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A Rare Case of Ataxia Telangiectasia in a 5-Year-Old Male: A Multisystem Challenge

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ABSTRACT
Ataxia Telangiectasia (AT) is a rare, autosomal recessive, neurodegenerative disorder primarily affecting children, characterized by progressive cerebellar ataxia, oculocutaneous telangiectasias, and immune dysfunction. A 5-year-old male child presented with speech slurring and gait imbalance from 1 year of age. Clinical and systemic evaluations revealed telangiectatic lesions, neurodevelopmental delay, and swaying while walking. Despite normal MRI and AFP levels, AT was suspected based on classical clinical findings. This report highlights the challenges in diagnosing AT in early stages and underlines the importance of a multidisciplinary approach.

Keywords: Ataxia Telangiectasia (AT), Telangiectasia, Cerebellar Ataxia, Neuro-degeneration, Immunodeficiency

I. INTRODUCTION

Ataxia Telangiectasia (AT) is a rare, autosomal recessive, neurodegenerative disorder caused by biallelic mutations in the **ATM (Ataxia-Telangiectasia Mutated)** gene, located on chromosome 11q22.3. The ATM gene encodes a serine/threonine protein kinase that plays a critical role in the cellular response to DNA double-strand breaks, maintaining genomic stability through cell cycle checkpoint control and DNA repair mechanisms. Deficiency or dysfunction of this protein leads to progressive neurological deterioration, immunodeficiency, and increased cancer susceptibility—primarily hematological malignancies [1,2] .

AT typically presents in early childhood, often between 1 and 4 years of age, with **progressive cerebellar ataxia** being the earliest and most prominent clinical feature. Children exhibit unsteady gait, frequent falls, dysarthria, and oculomotor apraxia. Over time, most patients develop **oculocutaneous telangiectasias**, particularly over the bulbar conjunctiva and sun-exposed skin. These vascular anomalies generally appear several years after the onset of ataxia [3] .

Immunodeficiency, particularly affecting both cellular and humoral immunity, is a defining feature of AT, predisposing patients to recurrent sinopulmonary infections. Laboratory findings often reveal **lymphopenia**, reduced serum levels of **IgA, IgG2, or IgE**, and increased serum **alpha-fetoprotein (AFP)** in over 85% of patients [4,5] . Despite its diagnostic utility, AFP may remain normal in early or variant presentations of the disease [6] .

Characteristic neuroimaging findings include **progressive cerebellar atrophy**, best visualized on MRI, even before significant clinical ataxia is apparent. Diagnosis is further confirmed by demonstrating absent or markedly reduced ATM protein activity, chromosomal breakage studies, and molecular genetic testing [7,8] .

Although there is currently **no definitive cure** for AT, multidisciplinary management involving neurologists, immunologists, physical therapists, and genetic counselors can significantly improve quality of life. Therapeutic strategies focus on infection control, nutritional support, surveillance for malignancies, and symptomatic care [9] .

This case report highlights the clinical presentation, diagnostic challenges, and neuroimaging features of a child diagnosed with Ataxia Telangiectasia, reinforcing the importance of early recognition and comprehensive care.

II. CASE PRESENTATION

A 5-year-old male child was brought to the Pediatric Ward of Karnataka Medical College and Research Institute (KMCRI), Hubballi, on 3rd April 2025, with parental concerns regarding persistent neurological symptoms. The primary complaints included slurring of speech and difficulty in maintaining balance while walking, both of which had been progressively evident since the child was approximately one year old.

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According to the caregivers, the child initially achieved early developmental milestones within normal limits; however, by the end of the first year, they began to observe speech articulation issues and an unsteady gait. Over time, these symptoms became more pronounced, with the child exhibiting frequent stumbling, swaying movements while walking, and delayed speech development. There was no history of seizures, trauma, or regression of previously acquired milestones. Given the chronic and progressively worsening nature of these symptoms, the child was referred for further evaluation and management.

Developmental History

- There **was no history of consanguineous marriage**, and the antenatal, natal, and immediate postnatal periods were uneventful, with no reported maternal infections, exposure to teratogens, or complications during delivery. The child was born full-term through an uncomplicated vaginal delivery with a normal birth weight and cried immediately after birth, indicating no perinatal hypoxic insult.
- Developmental **history revealed global delay**, particularly in motor coordination and language acquisition. Milestone achievement was notably delayed across gross motor, fine motor, speech, and social domains. The child exhibited delayed head control, late attainment of sitting and independent ambulation, and impaired fine motor skills such as grasping and self-feeding. Language development was significantly affected, with slurred and poorly articulated speech noted from early toddler years. Social interaction remained below expected levels for age, with limited peer engagement and reduced adaptive behaviors.
- Immunization **records confirmed the child was appropriately vaccinated** as per the national immunization schedule. However, given the suspected immunodeficiency associated with Ataxia Telangiectasia, further evaluation of humoral and cellular immune function was considered necessary.

