CASE REPORT

Friedreich's Ataxia

by

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Abstract

A girl of the Batak Mandailing sect suffering from Friedreich's Ataxia who was admitted to the Department of Child Health, Medical School, University of North Sumatera/RSUPP, Medan, is reported. Her four sisters also had the same disease, where three of them died. The most important symptoms were ataxia, kyphoscoliosis, positive Babinsky's sign, dysmetria, adiadochokinesis, positive Bomberg's sign, and absence of knee and ankle jerks. In this case and her four sisters the symptoms started at about the age of 10 years. To our knowledge this is the first report on Friedreich's Ataxia occurring in an Indonesian family from North Sumatera.

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Introduction

Friedreich's Ataxia is a term applied to a rather heterogenous group of disorders which manifests commonly in the late childhood or adolescence as progressive cerebellar and spinal cord dysfunction. In most families this so called Friedreich's Ataxia is transmitted on autosomal recessive basis, with spinocerebellar dysfunction as the chief signs. The group of predominantly spinal form is dominated by Friedreich's Ataxia, characterized by progressive degeneration of the dorsal column and dorsal roots especially in the lumbosacral region.

The clinical history is progressive gait disturbances, followed by incoordination of the upper limbs, initially associated with skeletal deformities, pes cavus (highly arched foot), hammer toes, and scoliosis. Clinical signs of the cerebellar disorders include gait ataxis, dysarthria, dysmetria intention tremor, and less commonly, nystagmus. Friedreich's Ataxia has evidence of corticospinal tract dysfunction which leads to a positive Babinsky's sign, and of peripheral neuropathia such as loss of tendon reflexes leading to distal weakness and muscles atrophy.

The combination of ataxia, Babinsky's sign, and absence of knee and ankle jerk are pathognomonic for the disease. The sensory loss occurs especially in the fect with positional and vibratory senses most severely affected. The child's balance is unsteady and Romberg's sign is positive, and sometimes cardiac failure is also present. The criteria are based upon :

1. The onset in the first decade
2. A positive family history
3. The degenerative signs of posterior column of the spinal cord and the cerebellar apparatus
4. Skeletal deformities with kyphoscoliosis, pes cavus, and hammer toes.

Case report

R.H., 14 years of age, was admitted to the Department of Child Health, Medical School, University of North Sumatera/RSUPP, Medan, on 4th April 1977 with the complaints of muscles pain and weakness and unstableness of the lower limbs. The weakness and unstableness developed gradually starting about 3 years ago. She fell quite often, and had difficulties in walking. These symptoms progressed eventually and became worse during the last year. No abnormalities of birth history and nutrition could be seen. The growth and mental development seemed normal up to the age of 10 years, at which time the weakness and unstableness began.

Family history shows that she is the youngest of 9 siblings. Numbers 2, 4, and 8 are all healthy, but number 3 died during the perinatal period. Her other sisters, number 1 and 6, and her brother, number 5 died apparently suffering from a disease with the

same symptoms as this patient. Sibs numbers 1, 5, and 6 suffered from a disease with the same symptoms as hers. Their onset were respectively at the ages of 18, 20, and 24 years. Her brother number 7, 20 years old and still alive, is said to have the same symptoms as this patient. He never had any convulsion.

Physical examination on admission revealed a girl of 14 years old with a body weight of 32 kg., height 135 cm., body temperature 37°C, heart rate 100/minute, RR 28/minute. The patient looked shy and weak, otherwise she was alert and quite active. No abnormalities of the head, neck, thorax, abdomen, and genital were noted; columna vertebrae: kyphoscoliosis.

Neurological examination shows that she is active and alert, no symptoms of increased intracranial pressure, and no abnormalities of cranial nerves. Motoric movement revealed a good muscle strength of the upper and lower limbs. Sensibilities showed a slight hypaesthesia of the skin from the umbilicus downwards to both legs. Vibratory senses was negative on both arms and legs. Proprioceptive disturbances were positive. Reflexes of biceps and triceps were normal, but of ankle and knees were negative. Babinsky's sign was positive; coordination — swinging to the left and right: Romberg's sign (+); post-pointing phenomenon (dysmetria) positive: cerebellar sign — ataxia (+), dysmetria (+); extrapyramidal sign — coordination bad, tremor (—), intention tremor (—).

Summary of physical examination :

1. Onset at 10 years of age (first decade)
2. Strong family history
3. Proprioception and gait disturbances, ataxia (+), dysmetria, Romberg's sign (+)
4. Knee tendon reflexes (—), ankle tendon reflexes (—), Babinsky's sign (+)
5. Kyphoscoliosis
6. Absence of muscle weakness and atrophy
7. Disturbance of skin and deep sense

Laboratory findings :

Urine : no abnormality,

Feces : ascaris (+),

Blood : Hb 10 gm.%, leucocytes 6400/mm^3, AT (—),

CSF : clear, Nonne (—), Pandy (—), cells 11/3,

Biopsy of mm. gastrocnemius (right and left) on 25th April 1977 : no abnormalities,

Radiological examination : no abnormalities of the thorax, skull, and articulatio coxae, kyphoscoliosis of the spine.

Other examination :

Blood CPK : 53 U/ml.

Blood cholesterol : 200 mg.%

SGOT : 7 mU/ml.

SGPT : 8 mU/ml.

ECG : normal with right axis deviation.

Ophthalmologic examination : no abnormalities.

