Two Sudanese siblings with ataxia telangiectasia: A case report

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Online 7th ASID Congress October 1st -2nd 2021

Background

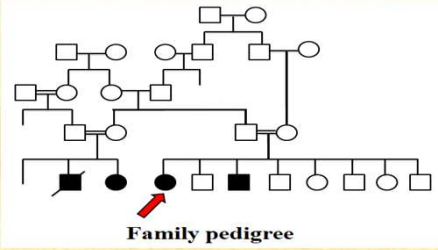
Ataxia Telangiectasia (AT) is an autosomal recessive disorder caused by ATM gene mutations & presents with cerebellar ataxia, telangiectasia & immunodeficiency.

Purpose:

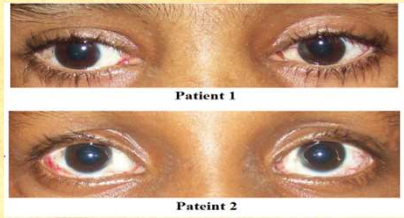
To highlight the importance of early specific management, multi-disciplinary team (MDT) approach and transition care in two siblings with AT.

Methods:

Case presentation of 2 siblings with unsteady gait and frequent falls since an early age. They were diagnosed with AT aged 11 and 7 years, without subsequent regular follow up. At 19 years, the female (*patient 1*) presented to adult respiratory service with acute infective exacerbation of bronchiectasis, preceded by 5 hospitalizations in the span of 8 months. Her brother (*patient 2*) had recurrent otitis media and febrile episodes requiring IV antibiotics. Both had ocular telangiectasia and ataxia. Parents were second degree cousins. The mother identified two paternal cousins with a similar presentation; a male who died (undiagnosed) aged 14 years, and a female who was later on diagnosed with AT and died aged 9 years.



Family pedigree



▪Investigations:

Patient 1: leukocytosis with neutrophilia, high inflammatory markers, sputum C/S: multi-drug resistant *Klebsiella spp.,* IgG 25.5 g/L, IgA 1.24 g/L, HRCT chest: Extensive bronchiectasis, MRI brain: mild generalized cerebellar atrophy.

Patient 2: IgG 25.2 g/L, IgA < 0.06 g/L, MRI brain: normal.

Genetic testing was not feasible.

▪Treatment:

Patient 1: antibiotics, intravenous immunoglobulin (IVIG), supportive care and long term oxygen therapy.

Patient 2: prophylactic antibiotics & IVIG.

Results:

Patient 1 and *patient 2* succumbed 3 months and 3 years respectively.

Conclusion:

AT can be easily diagnosed based on the clinical phenotype, however, both patients were diagnosed rather late. Early diagnosis, MDT approach and smooth transition care might have improved the outcome. In this family with many members with AT, genetic testing could have confirmed the diagnosis and provided adequate counselling for the extended family.

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