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CASE REPORT

Pulmonary tuberculosis in a case of Rubinstein–Taybi syndrome

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Summary
Rubinstein–Taybi syndrome is a rare genetic disease characterized by mental deficiency, broad thumbs and toes, short stature, and characteristic facial features. The syndrome has been linked to microdeletion at 16p13.3 encoding CREB-binding protein gene (CREBBP). Most reported cases of Rubinstein–Taybi syndrome are sporadic, with no firm evidence of Mendelian inheritance. We report a case of a 26-year-old male patient of Rubinstein–Taybi syndrome with pulmonary tuberculosis. The occurrence of respiratory infections in patients with this syndrome has been mentioned in the past. Systemic problems involving the respiratory system, feeding and the cardiovascular system have been noted in some individuals with this rare syndrome.

Introduction

Rubinstein–Taybi syndrome (RSTS) is a genetic multisystem disorder characterized by facial dysmorphism, broad thumbs and halluces, growth retardation and mental deficiency. Though first described by Michael in 1957 in the French literature, it was named after Jack Herbert Rubinstein and Hooshang Taybi who identified the constellations of a recognizable syndrome in 1963. The syndrome is estimated to occur in about 1 in 1 000 000–300 000 births. Rubinstein–Taybi syndrome is a rare multiple congenital anomaly syndrome with only about 600 cases reported worldwide. The current case reports a case of Rubinstein–Taybi syndrome. Diagnosis of RSTS was first established in the patient at the age of 26 years.

Clinical presentation

A 26-year-old male patient, with multiple malformations, short stature and mental retardation presented to the outpatient department with complaints of fever, cough and dyspnea for the last 2 months.

The prenatal and natal history of the child was uneventful. The mother revealed that the patient had delayed developmental milestones, feeding and speech difficulty

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since childhood. The patient had history of gastro-esophageal-reflux disease (GERD) with severe constipation. History of keloid formation with only minimal trauma to the skin was present. The patient did not go to school and appeared mentally challenged. He had been operated for club feet at the age of 2 years. The patient was the second, of 3 children of parents, with no history of consanguinity. There was no history of similar complaints in the siblings. The patient appeared to have a short stature for his age.

His craniofacial features included a prominent forehead, downward slanting of the palpebral fissure, epicanthal folds, beaked nose, grimacing smile and micrognathia (Figure 1). Head circumference was 48 cm which was under the 50th percentile for his age and sex. Examination of the ears and ophthalmologic system did not reveal any abnormality. Dental examination revealed crowding of teeth, malocclusion and talon cusps on the upper incisors of the secondary dentition. The patient had a stiff gait with scoliosis (Cobb's angle of 68°). Hypotonia and pectus excavatum was present. Examination of the limbs revealed large first toes with broad terminal phalanges, abnormal shape and angulation of the first metatarsal and fifth finger clinodactyly (Figures 2 and 3).

Loss of terminal phalanges of the hands secondary to minor trauma was present. Examination of the genitourinary system was remarkable for presence of bilateral undescended testes. The patient had speech difficulty, short attention span with an intelligent quotient (IQ) of 54. Examination of the abdominal and cardiovascular system was unremarkable. Capillary hemangiomas at multiple sites, keloid formation at the sites of trauma and hyperpigmented skin at peripheries of upper limbs and lower limbs was present. Height of the patient was 147 cm. Routine laboratory investigations of the patient were all within normal limits. The patient tested negative for human immunodeficiency virus (HIV). The CD4 and CD8 levels were normal. Radiographs of the patient confirmed the above findings. Bone age in this patient was normal. Karyotyping revealed no chromosomal abnormality.

