	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.	There are no effective neurological disease-modifying therapies available yet. 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 of 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. Counselling for young adult people of reproductive age. Genetic testing for partners of those with FA or FA carriers.	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) 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) Developments of any other symptoms or signs, assess if they are due to FA.
Challenges	1) Easy to confuse the clumsiness associated with a growth spurt to that due to FA 2) Changes are insidious in FA and may not be apparent to the individual. The above factors lead to frequent delayed or misdiagnosis.	As FA is not easily identified on the new NGS technology, it may be missed unless a neurologist specifically requests the test.	There are several clinical trials internationally but no effective treatment so far. 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 possibilities for the individual with FA - adolescents to maintain autonomy	The child/adolescent may not be able to compete with their peers and may retreat into themselves. The parents are often traumatised and unsure how to treat the individual with FA. Parents should be supported and advised about how to communicate the diagnosis to their affected child.
Goals	Multi-system complaints (especially in children and adolescents) should be taken seriously, i.e. poor balance, fatigue (heart problems), back pain (scoliosis) irritability and anxiety. Get a 2nd opinion in those with the above multi-system and 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.	Care guidelines are available and shared with the person so that they can bring them to medical appointments and have if a medical emergency arises and they have to go to Accident & Emergency.	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