

## ATAXIC DISORDERS

### CLINICAL AND LABORATORY FEATURES OF ATAXIA TELANGIECTASIA

Clinical and immunological features of ataxia-telangiectasia (AT) in 104 Iranian patients were evaluated at the Children's Medical Center, and other centers in Tehran, Iran. Records were obtained from the Iranian Primary Immunodeficiency Registry. Patients at time of study were aged 1.6-23.5 years; 54 were male (median age, 9 yrs 4 mos) and 50 female (median age, 8 yrs 6 mos). Median age at symptom onset was 1 year for males and 1 yr 4 mos for females. The median delay to time of diagnosis was 58 months. Median follow-up was 2 years. Four patients had died and 48 were lost to follow-up. Average age at death was 12 yrs 5 mos. Clinical characteristics included progressive ataxia in all patients, telangiectasia of conjunctiva (83.8%) and ear (70.2%), eye movement disorder in 80.6% (apraxia of horizontal and vertical saccadic eye movements), choreoathetosis in 87.1%, speech dysarthria in almost all, epilepsy (8.7%), mental retardation (9.6%), and growth retardation (37.4%). Acute and recurrent sinopulmonary infections affected 78 (75%) of 104; 54% had pneumonia. Recurrent diarrhea was seen in 21%. *Pneumococcus* and *E. coli* were the most common organisms. Parental consanguinity was reported in 79 families. Eighteen sibling pairs had AT. Eleven patients had a positive family history of recurrent infections, and 35 had a positive family history of immunodeficiency and AT. Three patients developed leukemia and lymphoma, and 17 had a family history of malignancy.

Low levels of serum immunoglobulin included IgA (<10 mg/dL in 75.4%), IgM <25 mg/dL in 67.7%, and IgG2. Immunoglobulin levels were inversely correlated with incidence of respiratory infections. A relative lack of CD4+ T-cells was found in 75.8% (severe lack in 44.4%). Average serum a-fetoprotein level was 149+/-137ng/dL. (Moin M, Aghamohammadi A, Kouhi A et al. Ataxia-telangiectasia in Iran: clinical and laboratory features of 104 patients. *Pediatr Neurol* July 2007;37:21-28). (Respond: Dr Aghamohammadi, Children's Medical Center, No 62, Dr Gharib St, Keshavarz Blvd, PO Box 14185-863, Tehran 14194, Iran).

COMMENT. The unusually high incidence of ataxia-telangiectasia (AT) in Iran is attributed to frequency of consanguineous marriage. AT is an autosomal recessive, multisystem disorder of childhood manifested by progressive cerebellar ataxia, ocular and cutaneous telangiectasia, immunodeficiency with increased susceptibility to sinopulmonary infection, x-ray hypersensitivity, and predisposition to malignancy. Diagnosis is often delayed until after age 10 years, and death follows by age 20 years, from pneumonia or malignancy. Following initial reports of the disease by Louis Bar (1941) and previous French authors, Syllaba and Henner (1926), the condition was described in detail and given the name AT by Boder E and Sedgwick RP in 1958 (*Pediatrics* 21:526). An IgG2 deficiency in AT was reported by Oxelius VA et al (1982).