ECTRODACTYLY

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This subject has not excited the attention of local medical men and a patient seen with so called lobster claw-hand or claw-foot is generally ignored. This is because in spite of the gross defects present, function is reasonably good and surgery can do little to improve this. Prosthetic 'social' hands are expensive and are not available locally. Moreover by and large these people have taken to their defects very well and do not seem to want any artificial gloves to make the hands look better at the expense of function.

The following seven cases are recorded mainly for their genetic importance. The first six cases are in a Chinese family (Pedigree A) where this condition having arisen as a mutation is transmitted in true Mendelian dominant manner. The further genetic aspects of this family will be discussed later. Case VII is interesting in that only one member of a family is affected (Pedigree B) and he too shows some variations that are of interest.

CASE SUMMARIES

CASE I: (II_2: Pedigree A).

The propositus was a 38 year old male Chinese, who died of carcinoma of the stomach last year. This man's hands and feet showed a variety of anomalies *viz.* syndactyly, polydactyly and ectrodactyly (Figs. 1, 2, 3, & 4).

Case II: (II_6: Pedigree A).

The affected sibling of the propositus, a 30 year old male Chinese, who is the chief witness states that the father died ten years ago and had no congenital abnormality and that the mother who lives in Johore is free from any anomaly.

He has typical ectrodactyly in the hands and feet. Some degree of syndactylism is also present (Figs. 5-8). One of his children is affected—Case VI. (III_{22}: Pedigree A).

CASE III: (III_5: Pedigree A).

An affected daughter of II_2 with ectrodactyly. Syndactyly is present in the left hand (where the bones of the middle and ring fingers are fused) and the right foot (fusion of the 4th and 5th toes). (Figs. 9, & 10).

CASE IV: (III_7: Pedigree A).

Another affected daughter of II_2 with ectrodactyly.

CASE V: (III_9: Pedigree A).

Another affected daughter of II_2 with ectrodactyly.

CASE VI: (III_{22}: Pedigree A).

The affected son of II_6 showing ectrodactyly in the feet only (Fig. 11). The hands are almost normal (Fig. 12) except for a tiny extra digit on left middle finger and an abduction deformity of the terminal interphalangeal joint of the right middle finger.

CASE VII: (II_1: Pedigree B).

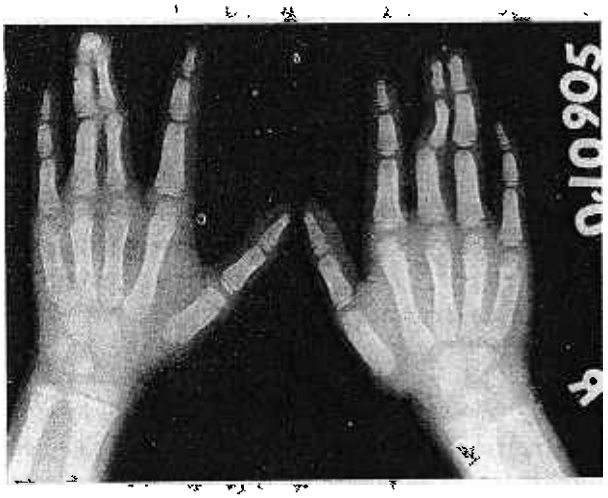


Fig. 13. Showing syndactly in the hands of Case VII.

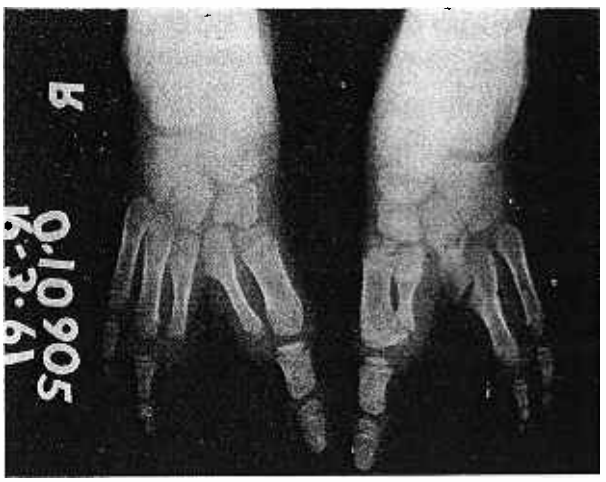


Fig. 14. Showing ectrodactyly in the feet of Case VII. Note the minor metatarsal aberrations.

