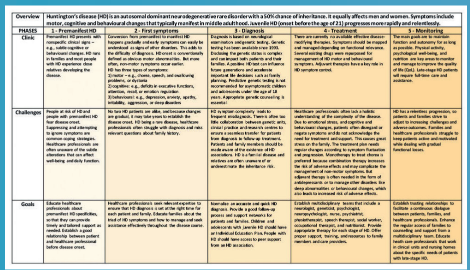
Patient Journeys are **info-graphical overviews** that visualize patients' needs in the care of their rare disease.

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

Find a detailed version of this patient journey on our website.



Overview	Huntington's disease (HD) is an autosomal dominant neurodegenerative rare disorder with a 50% chance of inheritance. It equally affects men and women. Symptoms include motor, cognitive and behavioural changes that typically manifest in middle adulthood. Juvenile HD (onset before the age of 21) progresses more rapidly and relentlessly.				
PHASES	**1 - Premanifest HD**	**2 - First symptoms**	**3 - Diagnosis**	**4 - Treatment**	**5 - Monitoring**
Clinic	Premanifest HD presents with nonspecific clinical signs – e.g., subtle cognitive or behavioural changes. HD runs in families and most people with HD experience close relatives developing the disease.	Conversion from premanifest to manifest HD happens gradually and early symptoms can easily be understood as signs of other disorders. This adds to the difficulty of diagnosis. HD onset is conventionally defined as obvious motor abnormalities. But more often, non-motor symptoms occur earlier. HD has three types of symptoms: 1) motor – e.g., chorea, speech, and swallowing problems, or dystonia 2) cognitive: e.g., deficits in executive functions, attention, recall, or emotion regulation 3) behavioural: e.g., depression, anxiety, apathy, irritability, aggression, or sleep disorders	Diagnosis is based on neurological examination and genetic testing. Genetic testing has been available since 1993. Disclosing the genetic status is complex and can impact both patients and their families. A positive HD test can influence future generations and accelerate important life decisions such as family planning. Predictive genetic testing is not recommended for asymptomatic children and adolescents under the age of 18 years. Appropriate genetic counselling is essential.	There are currently no available effective disease-modifying therapies. Symptoms should be mapped and managed depending on functional relevance. Several existing drugs were repurposed for management of HD motor and behavioural symptoms. Adjuvant therapies have a key role in HD symptom control.	The main goals are to maintain function and autonomy for as long as possible. Physical activity, psychological well-being, and nutrition are key areas to monitor and manage to improve the quality of life (QoL). Late-stage HD patients will require full-time care and assistance.
Challenges	People at risk of HD and people with premanifest HD fear disease onset. Suppressing and attempting to ignore symptoms are common coping strategies. Healthcare professionals are often unaware of the subtle alterations that can affect well-being and daily function.	No two HD patients are alike, and because changes are gradual, it may take years to establish the disease onset. HD being a rare disease, healthcare professionals often struggle with diagnosis and miss relevant questions about family history.	HD symptom complexity leads to frequent misdiagnosis. There is often too little collaboration between genetic units, clinical practice and research centres to ensure a seamless transfer for patients from diagnosis to follow-up treatment. Patients and family members should be made aware of the existence of HD associations. HD is a familial disease and relatives are often unaware of or underestimate the inheritance risk.	Healthcare professionals often lack a holistic understanding of the complexity of the disease. Due to emotional stress, and cognitive and behavioural changes, patients often disregard or negate symptoms and do not acknowledge the need for treatment and support. This causes great stress on the family. The treatment plan needs regular changes according to symptom fluctuation and progression. Monotherapy to treat chorea is preferred because combination therapy increases the risk of adverse effects and may complicate the management of non-motor symptoms. But adjuvant therapy is often needed in the form of antidepressants or to manage other disorders like sleep abnormalities or behavioural changes, which also leads to increased risk of adverse effects.	HD has a relentless progression, so patients and families strive to adjust to increasing challenges and adverse outcomes. Families and healthcare professionals struggle to keep patients active and motivated while dealing with gradual functional losses.
Goals	Educate healthcare professionals about premanifest HD specificities, so that they can provide timely and tailored support as needed. Establish a good relationship between patient and healthcare professional before disease onset.	Healthcare professionals seek relevant expertise to ensure that HD diagnosis is set at the right time for each patient and family. Educate families about the triad of HD symptoms and how to manage and seek assistance effectively throughout the disease course.	Normalize an accurate and quick HD diagnosis. Provide a good follow-up process and support networks for patients and families. Children and adolescents with juvenile HD should have an Individual Education Plan. People with HD should have access to peer support from an HD association.	Establish multidisciplinary teams that include a neurologist, geneticist, psychologist, neuropsychologist, nurse, psychiatrist, physiotherapist, speech therapist, social worker, occupational therapist, and nutritionist. Provide appropriate therapy for each stage of HD. Offer proper support, training, and resources to family members and care providers.	Establish trusting relationships to facilitate a continuous dialogue between patients, families, and healthcare professionals. Enhance the regular access of families to counselling and support from a multidisciplinary team. Educate heath care professionals that work in clinical units and nursing homes about the specific needs of patients with late-stage HD.

