

## CASE REPORT

**Friedreich's Ataxia – A Clinical Diagnosis***Md. Fekarul Islam<sup>1</sup>, Devdeep Mukherjee<sup>1\*</sup>, Ritabrata Kundu<sup>1</sup>, Joydeep Das<sup>1</sup>*<sup>1</sup>*Department of Pediatric Medicine, Institute of Child Health, Kolkata-700017, (West Bengal) India***Abstract:**

Friedreich's ataxia (FA) is an autosomal recessive spinocerebellar degenerative disease characterized by hyperexpansion of GAA triplets in Frataxin gene. The hallmark of this disorder is ataxic gait, areflexia, Babinski's sign and positive Romberg test. We report a 9 year old child who presented with all these features and was diagnosed with FA on the basis of these clinical features. There are few case reports of FA where the diagnosis was made so early.

**Keywords:** Friedreich's ataxia (FA), Frataxin, trinucleotide repeat

**Introduction:**

Friedreich's ataxia (FA) is an autosomal recessive neurodegenerative disease, usually manifested before adolescence involving both the central and the peripheral nervous system. It is the commonest of the inherited ataxias, affecting approximately 1–2 per 100,000 of the population and is caused by mutations in frataxin gene (mapped on chromosome 9q13) [1].

**Case Report:**

9 year female child born out of non consanguineous marriage was admitted with a history of progressive difficulty in walking since the last 2 years. She also complained of difficulty in climbing down the stairs and squatting. However she did not complain of muscle wasting or paraesthesia of lower limbs. Her birth history was insignificant. She also did not have any family history of similar illness.

On examination, she had normal sensorium. Her vitals were stable. There was neither any intellectual impairment nor any loss of vision or hearing. She was having dysarthria. She also had

diminished power and tone in both her upper and lower limbs. Her upper limb reflexes were diminished. She also had absence of knee jerk and ankle jerk. There was an extensor planter response. She had a wide based gait with ataxia and Romberg's test was positive. However she could walk around with support. Our patient also had intention tremor, past pointing and horizontal nystagmus in extremes of lateral gaze. Although pain and touch sensation was present, joint position and vibration sense was absent. She was also having kyphoscoliosis, pes cavus (Fig. 1) and hammer toes.



**Fig. 1: Pes Cavus with Hammer Toes**

There were no involuntary movements, calf muscle hypertrophy or fasciculations. Her sensory system and cranial nerves examination was normal. Fundus examination did not reveal any evidence of optic atrophy. She also did not have any difficulty in feeding.

Nerve conduction velocity was normal. Magnetic resonance imaging (M.R.I) brain revealed thickened cerebellar folia (Fig. 2). Her fasting and post prandial blood sugar were 89 mg/dl and 115 mg/dl respectively. Her electrocardiogram and echocardiography were both normal. There was no evidence of cardiomyopathy.



**Fig. 2: MRI of the Brain Revealed Thickened Cerebellar Folia**

In view of the clinical findings, a diagnosis of FA was made. Physiotherapy was advised to her. She is presently in regular follow up. 6 monthly ECG and ECHO are being done along with blood sugar monitoring.

#### **Discussion:**

Friedreich's ataxia is a spinocerebellar degenerative disease genetically characterized by hyperexpansion of GAA triplets in the first intron of the frataxin gene on chromosome 9 [2]. FA affects central and peripheral nervous systems, skeleton, heart and endocrine pancreas. The estimated carrier frequency ranges from 1:50 to 1:100 in people of European, North African, Middle Eastern and Indian origin.

FA is primarily a clinical diagnosis. It is typically a disease of young adults (around puberty) with no gender predilection. A family history of Friedreich's ataxia may lead to early diagnosis [3,4]. Our patient started developing the symptoms from 7 years of age. She however did not have a family history of FA.

The most common clinical manifestations are limb and postural ataxia, muscle weakness,

dysarthria, dysmetria, volitional tremor, loss of vibration and position sense, extensor planter response, lower limb areflexia and distal amyotrophy, often associated with osteomuscular deformities such as scoliosis, pes cavus and hammer toes [2, 5]. Gait ataxia is the earliest symptom in the vast majority. Our patient, even though she presented at a very early age, had all the above mentioned features.

According to Harding's criteria an age of onset before 25 years, progressive ataxia, absent deep tendon reflexes and dysarthria are considered to be essential criteria. Scoliosis, pyramidal weakness in lower limbs, absent tendon reflexes in arms, impairment of vibration and joint position sense and cardiomyopathy are additional criteria. Nystagmus, optic atrophy, fixation instability, deafness, distal amyotrophy, pes cavus and diabetes mellitus are other criteria found in less than 50% cases [4, 6].

Cardiomyopathy is the most important non neurological feature of FA. The exact proportion of patients with cardiomyopathy is still not known. However, in a study where hearts were examined in detail, over 90% were found to have abnormalities, though the clinical significance of some of the lesser changes is unclear [7]. About 65% of patients have an abnormal electrocardiogram with widespread T wave inversion in the inferolateral chest leads. The most frequent echocardiographic abnormality is concentric ventricular hypertrophy [8]. Cardiac involvement however appears a bit late. Cardiac conduction abnormalities are also common. Our index case had not yet developed cardiomyopathy. The incidence and progression of cardiomyopathy is directly proportional to the length GAA expansion.

Abnormalities in nerve conduction studies are severely slowed or absent sensory conduction

with normal motor conduction velocity. MRI may also be abnormal. The final confirmation is by molecular analysis. However, due to financial constraints, this was not possible in our patient.

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