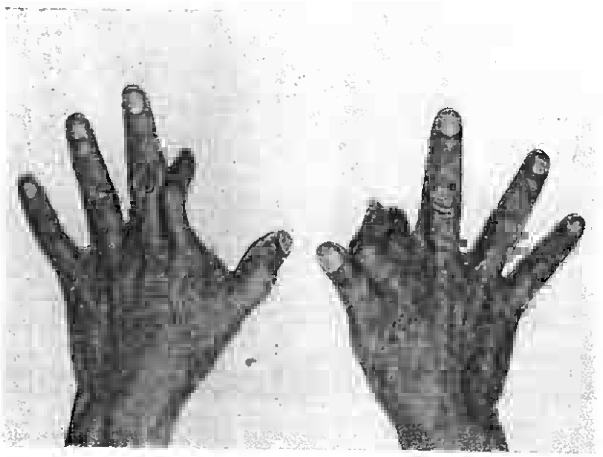


Fig. 1. The hands of Case I showing syndactyly in the left hand and syndactyly — polydactyly in the right hand.

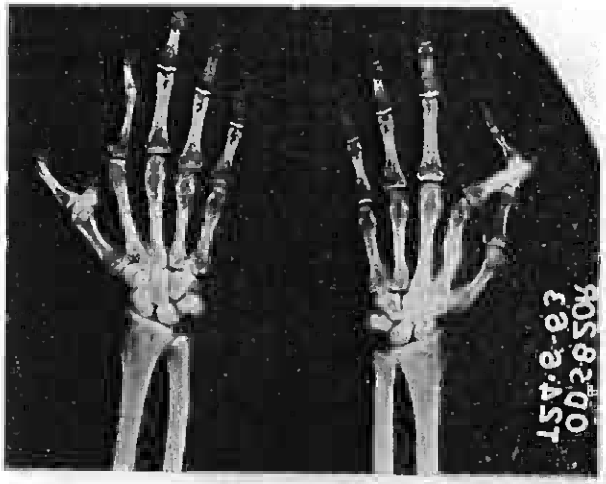


Fig. 2. The X-rays of the hands of Case I. Note that in the left thumb there are three phalanges and a tendency towards a bifid thumb (polydactyly). In the right hand note that 'bifid' index.

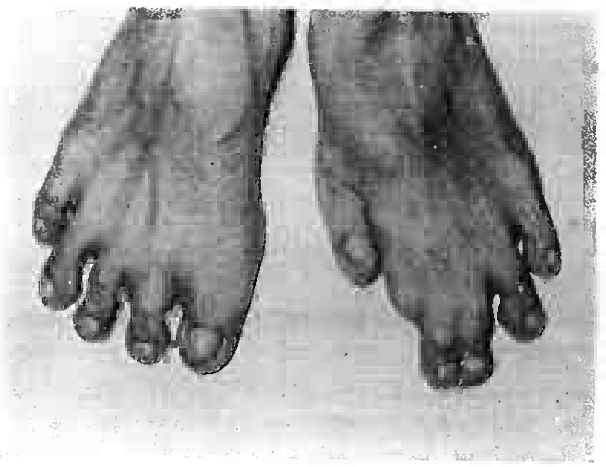


Fig. 3. The feet of Case I showing a normal right foot, a short left big toe and syndactyly of the left 2nd and 3rd toes.

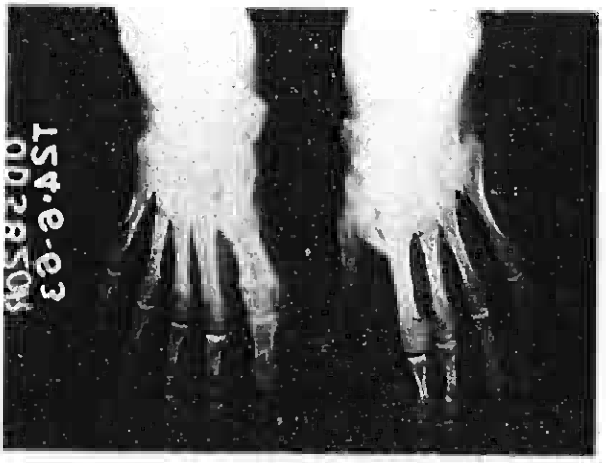


Fig. 4. The X-rays of the feet of Case I. Note the partial absence and abnormality of the left 1st metatarsal. There is also a partial duplication of this metatarsal.

Fig. 5. The hands of Case II showing ectrodactyly.