D.D. : 1. Levy - Roussy syndrome
2 Bassen - Kornzweig syndrome
3. Hypertrophic interstitial neuritis
4. Peroneal muscular atrophy
5. Juvenile tabes (Tabes dorsalis)

1. Levy - Roussy syndrome :

This disorder may be a variant of Friedreich's ataxia with muscular wasting. Symptoms develop early in childhood and include impairment of equilibrium in walking and standing; knee and ankle jerks are lost. Atrophy of the muscle of the lower extremities is seen, and slowly progressing.

2. Bassen - Kornzweig syndrome :

This is a recessive inherited disease with malabsorption of fat and a beta-lipoproteinemia which is associated with progressive cerebellar ataxia and pigmentary degeneration of retinae. Clinical pattern resembles that of Friedreich's ataxia. Laboratory findings show decreased of serum cholesterol and blood carotene. The red blood cells have multiple spiny projection (acanthocytosis). The therapy at present is limited to supplementary administration of the fat soluble vitamins, including vitamins A, D, and E.

3. Hypertrophic interstitial neuritis :

It is a heredo-familial chronically progressing polyneuritis with both motor and sensory symptoms. The onset is in childhood and the disease progresses slowly. It usually begins in the lower extremities with progressive weakness and difficulty in walking. Distal atrophy of the extremities and loss of tendon reflexes, gait disturbance and ataxia caused by loss of position sense are present. Pes cavus is positive. Peripheral nerves become palpable (enlarged): CSF — protein level is increased.

4. Peroneal muscular atrophy (Charcot -- Marie - Tooth disease) :

Motor neuropathy which affects the nerves of the legs. Inheritance is usually on dominant basis, and is a slow progressive disease. Onset occurs in late childhood, and characterized by clubbing of the feet, muscular wasting which begins in the legs. Peroneal myotrophy and foot drop give the appearance of "stork legs". Mild distal sensory impairment may be present: CSF is normal.

5. Juvenile tabes (locomotor ataxia):

It is characterized by marked ataxia due to a loss of proprioceptive sense. Tabetic crises and "charcot" joints are often seen. Perforating ulcer of the ball of the foot occurs. Muscular hypotonia, atrophy, and hypaesthesia are present. Argyll - Robertson pupils and optic atrophy are positive. Deep tendon reflexes are either decreased or negative. Romberg's sign, Abadie's sign, and Biernac-

ky's sign are positive. The syndrome is the result of syphylitic infection of the posterior roots and the roots entrance zone of the spinal cord. The spinal cord is smaller and atrophied. Laboratory findings of CSF revealed clear, normal or slightly increased cells. Protein is slightly increased, Wasserman reaction, VDRL, and Gold curve are all positive; microscopically: decrease of number of normal nerve fibers of the lumbo-sacral roots, cellular infiltration of the dorsal roots ganglia and degeneration of posterior column.

Discussion

The presence of Friedreich's ataxia in Medan proved that the disease is not impossibly found in Indonesia. In the literature this disease is rarely found and widespread all over the world. There are no climatic nor geographical influence in the distribution of the disease. Friedreich's ataxia is however a fairly well-defined syndrome where the spino-cerebellar ataxia forms the main sign, but the spinal form is more dominating characterized by progressive degeneration of the dorsal column and dorsal roots especially in the lumbo-sacral region. As a rule the cerebellum is unaffected, but involvement in the form of loss of Purkinje cells has been reported. The symptoms found in those 2 cases are listed as follows.

Symptoms	Case 1 : female 14 years of age	Case 2 : male 20 years of age
1. Onset in the first decade	10 years of age	12 years of age
2. Positive family history	+	+
3. Indicative signs of degeneration of posterior column of the spinal cord :	+	+
a) ataxia	—	—
b) knee tendon reflexes	—	—
c) Achilles tendon reflexes	+	+
e) loss of vibratory sense	+	+
4. Disturbances of cerebellar apparatus		
a) Romberg's sign	+	+
b) dysdiadochokinesis	+	+
c) dysmetria	+	+
d) scanning speech	+	+
5. Disturbances of cortico-spinal tract : Babinsky's sign	+	+
6. Skeletal deformities with :		
a) kyphoscoliosis	+	+
b) pes cavus	—	+
7. Electrocardiogram examination	R.A.D.	R.A.D. and sinus tachycardia

The diagnosis depend almost totally on the clinical findings. Laboratory examinations give no values except for electrocardiographic changes suggestive of myocarditis; however, in advanced cases there are abnormalities in the peripheral nerve conducting velocity owing to peripheral neuropathy. R.A.D. and sinus tachycardia are common in Friedreich's ataxia, which seem to increase with the degree of severity of the disease (Thoren, 1964).

According to Thoren's study (1964), in both clinical and physiological events it is convenient to group them according to the degree of movement impairment which is limited chiefly by the degree of ataxia. The patients are then classified in the following groups :

Group I : slight physical handicap, i.e. slightly unsteady gait with ability to run.

Group II : moderate, i.e. ability to walk without support, but inability to run.

Group III : severe, i.e. inability to walk without support, wheelchair mostly required.

Group IV : very severe,i.e. wheel chair definitely required owing to more or less totally paretic legs.

According to these classifications, these 2 patients are classified as Group II for case 1, and Group III for case 2. The degree of physical handicap increased as fast as the progression of the disease. Death is almost secondary to myocardiac failure and infection.

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