PATIENT JOURNEY

Huntington's Disease

different needs
at different times

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

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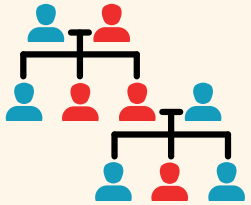
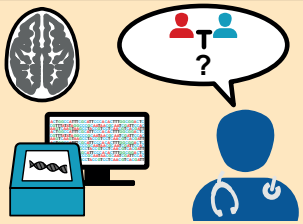
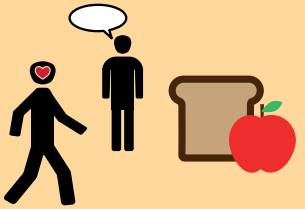
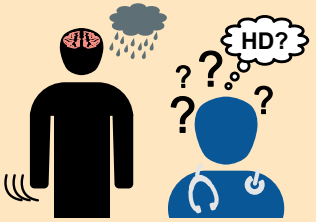
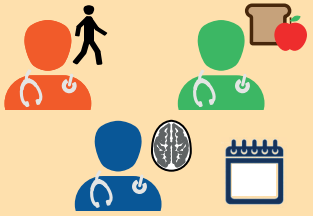
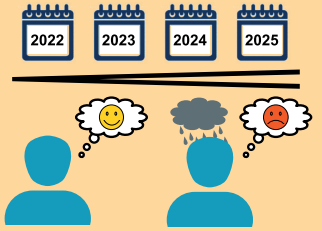
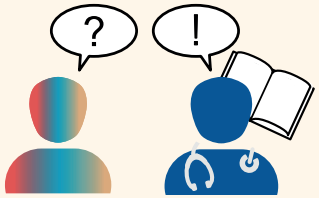
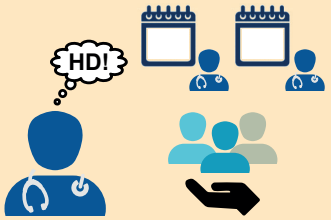
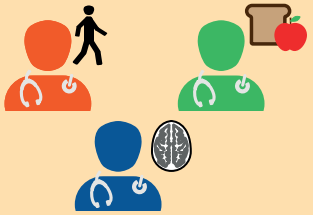
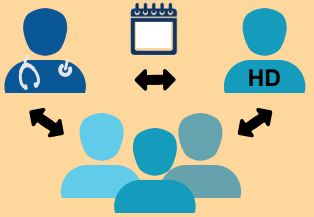
European Reference Network
for rare or low prevalence complex diseases

Network
Neurological Diseases
(ERN-RND)

EUROPEAN Huntington Association

	Premanifest HD	First Symptoms	Diagnosis	Treatment	Monitoring
Clinic	 Most people with HD experience several close relatives developing HD.	 Subtle and non-specific first symptoms fall in 3 categories: motor, cognitive, and behavioural.	 Confirm clinical diagnosis with genetic testing. Genetic counselling is essential.	 No disease modifying treatment. Symptoms managed and treated to maintain functionality and QoL.	 Physical activity, psychological wellbeing and nutrition maintain function and autonomy.
Challenges	 Fear of disease onset leads to ignoring symptoms.	 Differences in disease onset and first symptoms vary and lead to delay of diagnosis.	 Symptom complexity leads to frequent misdiagnosis.	 HD needs a multidisciplinary and holistic approach. A long-term perspective is essential.	 Disease progression leads to struggle to adjust.
Goals	 Educate clinicians about premanifest HD.	 Educate families on how to best cope and seek support.	 Accurate & quick diagnosis. Good follow-up process. Support network for patients.	 Establish multidisciplinary teams.	 Build trusting relationships between patients, families and clinicians.

HD Huntington's Disease
QoL Quality of Life

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.

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