The Turkish Journal of Pediatrics
1972 / Vol. 14 / No. 3 / pp. 118 - 124

Rubinstein-Taybi Syndrome

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This syndrome was first described by Rubinstein-Taybi in 1963. Several authors have published more than 120 cases since then. This syndrome is characterized by abnormal facial features, mental and motor retardation and broad thumbs and toes in most cases.[1-4] The case which is described below is the first one to be diagnosed and published in Turkey.

S. A., a 12 year old female, was admitted to Hacettepe Children's Hospital in March 1972, with complaints mainly of mental retardation and inability to talk. She is the third child of healthy parents, who were distantly related. She was born at home, after an uneventful pregnancy and normal delivery. There was no exposure to radiation or drugs during the pregnancy. Her birth weight is unknown, but she was regarded as an average baby. There was no history of cyanosis, asphyxia and jaundice. She had a weak cry at birth and her sucking was also reported weak. Her motor and mental development was slow. She walked at 7 years of age, and started to talk with some peculiar words and completed her toilet training when she was at 8 years of age. She is still unable to express her demands in regular speech and has a peculiar gait. Her past history is unremarkable. She has one younger and two elder brothers. The oldest brother is 16 years old and has some speech problems. He was reported to have repeated failures at school.

Physical examination showed the girl of 12 to be of short stature, with smiling face. Her weight was 21 kg, height 121 cm, head circumference 47 cm, chest circumference 60 cm. Her nose was beaked and long and eyes were small, with an antimongoloid slant. Her teeth were small and she had a high arched palate. Her ears were in normal position. She had pectus excavatum. Cardiorespiratory, gastrointestinal systems and external genitalia were normal. She had no breast development.

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Her hands and feet had peculiar manifestations of this syndrome. Both hands had short fingers, with larger than normal size thumbs, which were both deviated radially. Her toes were also considered larger than average. No other bone defects or abnormalities were noticed. Her neurological examination was within normal limits, except microcephaly and apparent mental retardation. She walked with a wide based gait. (Figures 1 a, b, c, d).

