CASE REPORT

Ataxia Telangiectasia with Giant Suprasellar Arachnoid Cyst - A Case Report and a Brief Review

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ABSTRACT

Ataxiatelangiectasia (A-T) is an infrequent genetic neurodegenerative disorder inherited autosomal recessively. It is mainly characterized by early-onset progressive cerebellar ataxia and dilated capillaries in the oculocutaneous regions especially conjunctivae so-called telangiectasia. A-T is a multisystem disorder and requires multi-disciplinary approach to management. Diagnosis is difficult in some cases because presentation is not in the same manner and showing a phenotypic spectrum. In atypical cases serum immunoglobulins and alfa fetoprotein are normal and telangiectasia is absent. We present a 5.5-year-old boy with progressive cerebellar ataxia and history of repeated sino-pulmonary infections that was homozygote for ataxia-telangiectasia mutated gene and had a giant arachnoid cyst in left hemisphere. It is important to keep in mind those cases with ataxia and repeated sino-pulmonary infections may be ataxia telangectasia patients. Genetic study is helpful and confirms the diagnosis by showing ataxia-telangiectasia mutated gene.

Introduction

Ataxia telangiectasia (A-T) [OMIM#208900] is a rare genetic neurodegenerative disorder (1). It is caused by an autosomal recessive mutation on ataxia-telangiectasia mutated (ATM) gene located on the chromosome 11q22.23 (2). This progressive inherited disorder characterized by cerebellar ataxia and telangiectasia mainly on bulbar conjunctiva. This mutation also involves other organs with variable features in different

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ages even in a family. Immune deficiency, malignancy, repeated sinopulmonary infections, radiosensitivity and hematologic malignancies are well recognized and described in many studies (3). ATM gene is a checkpoint in cell regeneration and has an important role in DNA repair. Patient with this chromosomal instability syndrome are radiation sensitive and ionizing radiation methods are not allowed routinely due to risk of hematologic malignancies especially lymphoma and leukemia (4).It is important to know that A-T is a twisted multi organ involvement and patients have variable features (5).Signs and symptoms are presenting differently in every age and laboratory findings are not conclusive in every case. Patients may start walking in normal age and telangiectasia is not seen in all cases so diagnosis may be a challenge when the presentation is not the classic type (6).We present a 5.5-year-old boy with genetically confirmed A-T who referred to our clinic with progressive cerebellar ataxia but without telangiectasia that had a giant suprasellar arachnoid cyst in left hemisphere.

Case Presentation

Our Patient was a 5.5-year-old Iranian boy who was born to non-related healthy parents after preterm delivery by cesarean section due to twin pregnancy with gestational age 32 weeks and birth weight 2300 grams. Perinatal insult and admission history were negative. He has presented with unsteady-ataxic gait since 2 years old with history of motor and language delay. Gradually, his gait was more ataxic and his speech fluency was abnormal until now. On neurologic examination, we found ataxic gait, slurred speech, oculomotor apraxia, positive finger to finger test, fine intention tremor of hands and horizental nystagmus. Deep tendon reflexes (DTRs) were normal. Telangiectasia in bulbar conjunctiva and pinna or nares was absent in our case (Fig 1).There is no abnormality on cardiovascular and respiratory systems in our patient. During routine laboratory investigations, complete blood count (CBC), electrolytes, thyroid function tests, erythrocyte sedimentation rate (ESR) was normal. Alfa fetoprotein level was high. (133ng/ml, Normal<8.5ng/ml). Based on these manifestations, whole exome sequencing (WES) was performed and a frameshift ATM gene mutation (pathogenic variant) was revealed and A-T was documented. He was evaluated for his immune system. His blood flowcytometry and complement system were normal. His serum immune globulin profile revealed normal IgG and IgM levels, but decreased IgA (2.6 mg/

Figure 1. Our case without ocular telangiectasia

dl) (with normal range of 25-154 mg/dl) which was unusual finding. Brain MRI was performed and we found cerebellar atrophy as expected finding associated with a very large left temporal suprasellar arachnoid cyst without mass effect as unexpected radiologic finding in this patient (Fig 2). The frequency of sino-pulmonary febrile infections was low in our case and he didn't need intravenous immunoglobulin (IVIG). Although, his twin sister with A-T has suffered from frequent sino-pulmonary infections since one year ago that after starting of IVIG since about six months ago, the frequency of these febrile infections was markedly decreased. Fortunately, there is no evidence for malignancy in our patient.