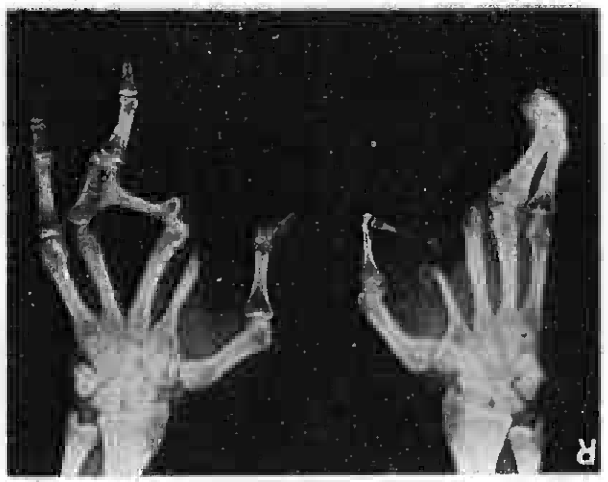


Fig. 6. The X-rays of the hands of Case II. Note the fusion of the proximal phalanges of the left middle and ring fingers and a fusion of the middle and terminal phalanges of the right ring and little fingers. There is also absence of phalanges in some fingers and extra phalanges in the thumbs.

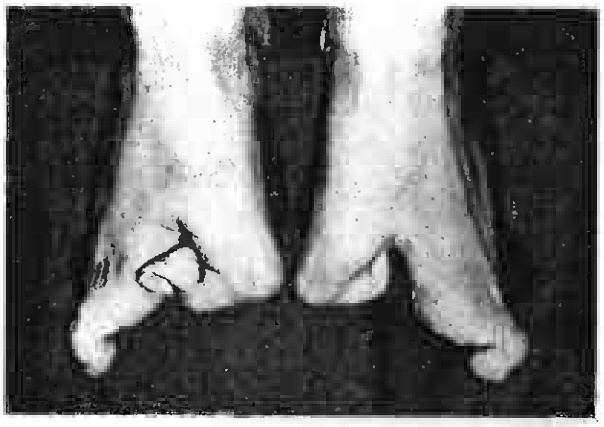


Fig. 7. The feet of Case II.

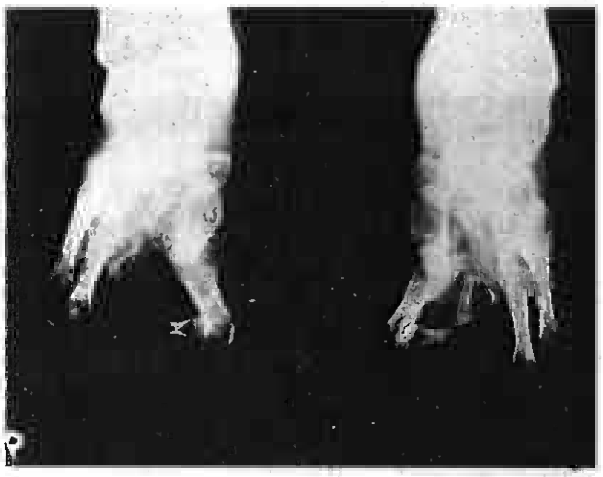


Fig. 8. X-rays of the feet of Case II where ectrodactyly is the dominant feature. Note that the heads of the 4th and 5th metatarsals articulate with the base of one phalanx.

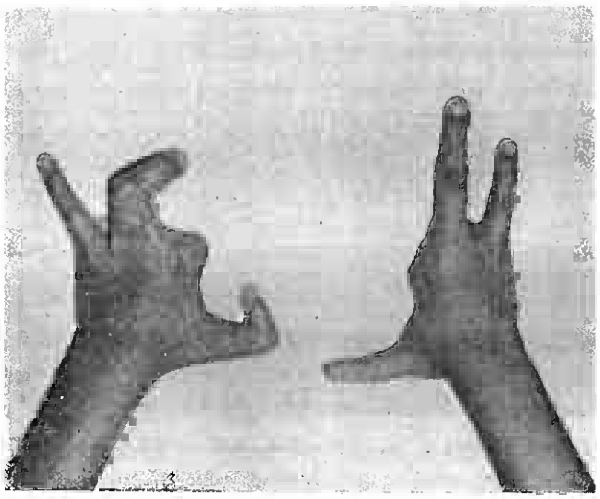


Fig. 9. The hands of Case III. An X-ray of the left hand showed a fusion of the phalanges of the middle and ring fingers.