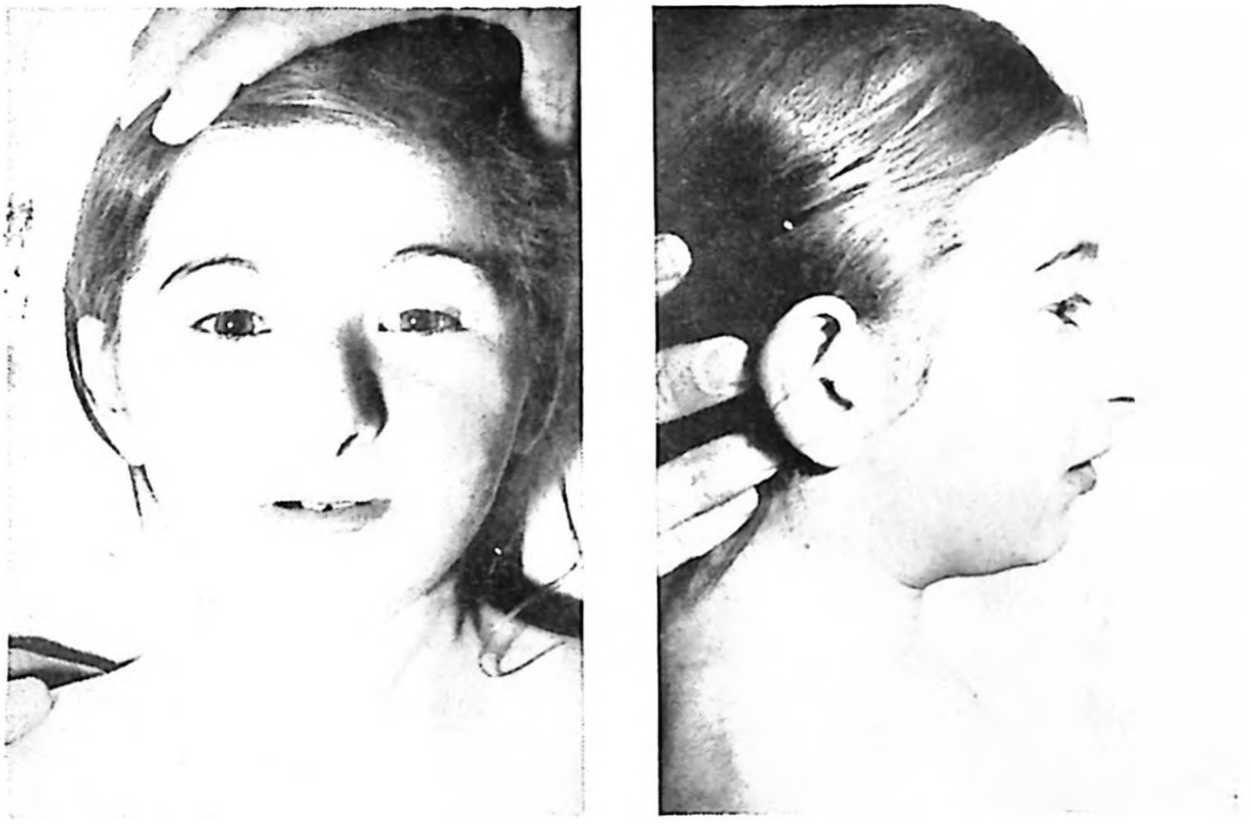

Figure 1 a, b
Front and lateral and profile of face

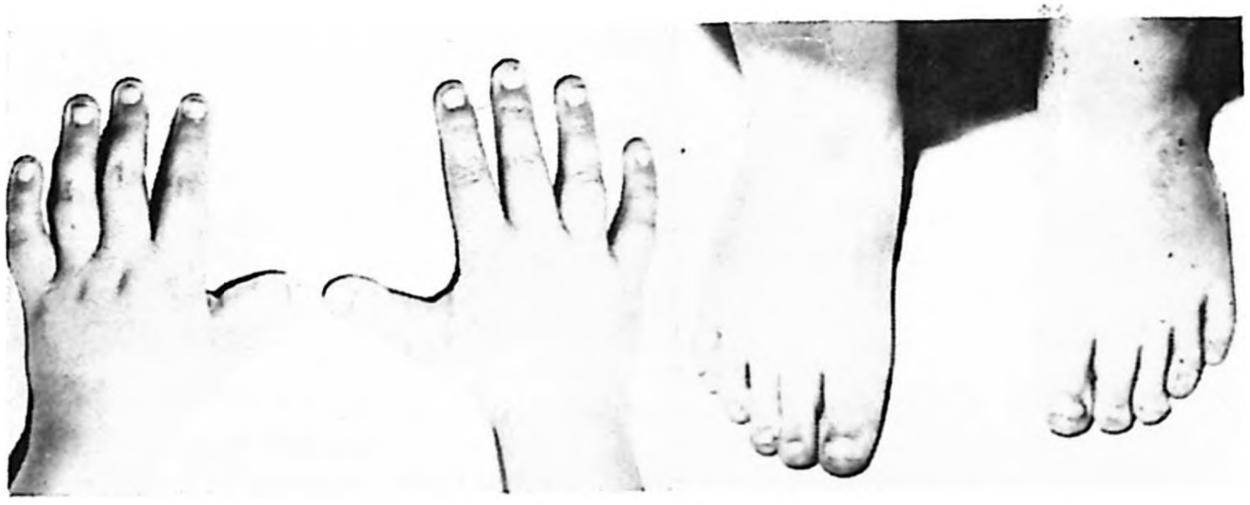

Figure 1 c, d
Dorsal view of hands and feet. Note the large thumbs and toes.