The patient and family described in this case report provided informed written and signed consent to participate of the case. There is no information revealing the patient's identity in this paper. The paper was written with ethical considerations in accordance with the Medical Ethics Group of Tehran University of Medical Sciences.

Discussion

Ataxia telangiectasia (A-T) is a rare autosomal recessive neurodegenerative disorder. ATM gene mutation on the chromosome 11q22.23 makes a multi organ involvement with different features even in affected twins. Features appear in spectrum with different presentation according the age so diagnosis is challenging in atypical forms of A-T (7).

A-T is characterized by cerebellar ataxia and oculocutaneous telangiectasia. Children may have not obvious gait disturbances till 4-5 years old and telangiectasia may not appear until the teenage years. Our patient has had global developmental delay since 2 years old without telangiectasia until 5.5 years. In this deceptive disease combination of neurologic features, postural instability, telangiectasia and repeated infections should propose A-T. Laboratory tests such as IgG and IgA deficiency, lymphopenia especially T lymphocytes and increased alpha-fetoprotein levels can help to confirm the

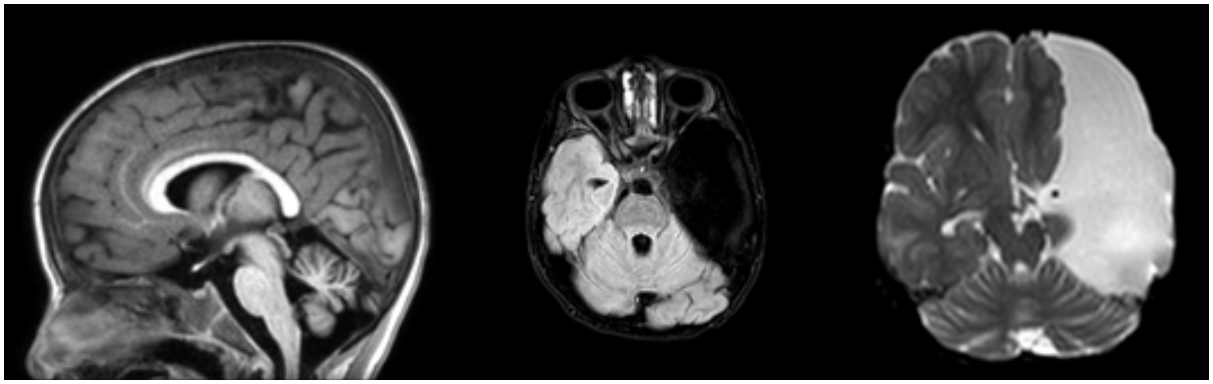

Figure 2. Sagittal FLAIR MR Image showing cerebellar atrophy (Left). Axial FlAIR MR Image showing a large suprasellar cystic lesion (arachnoid cyst) in left temporal region without mass effect (Middle). Axial T2 MR Image showing the same large arachnoid cyst (Right)

diagnosis. In our case, IgG was normal and only IgA was severely deficient. Also, there was no lymphopenia in our patient that is unusual finding in A-T. Alfa fetoprotein value was high. Genetic study is now available in advanced laboratories and makes the diagnosis certain as in our study (8).

A-T Patients are radiation sensitive because DNA repair mechanisms and genomic stability is impaired in ATM mutation. They are susceptible to hematologic malignancies such as leukemia and lymphoma. X-ray modalities are not performed because make patients more prone to malignancies. Nowadays impressive efforts and studies have been performed to improve our understanding of brain structural abnormalities with MRI. Progressive cerebellar atrophy predominantly in vermis, fourth ventricle dilatation, sinusitis and enlarged cisterna magna has been reported before. In cerebral hemisphere deep white matter, ischemic lesions due to small vessel involvement have been mentioned too(10). In our case, progressive gait disturbance and cerebellar features with history of motor delay arised our suspicion for hereditary ataxias such as A-T and A-T like disorders. Despite absence of parental consanguinity and telangiectasia, precise history and physical examination help to make the challenging diagnosis. Brain MRI showed a very large suprasellar arachnoid cyst in left hemisphere that as far as we know, it has not been reported before in patients with A-T. Also, his twin sister has the same picture and she receives IVIG monthly because of repeated sino-pulmonary infections.

