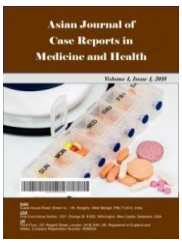

Asian Journal of Case Reports in Medicine and Health

1(1): 35-37, 2018; Article no.AJCRMH.46369

Tremulousness in a 7-yrs Old Child Diagnosed with Ataxia Telangiectasia: A Case Report

Abhijit Dutta[1*] and Pralay Sharma[2]

[1]Department of Organon of Medicine, National Institute of Homoeopathy, Kolkata, India.
[2]National Institute of Homoeopathy, Kolkata, India.

Authors' contributions

This work was carried out in collaboration between both authors. Both authors read and approved the final manuscript.

Article Information

DOI: 10.9734/AJCRMH/2018/46369
Editor(s):
(1) Dr. Arun Singh, Professor, Department of Community Medicine, Rohilkhand Medical College & Hospital Bareilly, Uttar Pradesh, India.
Reviewers:
(1) David Vance, University of Alabama at Birmingham, USA.
(2) Alina S. Fedorova, Belarusian Research Center for Pediatric Oncology, Belarus.
(3) Luis Rafael Moscote-Salazar, University of Cartagena, Cartagena de Indias, Colombia.
Complete Peer review History: http://www.sciencedomain.org/review-history/28205

Case Study

Received 14th October 2018
Accepted 30th December 2018
Published 10th January 2019

ABSTRACT

Ataxia telangiectasia is a rare autosomal recessive neurodegenerative disorder primarily characterized by cerebellar degeneration manifested as ataxia, telangiectasia, immunodeficiency, and cancer susceptibility and radiation sensitivity. Clinically A-T cases presents itself with a variability of symptoms but basic primary presentations are almost always present. This case demonstrates a 7yrs old girl who presented with tremulousness of limbs, difficulty to move the eye ball with command and dysarthria. On further clinical examination, a plain MRI of Brain and other relevant investigations suggested this as a case of A-T. Thus rationale of this typical case report gives a glimpse about a variant of A-T which is often misdiagnosed as other ataxic disorders.

Keywords: Ataxia telangiectasia; neurodegenerative disorder; ataxic disorders.

1. INTRODUCTION

Ataxia telangiectasia or Louis-Bar syndrome is a neurodegenerative disorder which is found to be autosomal recessive and primarily characterised by Ataxia, abnormal control of eye movement, and postural instability with variable symptoms like telangiectasia, dysarthria etc [1]. These

**Corresponding author: E-mail: drabhijitdutta1@gmail.com;*

symptoms are most likely to appear in the first year of life when the child learns to sit, walk and speak [2]. But sometimes the child learns for a period and then impairment starts and continues. A significant relationship has been found in pathological mutations in ATM gene and the development of A-T [3]. Sometimes some serological markers like Alpha Fetoprotein (AFP) also increased [4]. There are several other neurological conditions that the physician has to consider while diagnosing ataxic disorders such as cerebral palsy, autosomal recessive cerebellar ataxia, etc [5]. Several studies have found that A-T patients are prone to develop cancer and other immunological disorders, so proper diagnosis is needed not only for the symptomatic intervention but also for prognostic prevention [6]. A-T is still counted as a rare neurological condition having its world-wide prevalence estimated to be between 1 in 40,000 and 1 in 100,000 live births but definitely it is a serious concern for physicians because of its complexity and uncertainty in diagnosis [7].

2. CASE REPORT

A 7yrs old female child was brought in the Out Patient Department of National Institute of Homoeopathy, Kolkata with tremulousness of limbs, particularly while she was trying to take food into her mouth since 3 yrs of age. This problem was continuously increasing and suddenly observed by her parents. Before that she was apparently normal. After a few months, when the first symptom developed, she had difficulty in talking and difficulty in walking and since then, all those symptoms increased in severity. Her parents then noticed that she becomes easily irritable and withdraws herself from social affairs.

No significant intrauterine, past and family history were found.

On clinical examination vitals were normal (B.P.-126/74 mm of Hg, Pulse rate-78/ min and resp. rate-19/ min), anxious face, and oxygen saturation was 98%, No sign of icterus, clubbing, lymphadenopathy or neck vein engorgement was noted.

On neurological examination, slurred speech, hypophonia, restricted eyeball movement, impaired finger-nose-finger test, and wide-based imbalanced gait were noted but motor functioning was normal. Telangiectasia (dilated small blood vessels) in conjunctiva was noted during the eye examination (Fig. 1).

