Vegferðir sjúklinga eru myndrænar yfirlitsmyndir sem sýnir þarfir sjúklinga í tengslum við umönnun og meðferð sjaldgæfra sjúkdóma.

Þar sem þær eru mótaðar út frá sjónarhorni sjúklinga gera þær heilbrigðisstarfsfólki kleift að mæta með markvissum hætti þeim þörfum sem sjúklingar með sjaldgæfa sjúkdóma hafa.

Hægt er að finna ítarlegri útgáfu af þessari vegferð á ensku hér:

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 health care professionals that work in clinical units and nursing homes about the specific needs of patients with late-stage HD.

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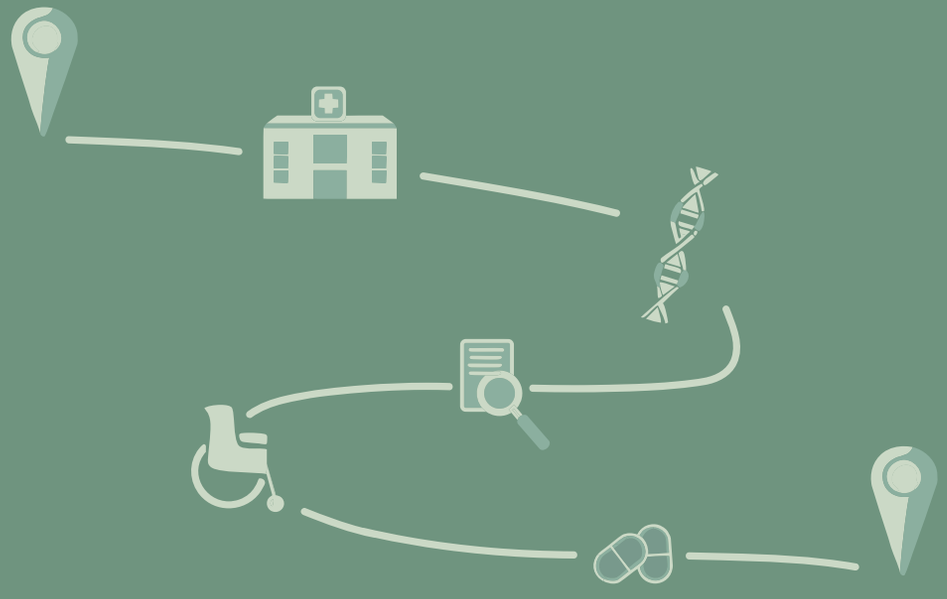
European Reference Network
for rare or low prevalence complex diseases