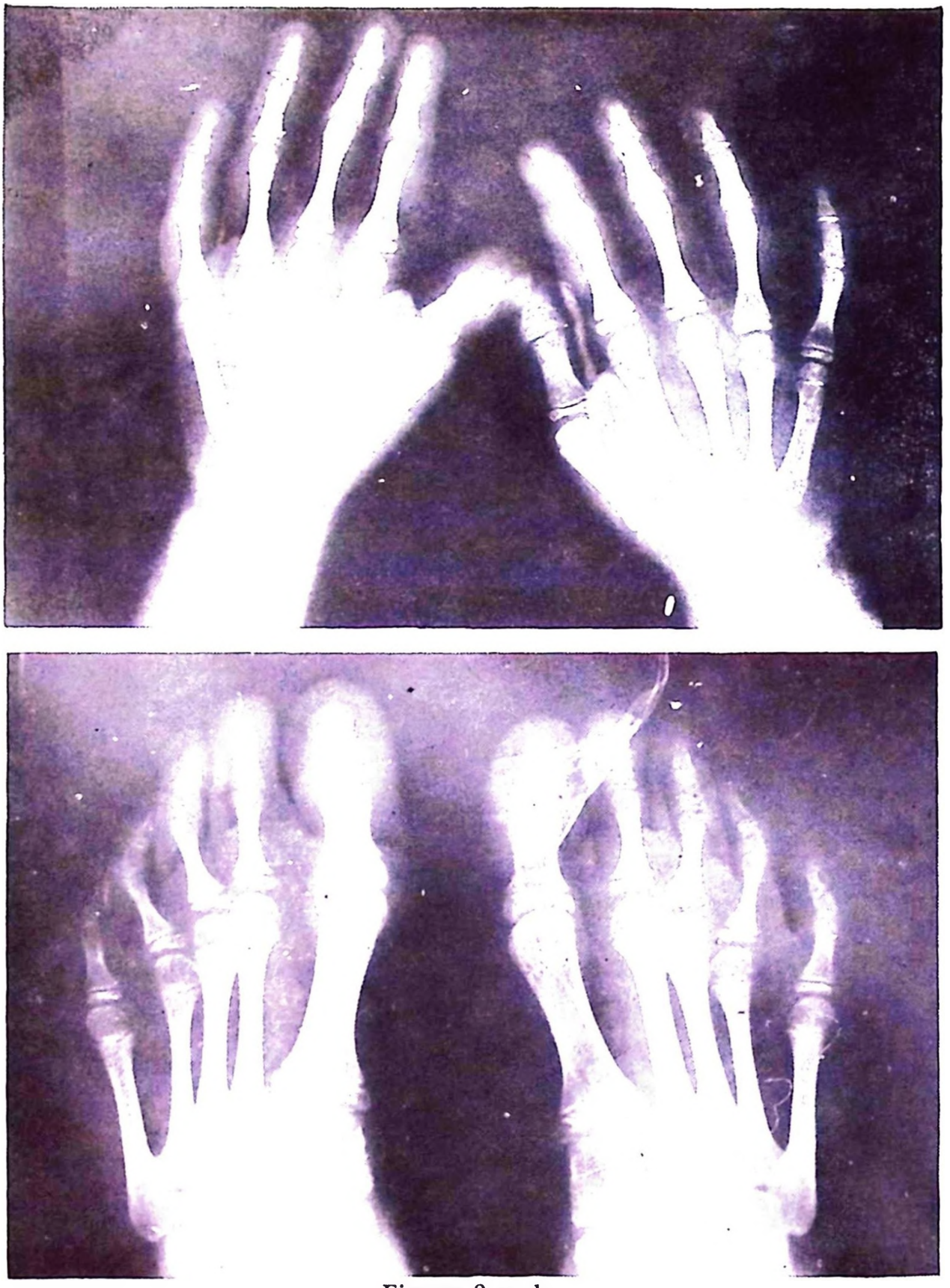

Figure 2 a, b
X-rays of hands, feet and basis cranii.

