PATIENT JOURNEY FRIEDREICH'S ATAXIA (FA)

PHASES	1 - First symptoms	2 - Diagnosis	3 - Treatment	4 - Monitoring
Disease	Difficulty walking in the dark, unsteadiness in standing or walking, followed by progressive gait and limb clumsiness. 91% of people present with poor balance or falls. 9% present with non-neurological symptoms i.e. scoliosis or heart trouble which may precede neurological symptoms.	Genetic testing for FA can be done since 1996 but is absent from standard next generation sequencing (NGS) and requires specific tools.	• One disease-specific therapy approved in Europe and USA - omaveloxolone • Several clinical trials ongoing using different disease modifying treatment • Scoliosis surgery may be indicated when the Cobb angle is >30 degrees • Cardiomyopathy can be treated with medication	Referral to expert centre with involvement of multi-disciplinary teams for monitoring scoliosis during growth and life-long monitoring of the heart and risk of diabetes mellitus.
Clinic	Assessment of symptoms and referral to relevant specialists. Multidisciplinary teams that include neurologist, orthopaedic, cardiology, psychology and other healthcare professionals depending on need.	Counselling: • parents regarding future pregnancies • Siblings unless symptomatic are usually not tested before the age of 18 years. Some centres offer the genetic test if delay is causing anxiety • Partners of those with FA • FA carriers • Relatives who are of reproductive age	1) Psychological and mental health support for the individual and all family members for this life-altering condition. Adolescents are at increased risk of suicidal ideation. 2) Annual neurological assessment **3)** Scoliosis and foot deformity assessment to see if physiotherapy, splints or surgery is indicated.	Annual visit to assess: 1) Mobility 2) Activities of daily living 3) Heart problems 4) Diabetes mellitus status 5) Development of any other symptoms or signs, assess if they are due to FA
Challenges	1) Easy to confuse the clumsiness of FA with a growth spurt, 2) Changes are insidious in FA and may not be apparent to the individual or their family at an early stage 3) FA can have atypical presentations The above factors lead to misdiagnosis or a delay in the diagnosis of FA.	As FA is not easily identified on the new gene sequencing (NGS) technology, it may be missed unless a neurologist specifically requests the test.	Access to omaveloxolone due to its high cost Encourage: • use of posterior walker to try and prolong their ability to walk • participation in social activities with peers • parents to avail of outside help if available, which gives them a short break and enlarges the social circle of the person with FA • adolescents to maintain autonomy	Parents should be supported and advised about how to communicate the diagnosis to their affected child. The child/adolescent may not be able to compete physically with their peers and may retreat into themselves. As a result, the parents are often traumatised and unsure how to treat/advise the person with FA.
Goals	Multi-system complaints (especially in children & adolescents) should be taken seriously, i.e. poor balance, fatigue, heart problems, back pain (scoliosis) & anxiety. Get a 2nd opinion in those with the above multi-system vague complaints, especially if the parents are very worried.	Asymptomatic siblings, aunts/uncles and grand-parents of the person with ataxia should be offered genetic counselling and testing to avoid FA presenting in cousins and future generations.	• Worldwide free access to omaveloxolone for all who have FA • Neurologists share the updated (2022) FA care guidelines with those who have FA which would enable the individual to bring them to other health care appointments and Accident & Emergency should they need arise.	Maximise the person's potential to live as normal a life as possible. In this respect, learning to drive and part-time work is very important.

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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