Patients with A-T need Multi-disciplinary treatment. Intelligence quittance (IQ) and school performance gets hurt as disease progresses. Speech therapy is essential while the child grows because dysarthria worsens after 5 years old (9).Infection control is important. Pulmonary insufficiency and sinusitis are important causes of disease morbidity and mortality. IVIG supports immune system and improves quality of life (10). Fortunately; our patient didn't need IVIG. Cancer screen is done by checking hematologic evaluation. Radiation is forbidden because DNA breakdown can cause malignant tumors. Mobility should be maintained as much as possible. Nutritional support and growth monitoring are noted because patients in older age, have feeding problem, repeated aspiration, pulmonary infection and failure to thrive (10).

In conclusion

A-T is a rare genetic multisystem disorder. Features are not presenting in a same picture even in a family and doctors may not be familiar in cases with atypical presentations such as absence of telangiectasia, normal alpha fetoprotein level, normal immunoglobulin level and slow course of the disease. We presented a case with progressive cerebellar ataxia without oculocutaneous telangiectasia. Our patient had a giant arachnoid cyst that has not been presented before in patients with A-T. Genetic study confirmed the diagnosis.

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Authors' contributions

Ashrafi stated that the report of this patient can be useful in identifying and treating theses patients faster, and the patient was identified by him. Nikkhah and Heidari helped in writing

and editing the article, and data was collected by ShahbodaghKhan, Sinaei and Aziz-ahari. Yousefimanesh helped in writing and correcting the article, recording the information and data collection.

Conflict of interest

The authors declare that they have no conflict of interest.

References

1.Rothblum-Oviatt C, Wright J, Lefton-Greif MA, McGrath-Morrow SA, Crawford TO, Lederman HM.Ataxia telangiectasia: a review. Orphanet J Rare Dis. 2016 Nov 25;11(1):159. doi: 10.1186/s13023-016-0543-7.

2.Sirajwala AA, Khan S, Rathod VM, Gevariya VC, Jansari JR, Patel YM.Ataxia-Telangiectasia: A Case Report and a Brief Review. Cureus. 2023 May 22;15(5):e39346. doi: 10.7759/cureus.39346. eCollection 2023 May.

3. Taylor AM, Groom A, Byrd PJ. Ataxia-telangiectasia-like disorder (ATLD)-its clinical presentation and molecular basis.DNA Repair (Amst). 2004 Aug-Sep;3(8-9):1219-25. [PubMed]

4. Van Os NJH, Haaxma CA, van der Flier M, Merkus PJFM, van Deuren M, de Groot IJM, et al. Ataxia-telangiectasia: recommendations for multidisciplinary treatment. Dev Med Child Neurol. 2017 Jul;59(7):680-689.

5. Raval DM, Rathod VM, Dobariya RK, Dave MP, Patel NS .A rare phenotype of inherited cerebellar ataxia. Cureus. 2022 Sep 6;14(9):e28831.

6. Amirifar P, Ranjouri MR, Lavin M, Abolhassani H, Yazdani R, Aghamohammadi A. Ataxia-telangiectasia: epidemiology, pathogenesis, clinical phenotype, diagnosis, prognosis and management. Expert Rev Clin Immunol. 2020;16:859-71.

7. Hadi Babikir HE. Classic ataxia-telangiectasia in a Sudanese boy: Case report and review of the literature. Sudan J Paediatr. 2011;11(1):60-3. PMID: 27493307; PMCID: PMC4949784.

8. Kashimada A, Hasegawa S, Nomura T, Shiraku H, Moriyama K, Suzuki T, et al. Genetic analysis of undiagnosed ataxia-telangiectasia-like disorders. Brain Dev. 2019 Feb;41(2):150-157. doi: 10.1016/j.braindev.2018.09.007. Epub 2018 Oct 6. PMID: 30301590.

9. Dineen RA, Raschke F, McGlashan HL, Pszczolkowski S, Hack L, Cooper AD, et al. Multiparametric cerebellar imaging and clinical phenotype in childhood ataxia telangiectasia. Neuroimage Clin. 2020;25:102110. doi: 10.1016/j.nicl.2019.102110. Epub 2019 Nov 26. PMID: 31855653; PMCID: PMC6926372.

10. Riboldi GM, Samanta D, Frucht S. Ataxia Telangiectasia. 2023 Jul 3. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2023 Jan–. PMID: 30137827.