A case of Friedreich ataxia

G N Lucas[1], T G Y R Gunapala[2]

Sri Lanka Journal of Child Health, 2001; **30**: 17-18

(Key words: Friedreich ataxia)

Introduction

Friedreich ataxia is the commonest of the inherited ataxias, accounting for at least 50% in most large series and affecting approximately one in 50,000 individuals[1]. The gene for Friedreich ataxia was mapped to chromosome 9ql3 in 1988 by Chamberlain and colleagues[2]. Harding's criteria are widely adopted in the diagnosis of Friedreich ataxia[3]. According to this, an age of onset before 25 years, progressive ataxia of gait and limbs, absent knee and ankle jerks and dysarthria are considered to be essential criteria. Scoliosis, pyramidal weakness in lower limbs, absent reflexes in arms, impairment of vibration and joint position sense and cardiomyopathy are additional criteria present in over 66% cases. Nystagmus, optic atrophy, deafness, distal amyotrophy, pes cavus and diabetes mellitus are other criteria found in less than 50% of cases. The natural history of this disease is one of relentless progression. The patient usually becomes wheelchair bound within 10-15 years of onset[3]. This is the first reported case of Friedreich ataxia in Sri Lanka.

Case report

A nine year old boy from Udugampola was admitted to the Lady Ridgeway Hospital in November 1998 with a history of polyuria, polydipsia and weight loss of 1 month duration, shortness of breath while walking of 1 year duration and unsteady gait of 4 year duration. He was the eldest child of non consanguineous parents. The other 2 children were normal. He was well up to the age of 5 years when he was noticed to have an unsteady wide based gait which progressively worsened over the past 4 years. There was no loss of hearing or vision and no intellectual impairment. All immunisations, including the measles vaccine had been given. There was no family history of similar problems.

On examination, the child was drowsy and moderately dehydrated with acidotic breathing. Scoliosis was present. There was no pes cavus but pes planus was present. There was cardiomegaly but no significant murmurs. There was dysarthria but no nystagmus and no optic atrophy. There were intention tremors and past pointing and the Romberg test was positive. The gait was ataxic. The power and tone were reduced in upper and lower limbs. The upper limb reflexes were diminished. The ankle and knee jerks were absent. An extensor plantar response was present. Joint position sense and vibration sense were absent. However, pain and touch sensations were intact.

The random blood sugar was 32.5 mmol/I (586 mg/dl) and ketone bodies were present in the urine. The ECG was normal. A 2-D echocardiogram showed hypertrophic cardiomyopathy.

The child was discharged home on lente insulin. In April, 2001 he was readmitted to the ward with a second episode of diabetic ketoacidosis. A repeat echocardiogram showed hypertrophic cardiomyopathy. There was no optic atrophy. Now the child is only able to walk with support.

In our patient ataxia, positive Romberg sign, intention tremor, past pointing, dysarthria, absent knee and ankle jerks, extensor plantar response, impairment of vibration sense and joint position sense together with scoliosis, diabetes mellitus and cardiomyopathy suggested a diagnosis of Friedreich ataxia.

Although pes cavus is the best known foot deformity found in Friedreich ataxia, pes planus and equinovarus are also often found[4]. The most important non-neurological feature of Friedreich ataxia is cardiomyopathy. The exact proportion of patients with cardiomyopathy is still debated. 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[5]. 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[6]

[1]*Consultant Paediatrician,* [2]*Paediatric Registrar, Lady Ridgeway Hospital, Colombo*

Recently, a 4 year old boy with Friedreich ataxia underwent cardiac transplantation because of cardiomyopathy with ischaemia[7]. Diabetes mellitus may occur with an incidence varying from 8-23% in different reports[8,9].

References

1. Geoffrey G, Barbeau A, Breton G. Clinical description and roentgenographic evaluation of patients with Friedreich's ataxia. *Can J Neurol Sci* 1976; **3**: 279-86.

2. Chamberlain S, Shaw J, Rowland A. Mapping of mutation causing Friedreich's ataxia to human chromosome 9. *Nature* 1988; **334**:248-50.

3. Harding A E. Friedreich's ataxia: a clinical and genetic study of 90 families with an analysis of early diagnostic criteria and intrafamilial clustering of clinical features. *Brain* 1981; **104**:589-620.

4. Wood NW. Diagnosing Friedreich's ataxia. *Arch Dis Child* 1998; **78 (3)**: 204-7.

5. Harding A E. The inherited ataxias. *Adv Neurol* 1988; **4** :37-46.

6. Pentland B, Fox KA. The heart in Friedreich's ataxia *Neurol Neurosurg Psychiatry* 1983; **46**:1138-42.

7. Leonard H, Forsyth R. Friedreich's ataxia presenting after cardiac transplantation. *Arch Dis Child* 2001; **84**:167-8.

8. Hewer R L. Study of fatal cases of Friedreich's ataxia *Br Med J* 1968; **3**: 649.

9. Hewer RL, Robinson N. Diabetes mellitus in Friedreich ataxia. *J Neurol Neurosurg Psychiatry* 1968; **31**: 226.