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Case Report

Early onset ataxia with Marfanoid features a new varient of Friedreich s ataxia

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ABSTRACT

A young male with ataxia since early childhood with Marfanoid features, normal intellect and no biochemical abnormality is reported. The syndrome has partial resemblance with previously described syndrome of arachnodactyle, cerebellar ataxia and other features, what has been named as "Bhaskar Syndrome". The documentation of such rare entities is worth for future research.

1. Introduction

Friedreich's ataxia, an autosomal recessive disease has prevalence of between 1 and 2 per 100,000, symptoms and signs being progressive ataxia, absent tendon reflexes in the legs, distal impairment of position and sense of vibration, Babinski reflexes, and dysarthria.[1] Other signs may be present, such as pes cavus, scoliosis, diabetes mellitus, and cardiomyopathy. Some non ataxic symptoms are also being described in it.[2] Some metabolic and morphological features like syndectyle etc are very rarely described and only from Indian subcontinent.[3,4] In fact the case reports were having some additional features to that of Merinesc- sJogren syndrome.[5] In some cases odd features like exaggerated jaw reflex was not against the diagnosis of Friedreich's ataxia.[6]

2. Case Report

A 15 year right handed boy, born of non-consanguineous marriage and normal home delivery, first of 3 sibs, student of eighth standard with average school performance, presented with history of unsteadiness of gait, deformities of feet and slurring of speech since early childhood. The patient started walking at the age of 2 years; thereafter used to walk and run normally but, with occasional falls. At the age of 6-7 years patient had febrile illness after which the difficulty in walking increased gradually with swaying on both sides while walking and turning with feeling of stiffness in both lower limbs and the patient was able to run normally as compared to his age matched classmates. Simultaneously difficulty in speaking was also noticed since early childhood. The patient takes time to initiate speech, is slow to speak, has to force more for speaking loudly and speaks the words syllable by syllable. Parents of the patient noticed at the age of 6-7 years that his fingers are longer and feet are deformed; whom they related to repeated falls and tall slender body. There was no history of weakness or tremulousness in upper limbs, difficulty in deglutition, hearing, and visual perception, vertigo, neck pain, occipital headache etc. There was no history of repeated infections. Family history was noncontributory and there was no illness to mother during antenatal period.

On examination vital parameters were normal. Regarding morphological features the neck was shorter, tall fingers, kyphoscoliosis, pes cavus with fanning of toes with high arched palate. His height measured 168cms, arm span 175 centimeters and the crown to symphysis pubis 78 centimeters. Liver and spleen were not palpable. On detailed

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neurological examination, higher mental functions were normal, speech was slurred, low pitch with scanning. There was bilateral coarse horizontal nystagmus with broken persuits. On fundus examination there was bilateral temporal pallor. Other examination was normal regarding cranial nerves. There was no K.F. ring on corneal examination by slit lamp. On motor examination the muscle bulk was normal, tone was normal in upper limbs and increased/spastic in lower limbs. Power was 5/5(Medical Research Council grading) in upper limbs and 4+/5 at hip and knee, 3/5 at ankle dorsiflexers and 4-/5 at ankle planter flexor. Deep tendon reflexes were all exaggerated except ankle with bilateral flexor planter response. The cerebellal signs were present in form of positive finger nose finger, finger to finger test, dis-diadokokinesia and knee-heel shin test with bilateral gross swaying on tendom walk. Touch, pain, temperature and fine cortical sensations were normal but position and vibration sensations were decreased by 25-30% in lower limbs.

On investigation the routine hemogram was normal, urine complete and microscopic examination, fasting blood sugar, kidney and liver function tests were normal. Copper profile was normal. There were no acanthocytes on freshly prepared peripheral blood smear, and nerve conduction studies were within normal limits. CT scan of the brain including cranio-vertebral junction was normal. Cyanide-nitroprusside urine test was negative.[7] Genetic analysis couldn't be performed because of lack of facilities and affordability.

3. Discussion

The present case who presented with early onset ataxia with corticospinal signs and had Marfanoid features is reported because of extreme paucity of such combinations being described in literature. Five patients were earlier described by Bhaskar from Chennai-India with positive family history.[3,4] Friedreich's ataxia, being more prevalent remains a possibility in this case along with some additional features like that of Bhaskar syndrome and Merinesco S Jogren syndrome.[3–5] The additional features were arachnodactyle, aminoaciduria, congenital cataracts and delayed milestones. My case may be a mere association of early onset ataxia and Marfanoid syndrome or a new variant of Friedreich's ataxia and will be worth documentation in an esteemed journal. The trial of certain therapeutic modalities like idebenone and riluzole are likely a promising drugs for Friedreich's ataxia and other diverse ataxias respectively.[8–10] Recognition of rarely observed morphological features may help narrow down the differentials and plan therapies in resource poor settings.

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5. Conflict of Interest

The authors declare that they have no conflict of interest.

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