Clinical Images

Spontaneous pneumothorax in ataxia telangectasia



Fig. Clinical photograph showing (A) ocular and (B) nail telangiectasia (arrows). Baseline X-ray chest posteroanterior (P/A) view (C) displayed right epidiaphragmatic and apical hyperlucencies (arrows) suggestive of pneumothorax with subsequent resolution of pneumothorax following insertion of intercostal tube (D) (arrow).

A 20 yr old male presented to Neurology Outpatient department, Postgraduate Institute of Medical Education and Research (PGIMER), in November 2012 with progressive cerebellar ataxia since the age of eight. The patient complained about a sudden onset shortness of breath for the past five days. Clinical examination revealed conjunctival and nail telangectasia (Figs. A and B). Chest roentgenogram showed right sided pneumothorax (Fig. C) that was managed with high flow oxygen and intercoastal drain insertion (Fig. D)

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Fig. Computerized tomograph of the chest (E) revealed presence of pneumothorax (arrow) with no parenchymal changes. Cranial MRI (F) depicted cerebellar atrophy (white arrow) without cerebral cortical parenchymal changes.

following which the patient had symptomatic relief. CT of chest (Fig. E) and cranial MRI (Fig. F) revealed findings as described. A diagnosis of ataxia telangectesia was made based on clinical and laboratory findings (elevated serum AFP and reduced serum IgA and IgE levels). The patient had no complaints of dyspnoea, and pulmonary function tests done six months later were normal.

Sudheer Chakravarthi^{*} & Manoj Kumar Goyal Department of Neurology, Postgraduate Institute of Medical Education & Research Sector-12, Chandigarh 160 012, India **For correspondence:* sudheerpgi@gmail.com