REHABILITATION APPROACHES OF A BANGLADESHI BOY WITH ATAXIA TELANGECTASIA: A CASE REPORT

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Abstract

Ataxia-Telangiectasia (AT) is a rare form of neurodegenerative disorder of hereditary origin. In Bangladesh we don't have any epidemiological study. Till today there is no gold standard of the diagnosis, further to its wide clinical heterogeneity, it often leads physicians to an incorrect or missed diagnosis. So is the importance of the insight into this disease. We report a 10 years old male child who presented with history of difficulty in posture and walking associated with frequent fever, delayed slurred speech and abnormal eye movements. Physical examination revealed delayed milestones of development involving neck control, sitting, walking, unsteady supported swaying gait and speech language dysfunctions. MRI of brain showed cerebellar atrophy and a raised serum Alfafeto protein level. The child was diagnosed as a case of AT and was presented in a clinical meeting at Bangabandhu Sheikh Mujib Medical University for discussions and further managements including medical rehab. We offered him a course of combined rehabilitation therapy program and discharged home.

Key words

Ataxia Telangiectasia; BSMMU; Rehabilitation.

Introduction

Ataxia Telangiectasia (AT) is an autosomal recessive disorder characterized by progressive cerebellar ataxia, occulocutaneous telangiectasia, immunodeficiency, recurrent sinopulmonary infections, premature aging, cancer susceptibility and radiation sensitivity[1]. The initial clinical description of A-T was reported by Syllaba and Henner during 1926 and a Belgian neuro-pathologist D Louis-Bar in 1941 reported initial cases of AT describing the syndrome[2]. AT is often referred to as a genome instability or DNA damage response syndrome and most of the cases are due to mutations of the gene (ATM). In Bangladesh we do not have any epidemiological study. World-wide prevalence is estimated between 1 in 40,000 to 1 in 100,000 live births[3]. Till today there is no gold standard diagnosis for AT has been stablished. Characteristic clinical presentations in most of the cases are progressive early onset cerebellar ataxia, oculomotor apraxia, oculocutaneus telangiectasia, recurrent sinopulmonary infections and proneness to cancer[3]. Other features include premature aging with graying of the hair, insulin-resistant diabetes mellitus[4]. A-T causes increased radio sensitivity may be a defect in their ability to respond to DNA damage rather than a defect in their ability to repair it. Almost all cases (99%) are associated with ATM mutations that cause ataxia-telangectasia[5]. Life expectancy of people with A-T is approximately 25 years on average.

Case Report

Master 'S' of 10 years old Bangladeshi boy presented on 11.03.2018 at the Department of Physical Medicine and Rehabilitation BSMMU, Dhaka with the complaints of unsteady walking with frequent falls associated with delayed started slurring of speech for 9 years. He was coming from a lower-middle class family with history of consanguinity of marriage. The child was born as a low birth weight baby (Weighing about 1 kg) at 9 months of gestational age with an uneventful birth history. His all milestones of development were delayed like neck control at 1 year, sitting at 1½ year, walking at 3 years, speaking at 4 years of age with stereotyped pattern his speech was more labored and less but last one year he was unable to speak. Previous medical record shows that he was diagnosed as a case of ataxic cerebral palsy with pulmonary TB taking of anti TB drugs for 1.5 years.

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His walking pattern has been deteriorated for last 6 months. Since the patient's attendant gives no history of convulsion, fever, trauma, back pain, bladder bowel abnormality. There was no history of radio sensitivity.

Physical examination revealed vital signs were normal and stable and his height and weight was less than 50th percentile of normal. His developmental milestone were delayed. Over the bulbar conjunctiva of both eye (Figure 1) telangiectasia were present with absence of nystagmus or squint. On systemic examination his standing posture were stooping with head and shoulder sunk forwards made an unstable ataxic gait. The arms and leg were thin associated with reduced muscle tone and power (4/5) in all 4 limbs. All deep tendon reflexes with flexor planter reflex bilaterally. No primitive reflex were found. Dysdiadokinesia and Romberg sign were positive. Functional Independence Measure (FIM) score was 55 out of 126 at admission.

Laboratory investigations revealed normal complete blood counts and elevated serum Alfa- feto protein {40.4 ng/ml (Normal:10ng/ml)}, CPK: 64U/L, Ig A:<0.262g/L,MRI of brain revealed generalized cerebellar atrophy. Identification of AT Mutation gene or evaluation of AT protein could not be done.

The patient was given supportive treatments including antibiotics, clonazepum, baclofen and pregabalin. A course of combined controlled rehabilitation therapy program involving physiotherapy, occupational therapy, speech language therapy was offered and discharged home. After one month he came for follow up and we found condition is static and speech capability has been improving.