General Examination and Clinical Observations

Parameter	Finding	Interpretation / Explanation
Temperature	36.6°C	Within normal range; afebrile status suggests no ongoing systemic infection at presentation.
Pulse Rate	96 bpm	Normal for age; reflects stable cardiovascular status.
Respiratory Rate	42 cpm	Slightly elevated for age, could be due to mild respiratory compensation or anxiety.
SpO_2 (Oxygen Saturation)	98% on room air	Indicates adequate peripheral oxygenation; no respiratory distress.
Weight	13 kg	Below expected for a 5-year-old; points to chronic undernutrition.
Anthropometry	Moderate Acute Malnutrition	Based on WHO growth charts; supports a diagnosis of nutritional compromise often seen in AT.
General Appearance	Alert, cooperative	Suggests preserved higher mental functions and stable systemic status.
Skin Findings	Telangiectatic lesions over conjunctiva and neck	Classic cutaneous sign of **Ataxia Telangiectasia**, due to dilated superficial blood vessels.
Lymphadenopathy	Not present	Absence of lymph node enlargement; no active infection or hematologic malignancy suspected.
Other Skin Lesions	None noted	No additional dermatological abnormalities were observed.
Nutritional Status	Evidence of undernutrition	Reinforces the need for nutritional intervention as part of multidisciplinary care.

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Conclusion Based on General Examination and Clinical Observations

The general examination and clinical observations offer critical early insights into the underlying diagnosis of **Ataxia Telangiectasia**. The child presented with **normal vital parameters**, indicating stable hemodynamic and respiratory status at the time of evaluation. However, **moderate acute malnutrition**, as evidenced by suboptimal weight-for-age, reflects the chronic nature of the condition and possible feeding or metabolic difficulties often associated with neurodevelopmental disorders.

Most notably, the presence of **telangiectatic lesions** over the conjunctiva and lateral neck regions is a defining clinical hallmark of Ataxia Telangiectasia, pointing to cutaneous vascular anomalies secondary to ATM gene dysfunction. These findings, in combination with the child's alert and cooperative general demeanor, suggest an early-stage systemic impact with preserved consciousness and cognition.

Together, the general and external clinical findings substantiate a preliminary diagnosis of **Ataxia Telangiectasia**, justifying further neurological, immunological, and genetic evaluations to confirm the diagnosis and guide comprehensive, multidisciplinary care.

Systemic and Neurological Examination

System	Findings	Interpretation / Explanation
Neurological	Gait ataxia (swaying while walking)	Classic sign of cerebellar involvement, commonly seen in early stages of Ataxia Telangiectasia.
Cardiovascular System	Normal S1 and S2 heart sounds; no murmurs	Indicates normal cardiac auscultation; no structural or functional abnormality suspected.
Respiratory System	Vesicular breath sounds; no added sounds	Normal lung fields; absence of wheeze or crepitations rules out respiratory infection.
Abdominal Examination	Soft, non-tender, no organomegaly	Suggests absence of hepatosplenomegaly or other intra-abdominal pathology.
Cranial Nerve Exam	CN I not assessed; CN II–XII intact	Normal cranial nerve function, excluding olfactory nerve which was not evaluated.
Higher Mental Functions	Alert and oriented	Suggests preserved cognition and consciousness; important baseline for neurological disorders.
ENT Examination	Audiological evaluation recommended	Due to known risk of sensorineural hearing loss or immune-related otitis in AT patients.
Dermatological Exam	Telangiectatic lesions confirmed over eyes and neck; no vascular malformations on USG neck	Ocular and cutaneous telangiectasias are hallmark signs of AT; no deeper vascular anomaly found.
Fundoscopy	Normal	Indicates no retinal hemorrhages, papilledema, or optic atrophy; helpful in excluding CNS pressure effects.

Conclusion Based on Systemic and Neurological Examination

The systemic and neurological examinations collectively highlight the classical clinical features of **Ataxia Telangiectasia**. The presence of **gait ataxia**, with swaying while walking, points toward **cerebellar involvement**, which is a hallmark neurological manifestation of the disorder. Despite this, higher mental functions and cranial nerve examination (CN II–XII) were found to be intact, indicating no gross cortical or cranial nerve deficits at this stage.

Systemic evaluation revealed **normal cardiovascular, respiratory, and abdominal findings**, suggesting that, at present, there are no overt complications affecting these organ systems. Dermatological assessment confirmed **telangiectatic lesions** over the conjunctiva and neck—classic cutaneous signs of AT—while ultrasonography ruled out deeper vascular

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malformations. A normal fundoscopic examination and audiological referral further support a multidisciplinary approach for ongoing assessment.