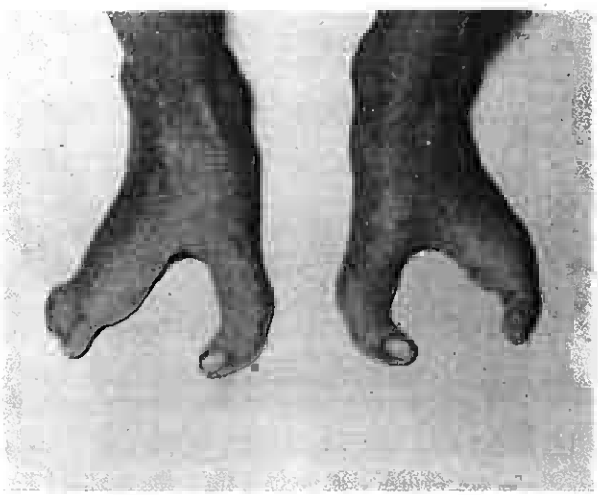


Fig. 10. The feet of Case III showing ectrodactyly and fusion of the right 4th and 5th toes.

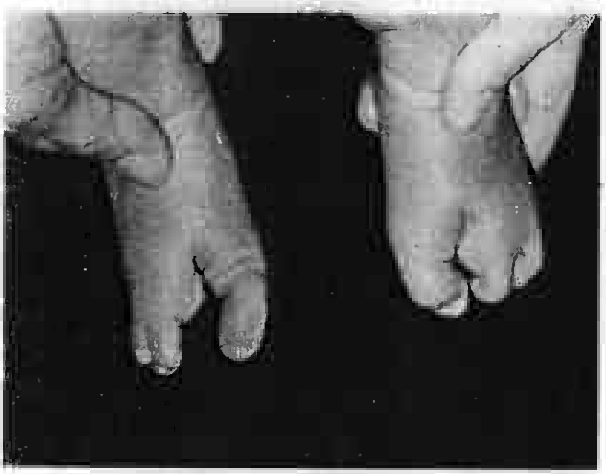


Fig. 11. The feet of Case VI showing ectrodactyly.

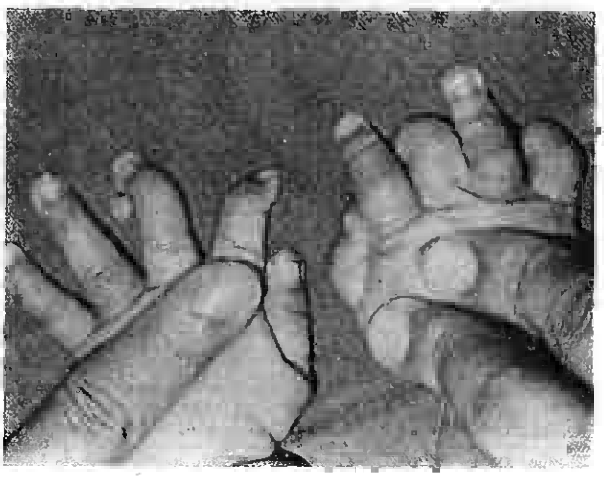


Fig. 12. The hands of Case VI showing an abduction deformity in the right middle finger and the small extra terminal phalanx of the left middle finger.

TABLE I

(PEDIGREE A). THE GENEALOGICAL TABLE FOR ECTRODACTYLY IN CASES I TO VI.