Laboratory examination revealed normal hemogram and urinanalysis. Her chest, skull, vertebrae and long bone films were normal. Foramen magnum was reported within normal limits. Her bone age was parallel

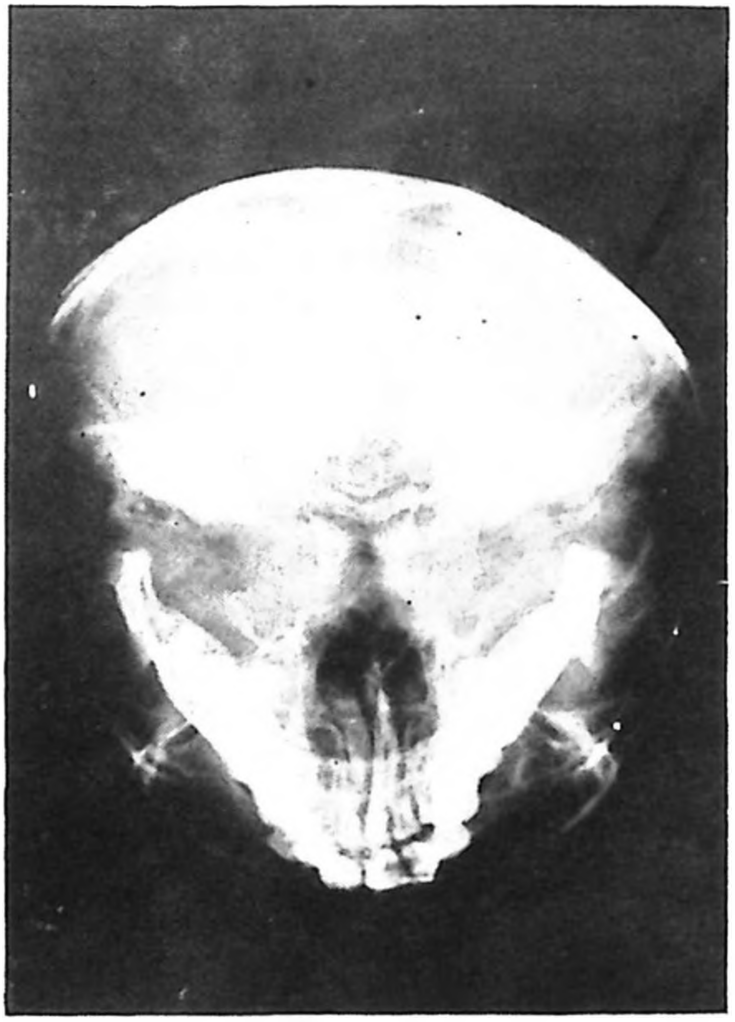

Figure 2 c

to that of an 8 year old child. (Figure 2 a, b and c). Chromosome analysis showed a 46+XX karyotype. Dermatoglyphic analysis was significantly different than normal controls. EEG showed a diffuse and mild disturbance. Her IQ was found to be around 30.

Discussion

The principal findings of this syndrome are listed in Table I.

TABLE I

CARDINAL SIGNS OF RUBINSTEIN-TAYBI SYNDROME

Motor and mental retardation	Grimacing smile
Small and short stature	Small chin
Broad toes and thumbs	High arched palate
Microcephaly	Long and beaked nose
Antimongoloid slants	Large forame magnum
Naevus flammeus	Associated congenital anomalies

Several authors have reported similar findings as far as the above manifestations are concerned.[2 4-6] The main features, however, were broad thumbs and toes. Rubinstein and Taybi were the first to discover 6 cases with broad thumbs and toes. Broad thumbs were found to be more consistent. These authors also have published the X-ray findings of 13 cases showing short and broad ends of distal phalanges of the toes and thumbs. Some of these showed segmentation and fragmentation, while the distal phalanges seemed clubbed. There were no reports of malar and mandibular defects.[2 6 7]

All of the cases reported had long, thin and beaked noses, grimacing smiles, antimongoloid slants and high palates. Mental and motor retardation have been common. Naevus flammeus and hemangiomas were frequent, undescended testes were also observed in some cases.[1 2 4-6 8 9]

Among ocular findings, Roy and his associates have reported antimongoloid slanting in 89 % of the cases, strabismus in 72 %, epicanthus in 58 % accommodation disorders in 55 % and long eyelashes in 28 %.[9]

Among the published studies there were very few pathological reports. Coffin has published the postmortem findings of one case: congenital fibroelastosis, abscence of the posterior part of corpus callosum, architectural changes of cerebral cortex, such as irregular lining of cortical cell levels, accumulation of neurons with scanty cytoplasm.[10]

Dermatoglyphic findings have been a point of curiosity. Giroux and Miller have reported extensive patterns (loops and whorls) in thenar /Interdigital I and hypothenar areas. They have also noted that the axial triradius was distally placed, and there were more than a single triradii and increased frequency of arches on the digital patterns (17.4 % compared to 5.3 % among controls).[11] Davison noticed that these cases lack -c- main lines.[8] Johnson has reported a case with bilateral simian creases[1] (Figure 3).

