

## First Symptoms

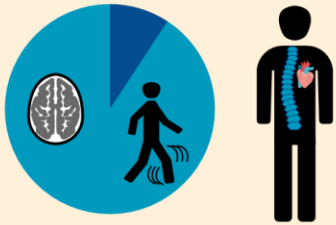
## Diagnosis

## Treatment

## Monitoring

Disease

91% neurological symptoms.  
9% non-neurological symptoms:  
scoliosis and heart trouble



Genetic testing is available, but  
gene changes in FA are not  
recognized using standard NGS



One disease-specific therapy in  
Europe and USA approved,  
several clinical trials ongoing



Referral to expert center.  
Multidisciplinary team for heart  
condition and diabetes



Clinic

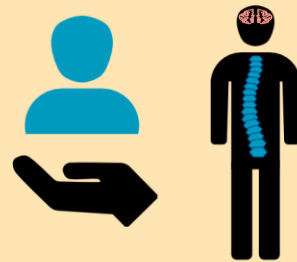
Assessment of symptoms and  
referral to relevant specialists



Genetic counselling for parents  
regarding future pregnancies  
and siblings above 18.



Mental health support,  
neurological and scoliosis  
assessment



Annual review of mobility,  
ability to do daily activities,  
heart, diabetic risk etc.



Challenges

Confusion and complexity of  
symptoms leads to frequent  
misdiagnosis



Consider diagnosis in all age  
groups, as 1% of those with FA  
are over 60 years old.



Maintaining personal autonomy  
and ability to walk, access to  
currently available treatments



Children may isolate themselves.  
Parents are often unsure how to  
treat their child with FA

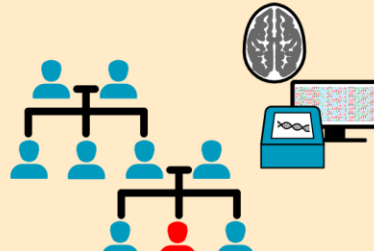


Goals

Take patients with multi-system  
complaints seriously, i.e.  
clumsiness, fatigue, back pain



Genetic counselling and testing  
for extended family to avoid FA  
presenting in cousins



Care guidelines should be  
shared with person with FA.  
Worldwide access to therapy



Maximize the potential to live  
as normal a life as possible, e.g.  
driving and part-time work.



FA  
NGS

Friedreich's Ataxia  
Next Generation Sequencing  
(mapping of entire genome)

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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