

Epidermal Ridge Configurations in a Boy with Triploid/Diploid Mosaicism *

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SUMMARY

The palm prints of a 2A,XY/3A,XXY patient displayed a scarcity of patterns and a wider than normal *atd* angle. Both hands had typical four-finger lines. The digits of the right hand had radial loops on II, III and V.

A series of observations on this case of triploid/diploid mosaicism (mixoploidy) has been reported earlier (Böök and Santesson, 1960*a* and *b*, 1961; Böök, 1961; Böök et al, 1962, 1963; Böök, 1964; Atkins et al, 1966). The patient, now 9 years of age, has been examined on several occasions since the discovery of his abnormal chromosome constitution at the age of one year. A comprehensive clinical and cytogenetical follow-up investigation was recently concluded. This report adds data on dermal patterns of fingers and palms.

The patient, M.G., at age 9 has a head circumference of 52 cm. The length of the skull is 20 cm. His ears are relatively large and sparsely moulded. His teeth are irregularly set with a frontal upper diastema and a deep bite. Physical examination of internal organs reveals nothing abnormal. General hypotonia of the muscles which are markedly underdeveloped. His penis and scrotum are normal, his testicles small and soft. He is severely mentally retarded and has shown no significant development as compared to earlier examinations. His IQ is estimated at 10-12. His motor age status corresponds to 13 months for the upper extremities and 17.5 months for the lower extremities, respectively.

His hands are small, oval and pawlike (Fig. 1). Major flexion creases, with the exception of marked four-finger lines, are practically lacking. His fingers are short and slender, particularly his thumbs. Major extension defects (clinodactyly) of the following fingers; left III, IV and V; right V. Syndactyly between III and IV right and left.

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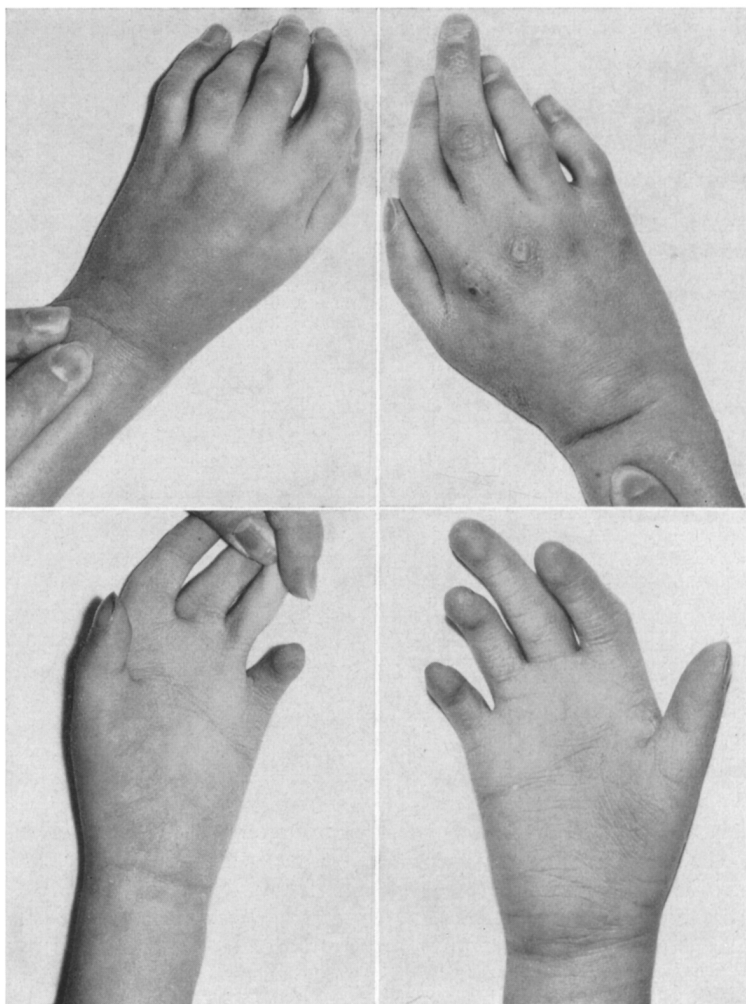


Fig. 1

Palm and finger prints. The analysis of the dermal ridge patterns has been summarized in Tab. I. The hand prints are primitive, in the sense of a scarcity of patterns. The left *atd* angle is wider than normal.

The digits of the left hand have no unusual patterns. Radial loops on V are quite rare in normal individuals, 0.20% according to Holt (1968), but sometimes occur in Down's syndrome and D-trisomy. Radial loops on left IV and V were also observed by Edwards et al (1967) in a premature baby of 32 weeks gestation who

showed XXY triploidy, only, in dermal cell cultures. Also this individual had radial loops on II which is extremely unusual in Down's syndrome.

The finger prints of two other triploid/diploid individuals, a 6 year old girl with XXX triploidy (Penrose, personal communication) and an 11 months old boy with 48, XXYY/71, XXXYY mosaicism (Schmidt and Vischer, 1967), showed no unusual patterns.

The two first mentioned cases had a very high percentage of triploid cells in dermal cultures at an early age, i.e., 90-100. The 6 year old girl (Ellis et al, 1963) had

Tab. I. Dermal ridge patterns

Digits and pattern areas	Right hand	Left hand	
Fingers	I	Whorl	Ulnar loop
	II	Radial loop	Whorl
	III	Radial loop	Ulnar loop
	IV	Ulnar loop	Ulnar loop
	V	Radial loop	Ulnar loop
Palms	Hypothenar	—	+
	Thenar and I	—	—
	II	—	—
	III	—	—
	IV	+	—
	Maximal <i>atd</i> angle	57°	65°
	Four finger lines	Typical	Typical

50%, which, however, in view of our observations (Böök et al, 1963), does not exclude the possibility that this percentage might have been considerably higher at an earlier age. The 11 months old boy had 26% triploid cells, only, in his dermal cultures.

The dermatoglyphic similarities of fingers and palms between this case and that of Edwards et al (1967) makes it likely that the observed abnormalities reflect, more closely, those of a triploid XXY dermis, e.g., rather of a dermis being of this constitution at the time of the formation of the ridge patterns.

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RIASSUNTO

I dermatoglifi palmari di un paziente 2A,XY/3A,XXY hanno rivelato scarsezza di disegni e un angolo *atd* più ampio della norma. Si sono riscontrate la linea delle quattro dita su ambedue le mani e anse radiali sul II, III e V dito della mano destra.

RÉSUMÉ

Les dermatoglyphes palmaires d'un patient 2A,XY/3A,XXY se présentent avec peu de figures et avec un élargissement de l'angle *atd*. Plis palmaire transversal sur les deux mains. Boucles radiales sur les doigts II, III et V de la main droite.

ZUSAMMENFASSUNG

In den Palmarlinien eines 2A,XY/3A,XXY-Patienten beobachtete man Mangel an Zeichnungen und einen grösser als normalen *atd*-Winkel. An beiden Händen fand man die Vierfingerfurche und am II, III, und V Finger der rechten Hand Radialschleifen.

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