No chromosomal abnormality has been reported up to the present. Cases with chromosomal analysis had normal karyotypes, such as 46,XY or 46,XX. Davison and her associates have reported some changes among group C chromosomes, such as delayed labeling one of the chromosomes by autoradiography.[8]

The mode of inheritance is most likely to be autosomal recessive.[12] There is no sex predilection. Aminoaciduria and maternal rubella were thought to be etiological causes.[4 6] More than one affected person in a family is reported quite often.[1 13] The rate of frequency is around 1/500 among the institutionalized cases.

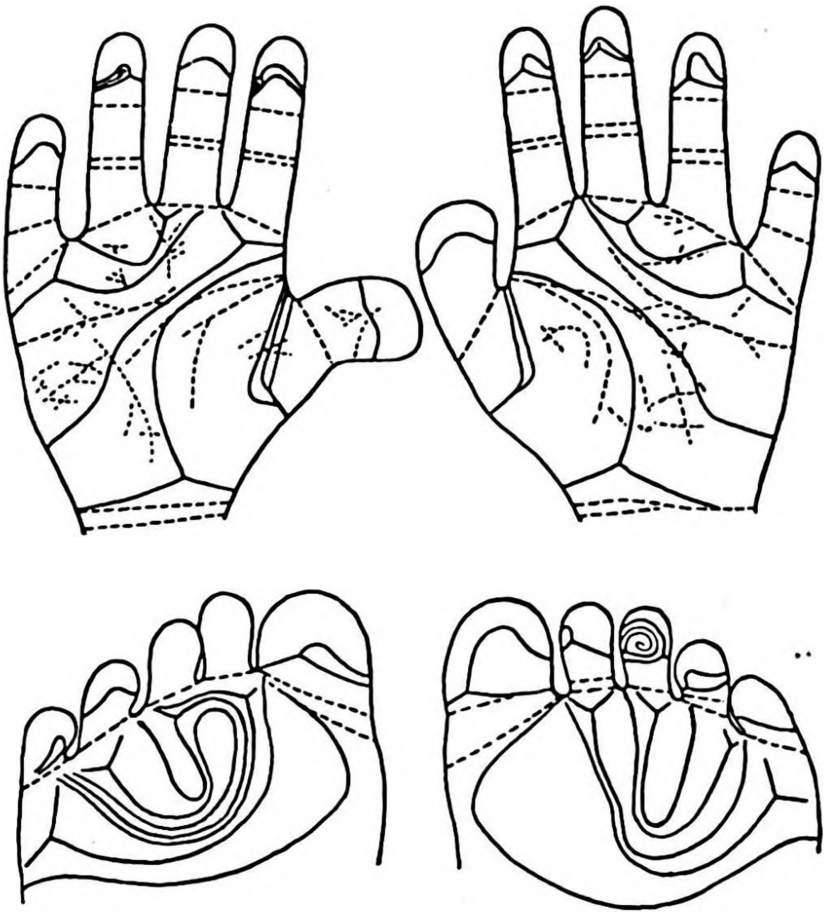

Figure 3
Dermatoglyphic patterns of hands and feet.

Pfeiffer has described 8 persons in a family, within 3 generations, with acrocephaly, partial syndactily and broad thumbs. But these cases differed from those with the Rubinstein-Taybi syndrome in their deafness, limited joint movements, visceral anomalies and normal intelligence[14-15] The Hallermann-Streif-syndrome and the Treacher-Collins syndrome can also be differentiated by their typical features.[17-18]

Summary

A case with mental and motor retardation, broad thumbs and toes, antimongoloid slanting, small posture and grimacing smile is presented. This case was regarded as having the typical findings of Rubinstein-Taybi syndrome, and also is the first one to be reported in Turkey.

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