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# FRIEDREICH'S ATAXIA: PRESENTATION OF A CASE

KEYWORDS: Autism, Psychological Well Being,

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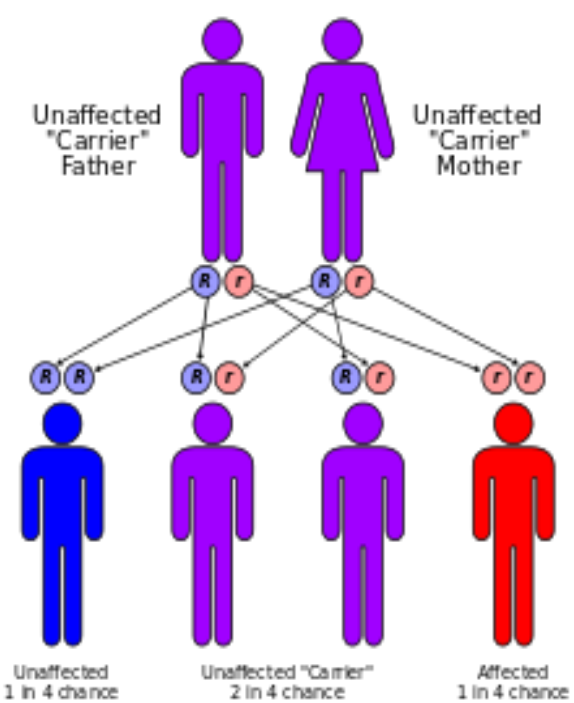
**ABSTRACT** *Friedreich's ataxia is rare genetic disorder with autosomal recessive inheritance. It is suspected with its clinical presentation and trinucleotide repeats. We present A 24 years old male patient who was admitted in this hospital for complaints of ataxia, dysarthria and dysphagia since 5 years. Patient also had chest pain, palpitations, breathlessness, titubation, pes equino varus, bilateral lower limb muscle atrophy, cardiomyopathy, staggering gait. After investigating and thorough examination a diagnosis of Friedreich's ataxia was made.*

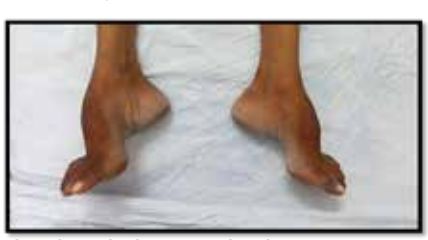
**INTRODUCTION:** Friedreich's ataxia also known as spinocerebellar ataxia is autosomal recessive disease and is most common form of inherited ataxias characterised by progressive staggering gait, titubation, dysarthria, foot deformity, nystagmus, cardiac involvement in 90% cases, more severe involvement of lower extremity. The diagnosis can be confirmed by gene mapping studies for expanded GAA trinucleotides repeats in mutant *frataxin* gene. Frataxin is mitochondrial protein involved in iron homeostasis. Normal persons have 7-22 GAA repeats ,and the patients have 200-900 GAA repeats. We diagnosed FA in 24 years old male on the basis of clinical findings.




**CASE DETAIL:**

**The clinical photograph showing wasting of both lower limbs.**

A 24 years male patient was admitted to the ward for complaints of breathlessness, palpitation, dysarthria, dysphagia, dizziness more since 15 days. Patient has inability to walk since childhood due to progressive ataxia and gradual worsening weakness of both lower limbs. The patient left school because of progressive ataxic paraparesis. There was no history of birth injury. The family history was also not significant.



**The clinical photograph showing pes cavus deformity.**

Examination revealed fully conscious patient with PR-72/

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min, RR-16/min, BP-110/70mm Hg , No signs of respiratory distress, No signs of CHF, Patient had bilateral Equinovarus deformity. His Neurological examination revealed evidence of mild mental retardation, normal cranial nerves. Both pupils were normal size but had bilateral horizontal nystagmus. The motor examination revealed gross wasting of both lower limbs as shown in fig **a**. The tone was normal in upper limbs but reduced in both lower limbs. The power was 5/5 in both upper limbs but 4/5 in both lower limbs. Patient had incoordination in upper and lower limbs. Titubation was present. The deep tendon reflexes were absent with bilateral Positive Babinskis response. The sensations were intact. Patient had Ataxic gait. The cvs examination: normal heart sounds without murmur. Other systems were normal.

- His Investigations revealed Hb-13gm%, TLC- 8700/ mm3, plt-2.4 lacs/mm3; rest blood indices were normal, liver function tests and kidney function tests were normal.
- ECG revealed sinus tachycardia.
- 2D ECHO showed: Dilated RA RV LA LV s/o DCM, No RWMA, No VEGETATIONS , CLOTS.
- MRI BRAIN revealed: Mild cerebral cortical atrophy. Few foci of T2W & FLAIR hyperintensities in bilateral corona radiata, bilateral centrum semiovale and in subcortical white matter of bilateral frontoparieatal lobes, s/o ?ischemic changes/ demyelinating changes.
- MRI of whole spine was normal

With above clinical findings and investigations diagnosis of Friedrich's Ataxia was made.

**DISCUSSION:**
Friedreich's Ataxia is an autosomal recessive disorder. The incidence being 1 in 50,000 with an estimated carrier prevalence of about 1 in 110.

Important clinical features: gait ataxia, titubation, extensor plantar response, absent deep tendon reflexes, normal tone in trunk and extremities, weakness, dysarthria, dysmetria, equinovarus deformity and cardiomyopathy.

Diagnosis is based on clinical findings and investigations and confirmed on genetic findings of GAA trinucleotide repeat sequence analysis.

Our patient also had similar clinical presentation hence clinical diagnosis of Friedricks Ataxia was suspected.

Differential diagnosis of the condition being other hereditary ataxias having same cerebellar symptoms . But these were excluded as the patient had no Telangiectasias. No Retinitis pigmentosa. No Pyramidal tract involvement. No peripheral neuropathy.

**GENES INVOLVED**
The classic form of FA has been mapped to 9q13-q21.1 and the mutant gene, *frataxin,* contains expanded GAA repeats in >95% of the patient. Normal person have 7-22 GAA repeats, and patients have 200-900 GAA repeats. Patients with FA have undetectable or extremely low levels of *frataxin* m RNA. It has autosomal recessive inheritance caused by an unstable mutation on chromosome 13. The primary sites of pathology are the spinal cord, dorsal root ganglia cells and peripheral nerves. Sclerosis and degeneration occur predominantly in the spinocerebellar tracts, lateral corticospinal tracts and posterior columns. Cardiac pathology consist of myocytic hypertrophy and fibrosis, focal vascular fibromuscular dysplasia with subintimal or medial deposition of PAS + material ,myocytopathy.

**SUMMARY:**
The reported case has majority features suggestive of Freidreich's Ataxia. It is autosomal recessive ataxia.

Important clinical features: gait ataxia, titubation, extensor plantar response, absent deep tendon reflexes, normal tone in trunk and extremities, weakness ,dysarthria, dysmetria, equinovarus, cardiomyopathy.

Diagnosis is based on clinical findings and investigations and confirmed on genetic findings of GAA trinucleotide repeat sequence analysis.

Our patient also had similar clinical presentation and hence diagnosis of Friedreich's Ataxia was made.

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