On laboratory investigations serum AFP was significantly raised (183.3 ng/ml; normally <5.3 ng/ml) but other haematological tests like complete blood count, liver function test, lipid profile, Plasma ceruloplasmin, Thyroid profile was normal.

Plain MRI brain scan revealed prominent superior cerebellar folia with widened cerebellar sulci favouring cerebellar atrophy.

Fig. 1. Telangiectasia in conjunctiva

3. DISCUSSION AND CONCLUSION

Although A-T is very rare disorder, it should be considered as a serious concern. Classically ataxia-telangiectasia (A-T) is characterized by ataxia primarily due to cerebellar dysfunction, telangiectasias, immunodeficiency, and an increased risk for malignancy, especially leukemia and lymphoma [1,8]. Lymphoid malignancy can alone may be the initial presenting feature of A-T and primarily described in some children [9]. In later life usually there is delayed puberty and gonadal problems [10]. In this case ataxia along with dysarthria, telangiectasia in conjunctiva and elevated serum AFP served as a lead to do further investigations and favoured the diagnosis of A-T. Every physician will have to do a proper examination and evaluation of the case. This case surely demonstrates a presentation of A-T which may help to evaluate such cases in future.

CONSENT

As per international standard or university standard, patient's consent has been collected and preserved by the authors.

ETHICAL APPROVAL

It is not applicable.

ACKNOWLEDGEMENT

We acknowledge *Dr. Abhijit Chattyopadhyay,* Director, NIH, Kolkata and we are heartily thankful to all the members of *BIN, Kolkata,* for doing all the preliminary investigations and progression of the diagnosis as the patient also visited there beforehand.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

REFERENCES

1. Riboldi GM, Frucht S. Ataxia telangiectasia (Louis-Bar Syndrome) [Updated 2018 Oct 27]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2018. Available:https://www.ncbi.nlm.nih.gov/books/NBK519542/
2. Rothblum-Oviatt C, Wright J, Lefton-Greif MA, McGrath-Morrow SA, Crawford TO, Lederman HM. Ataxia telangiectasia: A review. Orphanet Journal of Rare Diseases. 2016;11(1), 159. DOI: 10.1186/s13023-016-0543-7
3. Shiloh Y. ATM and related protein kinases: safeguarding genome integrity. Nat Rev Cancer. 2003;3(3):155–68. DOI: 10.1038/nrc1011
4. Stray-Pedersen A, Borresen-Dale AL, Paus E, Lindman CR, Burgers T, Abrahamsen TG. Alpha fetoprotein is increasing with age in ataxia-telangiectasia. Eur J Paediatr Neurol. 2007;11(6):375-80.
5. Anheim M, Tranchant C, Koenig M. The autosomal recessive cerebellar ataxias. N Engl J Med. 2012;366(7):636–46. DOI: 10.1056/NEJMra1006610.
6. Nowak-Wegrzyn A, et al. Immunodeficiency and infections in ataxia-telangiectasia. J Pediatr. 2004;144(4):505–11. DOI: 10.1016/j.jpeds.2003.12.046
7. Gilad S, Bar-Shira A, Harnik R, Shkedy D, Ziv Y, Khosravi R, Brown K, Vanagaite L, Xu G, Frydman M, Lavin MF, Hill D, Tagle DA, Shiloh Y. Ataxia-telangiectasia: founder effect among north African Jews. Hum. Mol. Genet. 1996;5(12):2033-7.
8. Bielorai B, Fisher T, Waldman D, Lerenthal Y, Nissenkorn A, Tohami T, et al. Acute lymphoblastic leukemia in early childhood as the presenting sign of ataxia-telangiectasia variant. Pediatric Hematology and Oncology. 2013;30:574–582.
9. Schoenaker MH, Suarez F, Szczepanski T, Mahlaoui N, Loeffen JL. Treatment of acute leukemia in children with ataxia telangiectasia (A-T). European Journal of Medical Genetics. 2016;59(12): 641-646.
10. Ehlayel M, Soliman A, De Sanctis V. Linear growth and endocrine function in children with ataxia telangiectasia. Indian J Endocrinol Metab. 2014;18(Suppl 1):S93-6.

Peer-review history:
The peer review history for this paper can be accessed here:
http://www.sciencedomain.org/review-history/28205