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**Broad Thumbs and Toes with Mental Retardation;
Syndrome of Rubinstein and Taybi.
(Case Report)**

by

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Introduction

In 1963 Rubinstein described how he was struck by the experience of finding two unrelated mentally retarded children who seemed to resemble each other so closely, that he wondered if they might not have the same "syndrome". After studying and reporting this, he got additional cases with the same clinical findings, and also saw other cases himself; he then suggested the possibility that the cases represented a single clinical entity. If this single entity was confirmed, the identification might lead to further investigations for a possible common pathogenesis.

The symptoms in the first cases were described as: Broad Thumbs and Toes and Facial Abnormalities, a possible Mental Retardation Syndrome (Rubinstein and Taybi, 1963). Since this publication more cases have been reported. (Coffin, 1964; Taybi and Rubinstein, 1965).

Seeing the unusual syndrome in an Indonesian baby we considered publication worthwhile.

The Rubinstein and Taybi syndrome consists of a large number of congenital anomalies. Some of these are common to all cases, many are seen in some cases only.

The abnormalities common to all cases are: 1. mental and motoric retardation; 2. broad thumbs and big toes; 3. highly arched palate; 4. eye abnormalities of different types.

The most prominent, consistent, and readily recognizable abnormality is that of the broad thumbs and big toes. These are variously described as: short and stubby, spatulate, clubbed, flat and rather wide, large and broad. The nails appear correspondingly flattened in some of the cases. The other fingers are variably involved, but much less than the thumbs and big toes. By roentgenographic examination the distal phalanges of

the thumbs and big toes are prominent and appear short and broad. This anomaly is often recognizable at birth and becomes more marked with growth.

The abnormalities of the eyes are described as exotropia, refractive error, cataract, strabismus, enophthalmus, ptosis, anti-mongoloid slant, epicanthus, highly arched eyebrow, and hypertelorism. The second group of abnormalities, present in some and absent in others are: 1. microcrania 2. delayed closure of large anterior fontanel, 3. beaked nose, 4. large foramen magnum, 5. abnormal segmentation with partial duplication of the phalanges of the thumbs and big toes, 6. broad terminal phalanges of other fingers, 7. short stature, 8. retarded skeletal maturation, 9. abnormal electroencephalograms, 10. frontal naevus flammeus, 11. neonatal feeding problems, 12. recurrent respiratory problems in infancy and early childhood, 13. allergies, 14. incompletely descended testis; and some more features.

The faces of the children are described as "odd", due to the presence of a combination of features: slanted palpebral fissures, the highly arched palate, and the prominent nose (beaked nose). The facial appearance sometimes slightly resembles that of mandibulofacial dysostosis (Franceschetti-Klein or Treacher-Collins syndrome), but mental retardation and broad thumbs and toes are not found in mandibulofacial dysostosis.

Also other features of mandibulofacial dysostosis i.e. molar and mandibular defects, deafness, lid colobomas, eyelash defects and macrostomia, are lacking in the cases described as Rubinstein-Taybi syndrome (Nelson, 1969).

Concerning investigations for a pathogenesis no consistent chemical or cytological abnormality has yet been detected. Amino-acid and chromosome findings in the children reported were normal, so the syndrome is not an inborn error of metabolism nor a chromosomal aberration.

Of hereditary interest was the finding of broad thumbs in the father, paternal uncle and aunts, and a paternal great-grand-uncle of one of the cases (Rubinstein and Taybi 1963).

The prenatal course of the cases reported could not be considered entirely normal, but did not show any consistent pattern, as described by Rubinstein and Taybi (1963): weak fetal movements, tensed abdomen, polyhydramnion, U.R.I. a.o. infections during pregnancy.

Case report

An Indonesian baby-girl, N.W., was born on June 26, 1971 as the first child in a family in which there was no history of parental consanguinity. The mother, a 35-year-old Sundanese, was asthmatic and had a miscarriage in February 1970. The father was a 30-year-old Javanese, who complained of "nervousness". His younger brother could be called

"queer", but there was no history of mental retardation in either the father's nor the mother's family. The baby was born fullterm after a normal pregnancy; delivery was spontaneous, the baby cried immediately.

Birth weight was 2200 gm; length 47 cm; head circumference 31 cm; and chest circumference 31 cm. She got 2 mg vit. K inj. for 2 days and drank some water. The third day the child had fever of 39.5° C, was cyanotic, restless, had twitching and could not drink. The next day temperature was 40.5° C, the baby was dyspnoeic and cyanotic, had three times convulsion and was transferred to the Bethesda hospital.

On admittance the child had generalized cyanosis and dyspnoea. Body weight was 1980 gm. Physical examination revealed tachypnoe, respiratory retraction of the chest, and prolongation of the expiratory phase of respiration. Moist rales were heard over the entire chest and also a systolic heart murmur. Liver-edge and spleen were just palpable. The child was spastic and twitching was present, it had the hands clenched. Anterior fontanel was large, but not tensed nor bulging. Liquor was normal.

The child had a highly arched palate. Thumbs and big toes were broad, the other terminal phalanges appeared flat and somewhat clubbed, nails were flat. Fingers were short.

The right big toe was doubled (polydactily). The ears had a slight anomaly of folding. White blood count revealed 13200 leucocytes. Urinalysis was normal.

The baby was placed in an incubator, received oxygen, penicilline and phenobarbital, and had to be tube-fed. After some days the temperature was normal again, although several times the baby was still dyspnoeic and had circumoral cyanosis. Sucking was weak. After 3 weeks she was taken home on request of the parents; body weight at that time was 2420 gm.

