physical, occupational, and psychological therapies. Limited patient involvement in decision-making about their care. 	<ul style="list-style-type: none"> Lack of long-term monitoring and data collection to better understand the disease's progression. Limited patient education and support resources. Barriers to participation in clinical research and trials.
Challenges	<ul style="list-style-type: none"> Early and reliable diagnosis remains a significant challenge. Developing effective therapies that target the underlying genetic defect. Improving patient access to specialized care and genetic testing. Enhancing patient education and support resources. Facilitating patient participation in research and clinical trials. 	<ul style="list-style-type: none"> Establishing a robust infrastructure for genetic testing and counseling. Developing standardized diagnostic and treatment protocols. Improving coordination and communication between healthcare providers. Increasing research funding and collaboration between scientists. 	<ul style="list-style-type: none"> Developing novel therapeutic approaches, such as gene therapy or CRISPR-based editing. Implementing a patient-centered care model that prioritizes quality of life. Establishing a strong patient advocacy and support network. 	<ul style="list-style-type: none"> Establishing a comprehensive long-term monitoring system. Developing patient education and support resources. Facilitating patient participation in research and clinical trials.



PATIENT JOURNEY

Hereditary Spastic Paraplegias (HSPs)

different needs
at different times



Was this patient journey helpful?
Help us improve patient care
and participate in our short survey!



European Reference Network
for Rare Neurological Diseases
Coordinator: Dr. Holm Graessner
University Hospital Tübingen

Centre for Rare Diseases
Calwerstr. 7 | 72076 Tübingen | Germany
www.ern-rnd.eu | info@ern-rnd.eu



European
Reference
Network

for rare or low prevalence
complex diseases



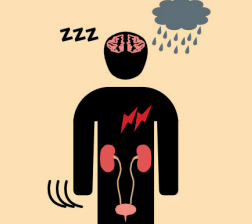
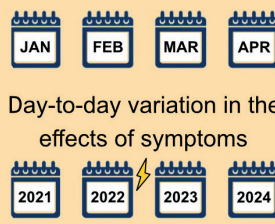



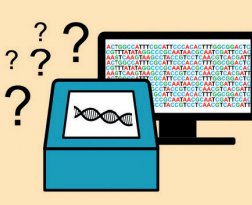
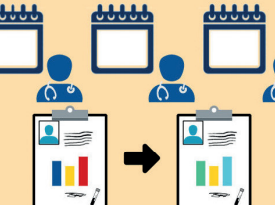
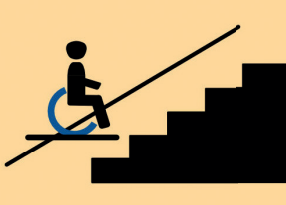

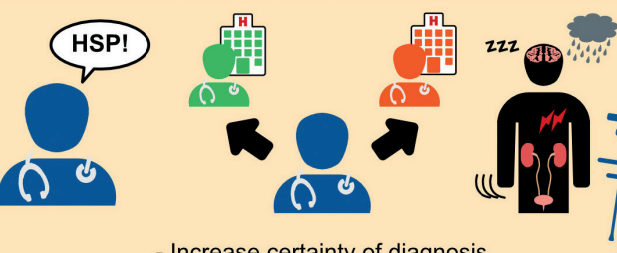





 **Network**
Neurological Diseases
(ERN-RND)



Euro-HSP
Federation of European HSP Associations



HEREDITARY SPASTIC PARAPLEGIA
Taking Steps Toward a Cure

	First symptoms	Diagnosis		Treatment	Monitoring
Disease	 <p>Childhood Age 30 - 50</p> <p>Early symptoms are often unspecific</p>	 <p>90+ different disease types. Misdiagnosis is common</p>	 <p>Possible symptoms: fatigue, urinary issues, pain, depression, spasms, cognitive problems</p>	 <p>Day-to-day variation in the effects of symptoms</p> <p>Slow progression of symptoms. New symptoms can develop</p>	 <p>Understand how to accept life with HSP</p>
Clinic	 <p>Early symptoms in people with HSP can include balance issues and tripping</p>	 <p>Clinical diagnosis after excluding other conditions</p>	 <p>Genetic diagnosis might be inconclusive</p>	 <p>Regular follow-up. Personalized plan changes over time with progression</p>	 <p>Plan to consider: future generations; changes at work; modifications at home</p>
Challenges	 <p>Knowledge of HSP is low in many healthcare professionals</p>	 <ul style="list-style-type: none">- Increase certainty of diagnosis- Referral of people with HSP to different expert centres- Awareness and prediction of all HSP-aspects		 <p>No cure for HSP, only symptomatic treatment available. Research & clinical trials needed</p>	 <p>Not all people with HSP want to plan. Need for personalized support</p>
Goals	 <ul style="list-style-type: none">- Clinicians should be able diagnose HSP and know experts to refer people with HSP to- Support for people with HSP after diagnosis, including physiotherapy and stretching- Providing people with HSP with information and treatment options			 <p>Get people with HSP to maintain a routine with physical activity. Best quality of life possible.</p>	 <p>Providing information about support networks; current research work; patient registries</p>

HSP Hereditary Spastic Paraplegias

Please note that specific terms (e.g. home care services, general physician, physiotherapy) do not include the same services in all EU countries and might differ from country to country. Patient advocacy groups can often provide support and resources for patients and families.

Disclaimer

ERN-RND specifically disclaims any warranties of merchantability or fitness for a particular use or purpose. ERN-RND assumes no responsibility for any injury or damage to persons or property arising out of or related to any use of this information or for any errors or omissions.

Updated on November 2022.

