PHASES	First Symptoms	Diagnosis		Treatment	Monitoring
Disease	• Early symptoms are not very specific. They may include difficulties in walking or running, stiff leg muscles, or difficulties controlling leg movements. • Symptoms can start at any age, but most often they start either in childhood or in adults aged 30-50.	**HSP diagnosis** • HSP is an umbrella for many different disease types. More than 90 types have been identified, and this number increases each year. Different types of HSP have different inheritance patterns, ages of onset, symptoms and rates of progression. • Misdiagnosis is common. HSP has been mis-diagnosed as cerebral palsy, multiple sclerosis, ataxia, arthritis, muscular dystrophy, other types of motor neuron disease and many other conditions.	**Non-motor symptoms** • Other symptoms often include: urinary or bowel problems, pain, depression, fatigue, spasms, and skin problems. • Symptoms of less common types of HSP can include: learning difficulties, speech/hearing/vision problems, dementia, ataxia, and upper body involvement. • Non HSP factors can be important (depression, isolation, loneliness etc.) but are often not discussed at diagnosis or later doctor appointments.	• Symptoms usually progress slowly over a number of years and new symptoms can develop over time. Day-to-day variation in the effects of symptoms can be larger than the year-on-year progression of HSP. • Physiotherapy and/or stretches are important. Depending on symptoms, treatments for spasticity, pain, bladder/bowel problems and depression are available. People with HSP may require mobility aids which change over time.	• Big life changes in employment or the home may be needed as a result of HSP. • At home, people with HSP may need to have ramps or grab-rails fitted. Should people require a wheelchair they may need bathrooms converting to wet-rooms, aids to help get up/down stairs, aids to help transfer between chairs and to/from bed. They may also need modificaitons in the kitchen. • At work, people may not be able to carry on with their roles as their HSP progresses. They may require adjustments to allow them to continue. Ultimately they may need to change roles or career to maintain their income.
Clinic	**Range of (inconclusive) symptoms** • In clinic symptoms that are often seen in people at the start of their HSP journies are walking, tripping and/or balance issues. People with HSP may have fatigue, pain, bladder issues or low mood and not realise these could be HSP symptoms. • Investigations are needed to confirm the diagnosis. The European Reference Network has produced a diagnostic flow chart for HSP. https://www.ern-rnd.eu/wp-content/uploads/2019/02/ERN-RND-Diagnostic-Flowchart-HSP_final-1.pdf	• A clinical diagnosis of HSP can be given after excluding a range of other conditions. Clinical diagnosis is likely to involve a range of tests and/or MRI scanning of brain and spine. • Genetic diagnosis for HSP is available, but results might be inconclusive. Whilst reliable genetic testing is available for the majority of the more common types of HSP, the main issues around testing results are that: - there may not be a genetic test for that type of HSP - there may not be evidence that any mutation found is associated with HSP - there is a 30-40% diagnostic gap in genetic testing If genetic diagnosis is achieved, the process is straightforward. Family members should be offered counseling. Predictive testing may be offered. • There may be clinical and/or genetic overlap with other conditions.		**Personalised management of motor and non-motor symptoms** •Referral of people with HSP to different expert centres according to their needs and symptoms. • The plan will need to change over time as the condition progresses and symptoms change. • The range of specialists will depend on the specific motor and non-motor symptoms, and may include: auxiliary support for deambulation; orthopedic assessment for secondary spine and foot deformities. • People with HSP are likely to need advice on controlling pain, managing fatigue, improving wellbeing and dealing with bladder/bowel incontinence/urgency.	**Personalised follow-up** • Whilst there are some general patters with speciic types of HSP or for specific age groups, the prediction of HSP progression for an individual is difficult. It is challenging to be able to answer questions like "when will I need a wheelchair?". • Regular follow-up is helpful and there should be personalised adaption of management plans, particularly as symptoms change over time. • Plans will need support and buy-in from the person with HSP, and they will need to be able to understand how making decisions on symptom treatment/management can help them in the future.

<table>
<tr><td rowspan="2">Challenges</td><td>Early and reliable diagnosis

• The knowledge of HSP is low in many healthcare professionals, and getting a confirmed diagnosis can include seeing several specliaists. The uncertainty of diagnosis can affect wellbeing.</td><td colspan="2">Physicians considering aspects outside their expertise

• Certainty of diagnosis needs to be increased to improve diagnosis time and reduce mis-diagnosis.

• After diagnosis, experts should be aware of all aspects of HSP - both direct mobility and non-mobility symptoms, and other impacts on wellbeing.

• Referral to different expert centres according to needs and symptoms should be routine so that people with HSP can get co-ordinated care.

• The need for a multi-disciplinary team depends on which symptoms occur over time. Specialists near time of diagnosis are likely to include: neurologist, geneticist, radiologist for MRI or other scans. After diagnosis, for mobility issues people may need: physiotherapist, orthotics, rehabilitation, mobility equipment, falls and balance problem specialists.

• Physicians should be able to help people with HSP understand the information given by specialists, and interpret what it means for them.

• Many people with HSP have to repeat their story to each new specialist, information transfer between specialists should be improved.</td><td>Finding a cure
• There is no cure for HSP, all treatment is symptomatic.

• Current research work in HSP includes looking at biomarkers, considering celluar and animal models of HSP, identifying potential treatment drugs, and increasing the understanding of HSP.

• Further research efforts and clinical trials are needed.</td><td>Personalised support
• Some people with HSP want to plan for their future, but others do not. There is a need for personalised support based on peoples needs and wishes.

• Those who do not wish to plan may benefit from seeing a counsellor to help them with the acceptance of their HSP - they may be affected by grief for the loss of their former selves or anger about what they feel unable to do in the future.

• Those with HSP who wish to have a family are likely to benefit from seeing a geneticist to understand risks and consider options for IVF and/or prenatal testing.</td></tr>
<tr><td>Goals</td><td colspan="2">Awareness and education about HSP (and other similar neurological conditions) in relevant healthcare professionals

• Clincians should be able to diagnose HSP - or refer to an appropriate specialist to do so. Clinicians should know experts to refer people with HSP to - this is likely to include a neurologist, a physiotherapist, and orthotist. Other specialists may be needed if other symptoms are present.

• Given there is no cure for HSP, psychological support for people with HSP after diagnosis may be important

• Given that HSP is a rare disease, it can feel isolating and lonely feeling like the person is the only one with HSP. Connections with relevant communities can be important in reducing these feelings.

• It is difficult to find information about HSP. So, it is important to provide people with relevant information about options for treatment of symptoms, places to get advice, and genetic diagnosis.</td><td>Living with HSP

• People with HSP may meed help developing and maintaining a routine with appropriate levels of physical activity so that they can have the best quality of life possible.

• Beyond a healthcare routine with appropriate physiotherapy and medication, routines can include aspects on wellbeing, social connections and understanding the wider HSP world.</td><td>Patient empowerment

• Information about HSP can be hard to find, and telling people about three areas can help release information:
- information about support networks can provide access to others with HSP, which can help with solutions to everyday problems and to help feel less alone and isolated.
- an understanding about current research work can help people see the wider picture and get reassurance that there are many professionals working on HSP
- information about patient registries can allow people to take part in research projects and keep up to date about specific news for their types of HSP.</td></tr>
</table>