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ATAXIA TELANGIECTASIA IN SIBLINGS – A RARE CASE REPORT

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ABSTRACT
Ataxia telangiectasia (synonym: Louis-Bar syndrome) is a rare neurodegenerative disorder inherited in an autosomal recessive pattern. It causes progressive degeneration of multiple systems. Commonly presents in infancy or childhood. We report a case of ataxia telangiectasia in siblings with one child, associated with vitiligo.

INTRODUCTION

Ataxia telangiectasia is a hereditary genetic disorder. AT was first described in 1926 by Syllaba and Henner [1]. The first case was described in 1941 by Madame Louis Bar in a Belgian child. The incidence is 1 in 40,000 to 100,000 live births [2] with a carrier rate of 1%. Carriers with dominant negative missense mutations, have an increased risk of breast and hematologic malignancies. Life expectancy in carriers is decreased by 8 years approximately [3], deaths are mainly due to both ischemic heart disease and malignancies in such cases. Both females and males are equally affected. No regional or racial distribution.

CASE REPORT

12 year old boy born to non-consanguineous parents was brought to our OPD by his mother with asymptomatic light colored lesions over the lips and dorsum of left foot. History from the mother revealed that the child was delivered by full term, normal delivery with 2.6kg birth weight. Antenatal, natal and postnatal period was uneventful. Motor, language and social milestones were delayed. There was no history of hearing or visual disturbances or seizures. By 3 years mother noticed redness of eyes which also involved the ears. By 7 years the child had repeated falls and difficulty in walking and within 6 months he was bound to wheelchair and also had history of slowing of speech, head bobbing and difficulty in swallowing. No history of recurrent infections. His elder brother aged 14 years also has similar complaints of delayed milestones, redness of eyes and difficulty in walking.

On general examination child is poorly built and nourished. Genu valgum and pes cavus are the positive findings.

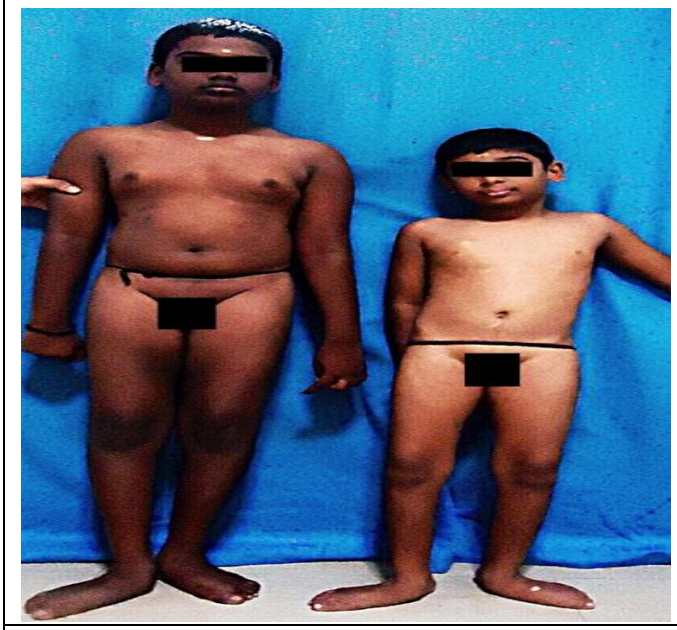
On dermatological examination, well circumscribed depigmented patch is seen over the lips, trunk and dorsum of left foot. Hypertrichosis of the trunk, both upper and lower limb present. Nails, teeth, palms and soles were normal (Figures 1, 4 & 5).

Eyes & Ears - telangiectasia in the bilateral cornea, conjunctiva and ears (Figures 2 & 3).

Cardiovascular system, respiratory system and per abdomen examination were normal. On CNS examination, titubation and dysarthria were present.