On February 6, 1972 we saw the baby again when she was admitted for the second time, now because of hyperpyrexia and bronchopneumonia. She was 7 months old and very ill. She had several time convulsions and was still somewhat "spastic". Physical examination revealed a retarded baby with a weight of 5350 gm (weight-age 3 months)*; a height of 60 cm (height-age 3½ months)*; and a head circumference of 37 cm (head age 2 months* microcrania). The occipital region was flat, the anterior fontanel large 3 X 4 cm, and reached as far as her forehead. The bridge of the nose seemed slightly widened. A suggestion of hypertelorism was present, as well as maxillary hypoplasia. The palate was highly arched. The child had not yet any control of the head, and just

* Because of unavailability of centiles of weight and height for Indonesian babies we used the standard mean values of measurements by Moh. Sugiono and Te Bek Siang (1964).

began to roll over; it could smile, but almost always kept her eyes closed and could not fix them because of cataract of the eyes. Feeding was difficult.

After discharge the child was not doing well. She kept sniffing and rattling, and several times she was febrile. She contracted diarrhoea and was readmitted for purulent meningitis. Weight gain was unsatisfactory.

On June 21, 1972 the baby was 12 months old and had a weight of 5900 gm, which is a weight-age of under 4 months. After that we did not see her for a long time. In July 1973 she was operated for congenital cataract on both eyes.

A last check-up was done on October 31, 1973 at the age of 28 months.

The child was doing rather well, her health condition was better. Body weight was 6700 gm (weight-age of 6 months), height 70 cm (height age of 12 months), head circumference 38 cm (head-age of 3 months). Bone age was 6 months (Caffey, 1967).

The anterior fontanel was closed; the hair grew low on her forehead. She had 8 teeth and 4 molars. Feeding was rather easy. She did not sniff nor rattle anymore. But she was still very retarded, mentally as well as motoric. She could not even control her head well, nor sit, although she could roll over. She could smile, but could not fixate; she was able to say 2 short words (pak, mam) and make some noises. She was weak and slightly anemic; hemoglobin content was 9,6 gm%.

Clinical findings are outlined in the table below:

Prenatal	:	mother asthmatic; had a miscarriage the year before.
Mother's age	:	35 years.
Father' age	:	30 years.
Duration of pregnancy	:	full term.
Birth	:	vertex.
Birth weight	:	2200 gm.
Neonatal	:	Respiratory distress, cyanosis, convulsions, hyperpyrexia. Tube fed. Poor sucking. Broad thumbs and big toes; other finger-tips and toe-tips broad (clubbed). Polydactily right big toe (Fig. 1 and 2). Large anterior fontanel. Systolic heart murmur.
First tooth	:	13 months.
Motoric retardation	:	can not sit at 2 years 4 months

Said only "pak, mam"	: 2 years 3 months.
Respiratory problems	: recurrent respiratory infections in the first year of life.
Ocular abnormalities	: cataract, slight anti-mongoloid slant, hypertelorism.
Odd facies	: low forehead, hair growing low on forehead.
Skull abnormalities	: microbrachycephaly (Fig. 3). Head circumference below average for chronologic age and sex.
Nose	: not remarkable.
Ears	: slight anomaly of folding.
Highly arched palate	: present.
Height	: short stature; below average for chronologic age and sex.
Weight	: below average for chronologic age and sex.
Skeletal maturation	: below average for chronologic age and sex.
Delayed closure of large anterior fontanel	: present.
Broad thumbs and big toes and other distal phalanges by roentgenogram	: present.
Seizures with fever	: present.
Mental, motor, language and social retardation	: present. Psychomotoric: idiocy.

Summary

A baby-girl with all the features of the Rubinstein and Taybi syndrome, i.e. broad thumbs and toes, facial abnormalities and severe mental retardation has been reported.

REFERENCES

1. CAFFEY, J.: *Pediatric X-Ray Diagnosis*, 5th ed. p. 731 (Yb. Publ., Chicago 1967).
2. COFFIN, G.S.: Brachydactily, peculiar facies and mental retardation. *Amer. J. Dis. Child.* 108 : 351 (1964).
3. MOH. SUGIONO and TE BEK SIANG: Standard values of weight, length a.o. measurements of Indonesian infants in two M.C.H. centres in Jakarta. *Pediatr. Indones.* 4 : 171 (1964).

- 4. NELSON, W.E. : Textbook of Pediatrics, 9th ed. p. 753 (Saunders, London Toronto 1969).
- 5. RUBINSTEIN, J.H. and TAYBI, H. : Broad thumbs and toes and facial abnormalities. A possible mental retardation syndrome.

- Amer. J. Dis. Child. 105 : 88 (1963).
- 6. TAYBI, H. and RUBINSTEIN, J.H. : Broad thumbs and toes, and unusual facial features. A probable mental retardation syndrome. Amer. J. Roentgenol. 93 : 362 (1965).

A baby-girl with all the features of the Rubinstein and Taybi syndrome (broad thumbs and toes and facial abnormalities and fever) has been reported.

REFERENCES

1. RUBINSTEIN, J.H. and TAYBI, H. : Broad thumbs and toes and facial abnormalities and fever. Amer. J. Dis. Child. 105 : 88 (1963).

2. TAYBI, H. and RUBINSTEIN, J.H. : Broad thumbs and toes, and unusual facial features. A probable mental retardation syndrome. Amer. J. Roentgenol. 93 : 362 (1965).

3. NELSON, W.E. : Textbook of Pediatrics, 9th ed. p. 753 (Saunders, London Toronto 1969).