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Case Report

Friedreich's Ataxia-A Case Report

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Friedrich's ataxia (FA) is an autosomal recessive disorder. It is the most common cause of inherited ataxia. It affects approximately 1-2 persons per 100,000 population.[1] It occurs due to a mutation that results in the homozygous expansion of Guanosine Adenine Adenine trinucleotide (GAAT) repeat units in intron 1 of FRATAXIN gene in chromosome 9.[2,3] Its pathophysiology can be explained by "Dying back phenomenon"; progressive damage to axons with ultimate neuronal death, mostly in spinal cord but can also involve cranial nerves VII, X and XII.[4] Apart from neurological manifestations cardiomyopathy[5,6] and endocrine pathologies also occur which contribute to a worsened prognosis of the disease i.e. within 15-20 years of onset, patient becomes wheel chair bound and has a shortened life expectancy. Cardiac pathologies are the most common cause of death in Friedreich's ataxia.[2]

Case report

11 years old boy, born of consanguineous marriage, presented with complaints of difficulty in swallowing and vomiting for 1 day. History dates back to 5 years with progressive regression of developmental milestones, frequent falls during walking, clumsy staggering gait, difficulty in climbing stairs, difficulty in holding objects and deterioration of hand writing. Patient's maternal uncle also had similar complaints which started at the age of 5 years. Unlike usual presentation of Friedrich Ataxia, this patient never noticed any change in perception of sensations, had no difficulty in hearing, no visual abnormalities or any other problems.

Examination showed vitally stable, bed ridden boy with marked pallor, aggressive scoliosis, visible muscular atrophy, diminished muscle power (3/5), areflexia of limbs, extensor plantar response, loss of co-ordination in all limbs, intention tremors, past pointing, dysdiadochokinesia, positive Romberg sign and scanning speech. The child was unable to stand or walk without support. Cranial nerve functions and sensations were intact. Fundus was healthy looking. Diagnosis of Friedreich's ataxia was made on the basis of history and examination as genetic confirmation facilities were not available. MRI Scan, Electromyography EMG and Nerve conduction velocity tests (NCV) were done. NCV showed that response to electrical stimulation is smaller in amplitude in our patient than normal individuals. Echocardiography showed no conduction blocks. Thyroid function tests and blood sugar profile were normal.

Physiotherapy and speech therapy was started. Parents were counseled about the disease, its mode of inheritance, its management, possible outcome and necessity for prolonged follow up for timely diagnosis of complications.

Discussion

Friederich's ataxia is an autosomal recessive disorder. Its gene is found on chromosome 9y13.[7] Carrier rate is 1:150. It usually occurs at 20-30 years but may manifest as early as 5-15 years. There is no sex predilection. It usually has neurologic, cardiac, skeletal and endocrine manifestations; most common of which is ataxia, weakness, spasticity, areflexia, extensor planter response, cerebellar lesion manifested as nystagmus, fast saccadic eye movements, truncal ataxia (drunken gait), sensory disturbances including loss of vibratory and proprioceptive sensations and cranial nerve palsies.

These neuronal findings are due to progressive degeneration of areas including dorsal column, posterior column, corticospinal tract, spinocerebellar tracts and other parts of spinal cord and cerebellum. Cranial nerve nuclei and nerve fibers are also targets of FA.[8,9,10] Neurological presentation in our patient was not typical of disease but was comparable to another case report published by Nitte University Journal of Health Science.[11]

About 75-91% of patients develop cardiac abnormalities which include dilated cardiomyopathy, hypertrophied cardiomyopathy, conduction defects and heart murmurs. It presents as chest pain, easy fatigability, dyspnea, headaches, palpitations, bipedal edema and sometimes unexplained fainting episodes. Echocardiography is recommended at every visit and the patient managed according to the results. Despite typical presentation and progression of signs and symptoms, there were no such complications in our patient.[12,13]

60-79% of patients develop skeletal abnormalities, most significant of which is progressive scoliosis, which is painful and associated with breathing difficulties as it interferes with normal lung expansion. In contrast to our case, other common skeletal abnormalities are Pes cavus (high arched foot) and inversion (inward deviation or deflection of forefoot). Both these conditions result into painful walking and repeated trauma to feet which ultimately cause formation of calluses and ulcers that have a long list of complications of their own.[14]

Diabetes mellitus develops in 10-20% of patients depending upon presence or absence of other precipitating factors of diabetes. This is due to FRATAXIN deficiency in pancreas which reduces insulin production and secretion resulting in progressive insulin insufficiency. Diabetes mellitus due to Friedreich's ataxia is associated with higher ophthalmological complications.[15] There was no such complication in our patient.

Diagnosis usually begins with detailed history of onset of symptoms and assessment of family history. In physical examination special attention is given to neurological performance of patient.

Confirmation is through genetic diagnosis by looking for FRATAXIN mutation. This is the most sensitive and specific test that can confirm or rule out FA. Other investigations, as mentioned in our case, are supportive to diagnosis.

Prognosis of FA is poor. Health related quality of life is significantly worse in these individuals.[16] No clear relationship is found between FRATAXIN mutation and cardiac dysfunction. Further research is needed in this regard.[17]

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