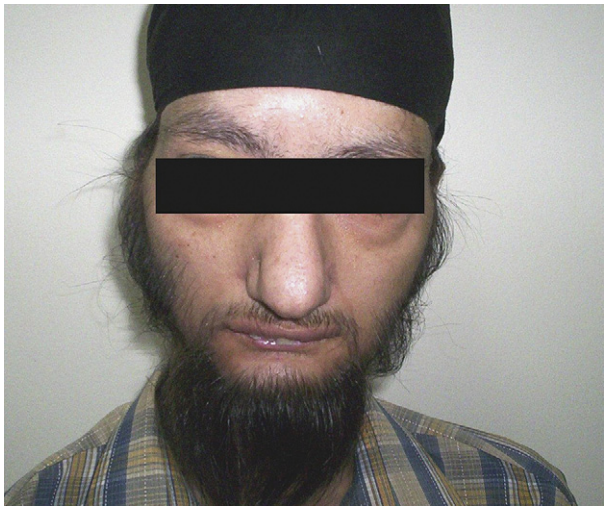

Figure 1 Photograph of the patient showing prominent forehead, downward slanting of the palpebral fissure, epicanthal folds, beaked nose, grimacing smile and micrognathia.

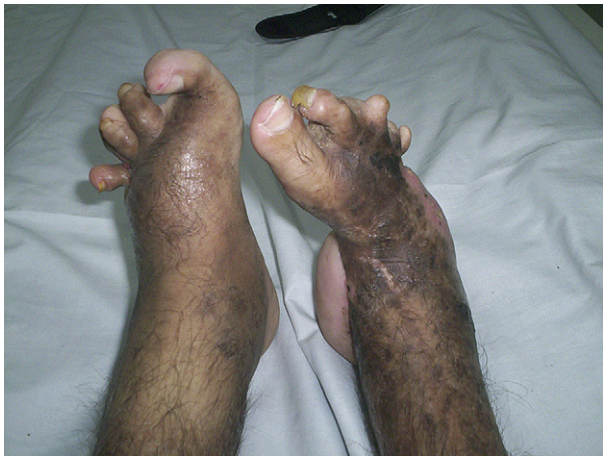

Figure 2 Photograph of both feet showing broad first toes with abnormal proximal phalanges consistent with Rubinstein–Taybi Syndrome.

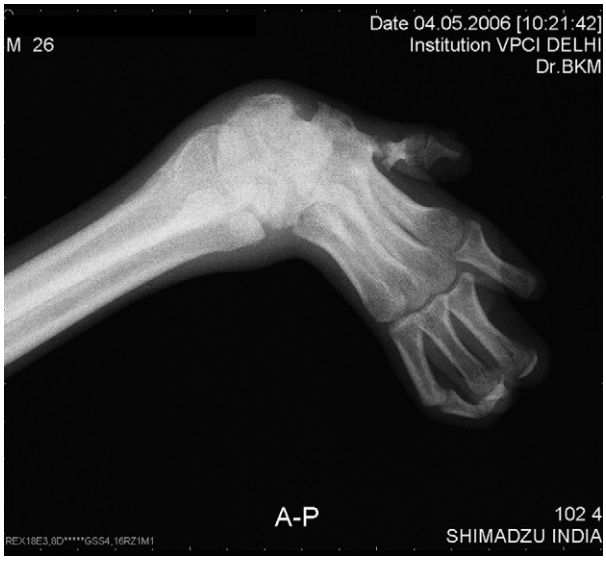

Figure 3 Radiograph of the hand (AP view) showing clinodactyly of the fifth finger with hypoplastic first metacarpal.

Examination of the respiratory system was unremarkable with a normal chest radiograph. Sputum for fungus and acid fast bacilli was negative. Computed tomography scan of the chest revealed areas of patchy consolidation in the left lower lobe with fibro infiltrative opacities in both lungs (Figure 4). Bronchoscopy was unremarkable. Bronchial aspirate for acid fast bacilli detection by culture on L-J medium was positive. BACTEC was positive for mycobacterium tuberculosis and drug susceptibility testing showed the bacilli to be sensitive to all the first line anti-tubeculosis drugs. MTb PCR was positive for the insertion sequence IS6110. The patient was started on anti-tuberculosis treatment. Patient responded to the treatment and bronchial aspirate for acid fast bacilli was negative at the end of treatment.Currently, the patient is being followed up for orthodontic management of feeding difficulty and is on medication for GERD and constipation. The patient is also undergoing speech and cognitive therapy.