Collectively, these findings reinforce the suspicion of **early-stage Ataxia Telangiectasia**, with localized neurological involvement and preserved systemic stability.

INVESTIGATIONS

Investigation	Finding	Clinical Interpretation / Explanation
MRI Brain	Cerebellar atrophy	A key radiological hallmark of Ataxia Telangiectasia, indicating progressive cerebellar neurodegeneration.
Alpha-Fetoprotein (AFP)	Normal	Although AFP is often elevated in AT, normal levels do not exclude the diagnosis, especially in early/mild cases.
Hemoglobin (Hb)	Decreased	Suggests anemia, potentially due to chronic illness or nutritional deficiency common in neurodegenerative disorders.
Packed Cell Volume (PCV)	Decreased	Consistent with anemia; may further contribute to fatigue or reduced physical endurance.
Differential Leukocyte Count (DLC)	Elevated	May indicate subclinical infection or immune system dysregulation, which are common in AT due to immunodeficiency.
Serum Urea	Normal	Indicates preserved renal function.
Serum Creatinine	Normal	Suggests no evidence of renal impairment.
Serum Sodium	Normal	Reflects stable electrolyte balance; no evidence of dehydration or renal-tubular dysfunction.
Serum Potassium	Normal	Indicates normal neuromuscular function; hypokalemia not present.
Serum Chloride	Normal	Supports overall electrolyte stability and metabolic balance.

III. DISCUSSION

AT is a complex disorder with neurological, dermatological, and immunological involvement. Although this child's MRI with Cerebellar atrophyand AFP were normal, the presence of cerebellar ataxia, telangiectatic lesions, and delayed milestones strongly pointed toward a diagnosis of AT. Diagnostic confirmation requires ATM protein activity assays or genetic testing, which were pending or unavailable. Nutritional status and immune system monitoring are essential in such patients to prevent complications.

Differential diagnosis includes:

- Cerebellar hypoplasia
- Congenital disorders of glycosylation
- Spinocerebellar ataxias

Supportive care remains the mainstay of treatment. A multidisciplinary team including neurologists, immunologists, physiotherapists, and genetic counselors is critical in managing this condition.

IV. CONCLUSION

This case highlights the diagnostic and management challenges associated with Ataxia Telangiectasia, a rare multisystem neurodegenerative disorder. The early presentation of gait ataxia, slurred speech, global developmental delay, and classical telangiectatic lesions strongly suggested AT, even in the absence of elevated AFP levels. Early recognition of these hallmark features is crucial, as timely diagnosis enables proactive multidisciplinary interventions aimed at improving quality of life and preventing severe complications such as recurrent infections and malignancies.

While there is no definitive cure, comprehensive management involving neurological rehabilitation, immunological monitoring, nutritional support, and genetic counseling plays a pivotal role in optimizing outcomes. This case underscores the need for heightened clinical awareness, especially in resource-limited settings, and emphasizes the importance of genetic confirmation for accurate diagnosis and family counseling. Early intervention and long-term follow-up are essential to mitigate disease progression and support both the patient and caregivers in navigating this lifelong condition.

REFERENCES

[1]. Rothblum-Oviatt C, Wright J, Lefton-Greif MA, et al. Ataxia telangiectasia: a review. Orphanet J Rare Dis. 2016;11(1):159.

[2]. Shiloh Y, Ziv Y. The ATM protein kinase: regulating the cellular response to genotoxic stress, and more. Nat Rev Mol Cell Biol. 2013;14(4):197–210.
[3]. Sedgwick RP, Boder E. Ataxia-telangiectasia. In: Vinken PJ, Bruyn GW, editors. Handbook of Clinical Neurology. 1975;21:347–423.
[4]. Nowak-Wegrzyn A, Crawford TO, Winkelstein JA, et al. Immunodeficiency and infections in ataxia-telangiectasia. J Pediatr. 2004;144(4):505–511.
[5]. Lavin MF, Kozlov S. ATM activation and DNA damage response. Cell Cycle. 2007;6(8):931–942.
[6]. Verhagen MM, Abdo WF, Willemsen MA, et al. Clinical spectrum of ataxia-telangiectasia in adulthood. Neurology. 2009;73(6):430–437.
[7]. McGrath-Morrow SA, Kollman TR, Keller M, et al. Evaluation and management of immune dysfunction in ataxia-telangiectasia. Pediatr Pulmonol. 2012;47(6):591–602.
[8]. Chessa L, Luciani M, Dolcetti R, et al. Ataxia-telangiectasia: diagnosis and treatment. Expert Rev Neurother. 2006;6(4):525–531.
[9]. Taylor AMR, Byrd PJ. Molecular pathology of ataxia telangiectasia. J Clin Pathol. 2005;58(10):1009–1015.