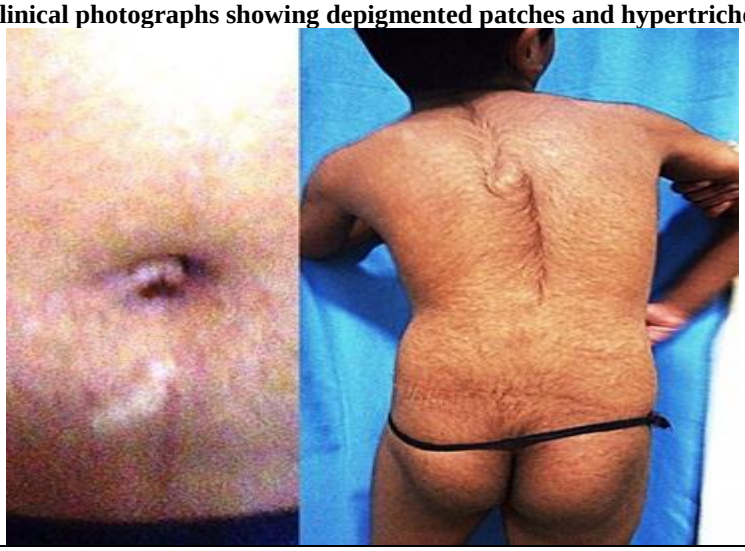
All routine investigations were within normal limits. Serum CEA (carcinoembryonic antigen) and AFP (alphafetoprotein) levels were elevated and IgA levels were decreased in both the siblings. MRI brain of both siblings revealed diffuse cerebellar atrophy and bilateral maxillary, sphenoid and ethmoidal sinusitis changes.

Figure 1. Clinical photograph of both the siblings.



Figures 2 & 3. Clinical photographs showing telangiectasia in the conjunctiva and external ear.

Figures 4 & 5. Clinical photographs showing depigmented patches and hypertrichosis in the trunk.



DISCUSSION

Ataxia telangiectasia (AT) is characterized by oculocutaneous telangiectasias, progressive cerebellar ataxia beginning in infancy, both cellular and humoral immunodeficiency with a tendency to develop sinopulmonary infections and decreased IgA levels, ionizing radiation hypersensitivity and increased risk of lymphoid malignancy. AT is caused by homozygous mutations in ATM gene (ataxia telangiectasia mutated) located in chromosome 11q22.3, which encodes a phosphatidylinositol 3-kinase like serine/threonine protein kinase which plays a important role in cell cycle responses to DNA damage and activating apoptosis. The MRN [4] (MRE11-RAD50-NBS1) complex senses DNA breakage and activate ATM. Autophosphorylated ATM monomers activates a variety of targets, including p53, FANCD2, BRCA1, MRE11 and NBS1 by phosphorylation. AT presents with telangiectasia which usually develop between the age of 3 and 5 years but it can present as early as 2 years of age in the eyes but it can also seen in ears, eyelids, malar prominence, limbs, trunk, popliteal and antecubital fossae. It can also be associated with petechiae. Cerebellar Ataxia is progressive and becomes apparent once the child

starts to walk and is characterized by swaying of the head and trunk, dysarthric speech, oculomotor abnormalities, choreoathetosis and myoclonic jerks (which usually become prominent during childhood). Patients are typically confined to a wheelchair in teenage, inspite of good muscle strength. AT also presents with mottled hyper and hypopigmentation [5] and café au lait macules. It is associated with seborrheic dermatitis, vitiligo [6], alopecia areata, hypertrichosis of arms and legs, atopic dermatitis, multiple warts, acanthosis nigricans, keratosis pilaris, nummular eczema, recurrent impetigo and cutaneous granulomas. Recurrent sinus and pulmonary infections are common. There is a strong association with malignancy (leukemia, lymphoma and breast cancer).

Diagnosis is usually made by clinical presentation. Laboratory investigations are serum CEA & AFP levels are usually increased, MRI done after 2 years of age shows cerebellar atropy, radiosensitivity testing done with the colony survival assay [7], analysis of radioresistant DNA synthesis, assessment of ATM kinase activity, immunoblotting for the ATM protein, karyotyping and molecular genetic testing for prenatal diagnosis. Management is by multidisciplinary approach – genetic counseling, physical therapy for neurological manifestations, antibiotics for infections, physiotherapy and corticosteroids for interstitial lung disease, sun exposure should be avoided and usage of sun screens and aggressive screening for malignancy. Death usually occurs by late childhood or early adolescence due to pulmonary infections and malignancy.

CONCLUSION

Patients with AT are at increased risk of recurrent sinopulmonary infections and malignancy, hence treating the infections and screening for malignancy can improve life expectancy. This case is reported because of the rare occurrence of AT in siblings and its association with vitiligo.

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CONFLICT OF INTEREST:

The authors declare that they have no conflict of interest.

STATEMENT OF HUMAN AND ANIMAL RIGHTS

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

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