Fig 1 : Telangectasia at bulbar conjuctiva

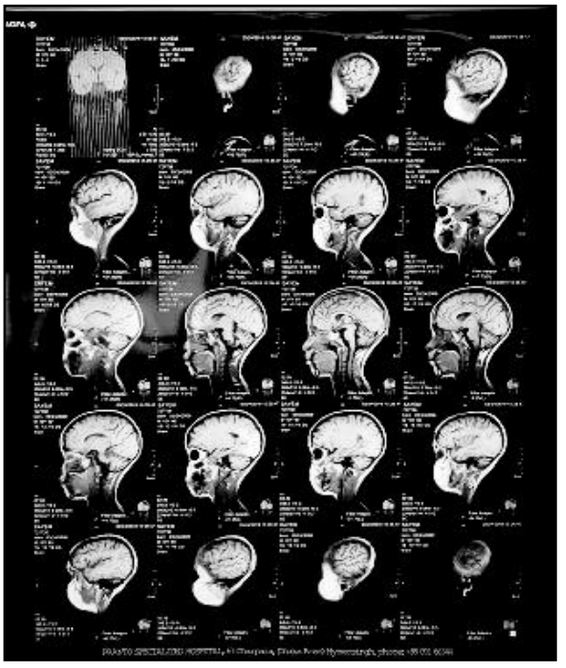

Fig 2 : Cerebellar atrophy on MRI

**Telangiectasia at bulbar conjunctiva was confirmed by ophthalmologist on slit lamp examination. Cerebellar atrophy was confirmed by Radiologist.

Discussion

AT is a rare multisystemic autosomal recessive neurodegenerative disease (1:10,000-1:40,000 new births per year over the world)[6]. According to an Iranian study of 104 patient, the median age of the patient was 9 years and our patient is 10 years old[7]. Characteristic clinical presentations in most of the cases are progressive early onset cerebellar ataxia, oculomotor apraxia, oculocutaneus telangiectasia, recurrent sinopulmonary infections which matched with this case[8]. Characteristic facies and postural attitude observed in this patient appeared to be primarily part of cerebral symptomatology[9]. Increased infections due to immune system disorder appear in about 70% of the case[10]. Our patient also have history of repeated respiratory tract infection. Elevated serum alpha-fetoprotein was described as useful screening test for A-T, reduced serum Ig A levels[11]. Our patient has MRI findings of cerebellar atrophy which seems like trifoliate appearance, suggesting to the point of possibility of AT[12]. The most common misdiagnosis was cerebral palsy[13]. A study showed that, AT cells fail to respond to the X-ray induced DNA damage that is normal cells inhibits

initiation of replicons and blocks chain elongation. One possibility is that AT cells lack a factor or process that in normal cells delays replication after irradiation[8]. Dietary changes, including the use of a gluten-free diet to treat ataxia, were outside the scope of this systematic review. On functional assessment he feels difficulty in speech, performing activities of daily living, walking, for these we gave him a comprehensive rehabilitation therapy which include following physical measures and exercise program. So far there is no curative strategy for this disease exists. Treatment has focused on slowing the progress of the neurodegeneration[14]. Treatment of AT is symptomatic and supportive[15]. Only the conservative treatment to avoid respiratory tract infection. Physical and speech therapy are another option[16]. For mobility improvement we gave him exercise program like strengthening exercise of both lower limb, stretching exercise of all four limbs and core muscle exercise like pelvic tilting. We also guided him for gradual and graded gait retraining and advised for home modification by widening of passage way, removal of obstacles from the path and fall prevention by proper lighting of the passage way, use of anti-slippery mattress and hand rail in bathroom. He was advised to have self-care activity with partial assistance. Speech and swallowing therapy was given. We also advised to admit him in specialized school for improve his communication ability and social interaction. In addition, historical treatment approaches, such as the use of acetazolamide for the treatment of EA2, can have clinical value even in the absence of clinical trial evidence[17]. The two most common causes of death are chronic lung disease (About one-third of cases) and cancer (About one-third of cases)[18]. As this type of patient prone to develop cancer and hypersensitive to ionizing radiation, we counsel him to avoid unnecessary repeated X-ray. Gradually feeding and swallowing become difficult for a patient with AT as they get older[19].

Conclusion

As ataxia telengectasia was less common, a tremendous amount has been learned about the clinical manifestations of AT, and advances in clinical care have significantly improved the average life span of individuals suffering from this disease. Multidisciplinary team approach is needed for the management of this disease.

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Contribution of authors

FN: Conception, designing, drafting and final approval
MFP: Designing, citing references and final approval.
HI: Design, collect other information and final approval.
TU: Conception, critical revision and final approval.
MSR: Designing, citing references and final approval.
MNK: Manuscript drafting, final approval.

Disclosure

All the authors declared no competing interest.

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