

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



Several clinical trials ongoing,
but no effective disease-
modifying therapies yet



Referral to expert centre. Multi-
disciplinary team for heart
condition and diabetes



Clinic

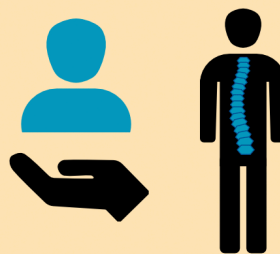
Assessment of symptoms and
referral to relevant specialists



Genetic counselling for parents
regarding future pregnancies
and young adults



Mental health support and
scoliosis assessment



Annual neurological and cardiac
assessment, diabetes check-up,
review mobility and daily living



Challenges

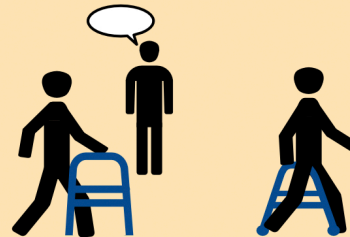
Confusion and complexity of
symptoms leads to frequent
misdiagnosis



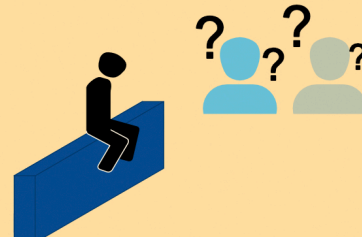
Consider diagnosis in all age
groups, as 1 % of those with FA
are over 60 years old



Enlarge social circle, maintain
personal autonomy, prolong
walking ability with use of aids



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

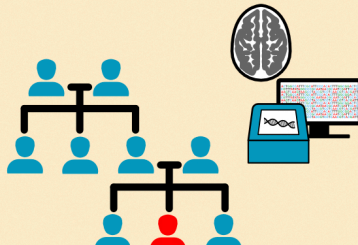


Goals

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 are available
and should be shared with
person with FA



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