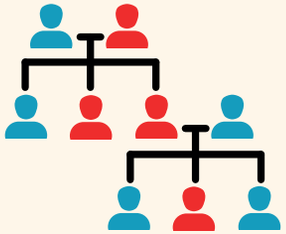
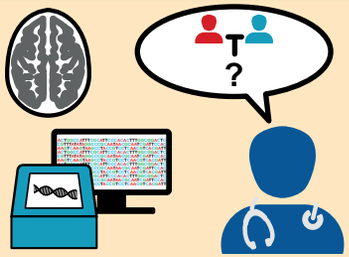
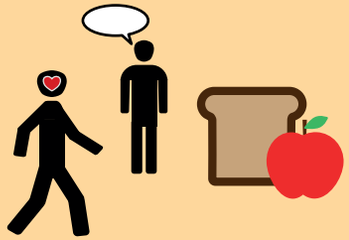
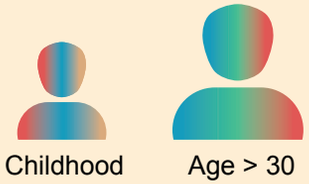
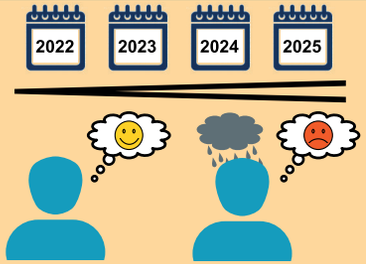
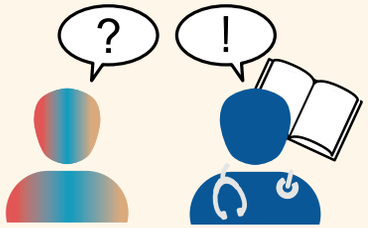
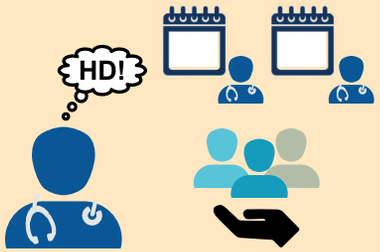
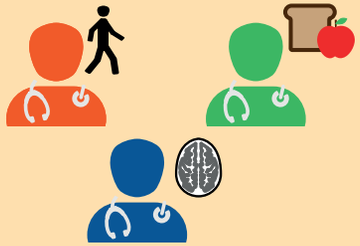
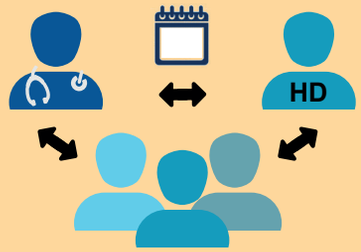
Network Neurological Diseases (ERN-NMD)

huntington.is
ern-rnd.eu

Vegferð sjúklings með Huntington-sjúkdóm



HD SAMTÖKIN
Á ÍSLANDI

	Forstig HD	Fyrstu einkenni	Greining	Meðferð	Eftirlit
Klínískt	 Flestir sem greinast með HD hafa þegar séð nána ættingja þróa með sér sjúkdóminn.	 Væg fyrstu einkenni falla í 3 flokka: hreyfi-, vitræn-, og hegðunareinkenni.	 Staðfesta klíníska greiningu með erfðagreiningu. Erfðaráðgjöf nauðsynleg.	 Engin sjúkdómshamlandi meðferð þekkt. Meðferð við einkennum til að viðhalda lífsgæðum og virkni.	 Hreyfing, andleg vellíðan og rétt næring viðheldur virkni og sjálfstæði.
Áskoranir	 Ótti við birtingu sjúkdómsins leiðir til hunsunar á einkennum.	 Mismunandi birtingarmyndir fyrstu einkenna sjúkdómsins leiðir til seinkunar á greiningu.	 Margbreytileiki einkenna leiðir oft til rangrar greiningar.	 Margbreytileiki einkenna leiðir oft til rangrar greiningar.	 Framvinda sjúkdómsins leiðir til aðlögunarvandamála.
Markmið	 Fræða lækna um forstig sjúkdómsins.	 Fræða fjölskyldur um hvernig best er að takast á við sjúkdóminn og leita stuðnings.	 Nákvæm og fljót greining. Góð eftirfylgni. Stuðningsnet fyrir sjúklinga.	 Stofna þverfagleg teymi.	 Byggja traust sambönd á milli sjúklinga, fjölskyldna og lækna.

HD = Huntington's Disease

Vinsamlegast athugið heiti þjónustu (t.d., heimaþjónusta, umönnun, heimilislæknir, sjúkraþjálfun) fela ekki í sér sömu þjónustu í öllum löndum Evrópu og gæti verið mismunandi eftir löndum. Hagsmunasamtök sjúklinga geta oft veitt sjúklingum og fjölskyldum stuðning og úrræði.

Fyrirvari

ERN-RND afsalar sér allri ábyrgð á sölu eða hæfi fyrir tiltekna notkun eða tilgang. ERN-RND tekur enga ábyrgð á meiðslum eða skemmdum á mönnum eða eignum sem stafa af eða tengjast notkun þessara upplýsinga eða fyrir hvers kyns villur eða vanrækslu.

HD SAMTÖKIN