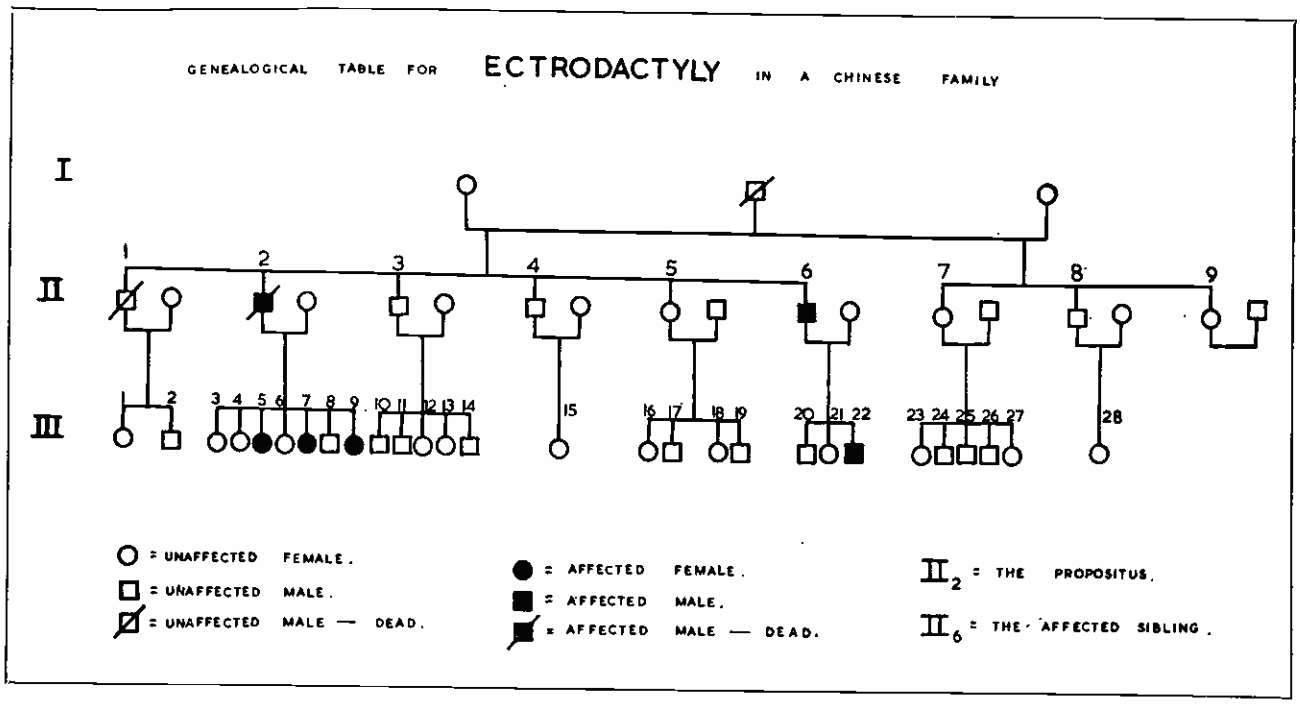
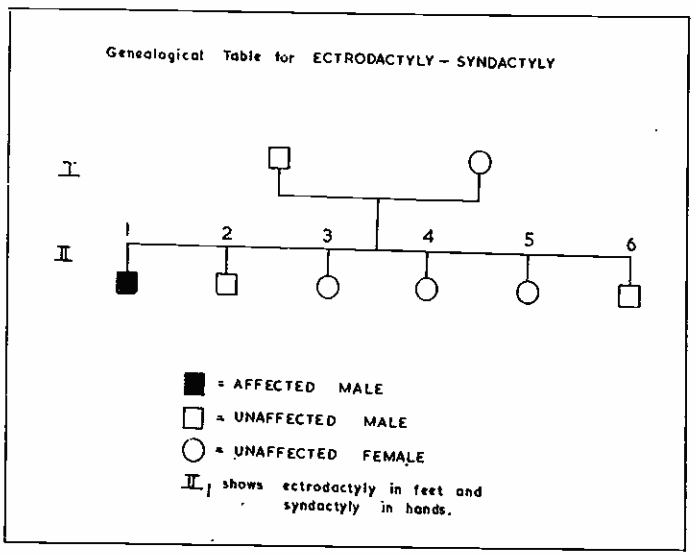


TABLE II

(PEDIGREE B). THE GENEALOGICAL TABLE FOR ECTRODACTYLY-SYNDACTYLY IN CASE VII.



A 10 year old male Indian showing syndactyly (of middle and ring fingers) in the hands and ectrodactyly in the feet. (Figs. 13 & 14). The fingers have been separated. No other member of his family is affected.

DISCUSSION

The term Ectrodactyly has been taken to mean the congenital absence of one or more digits (Barsky 1958). However to most people this is synonymous with claw (cleft) hand or claw foot. Birch-Jensen reported the incidence at birth to be 1:90,000. This condition is mainly inherited as a Medelian dominant trait, though it can manifest itself as a phenocopy—a congenital defect resembling one caused by genetic factors but due to other often indeterminable environmental factors.

In six of the reported cases (Pedigree A) genetic factors undoubtedly play a part. This pehigree is interesting from more than one aspect. It can be seen that the abnormality has manifested itself for the first time in the second generation—II_2—and that two members are afflicted—II_2 and II_6. Single mutations have been recorded many times in human genetics (Pillay 1964, Lee Kee Wee & Pillay 1964) but double mutations though common in animal genetics are rare in the human. However they have been previously recorded for Ectrodactyly (MacKenzie and Penrose 1951; Graham and Badgley 1955). For the nail patella syndrome (osteo-onychodysplasia) where absent or defective nails, dislocated radial heads, absent or small patellae and the presence of iliac horns form the characteristic tetrad, a double mutation has for the first time been noted (Pillay 1965). The explanation so far offered for this is that one of the parents is a gonadal mosaic *i.e.* the mutations involve only one sector of the gonad, the remainder being normal (MacKenzie & Penrose 1951) or that a delayed mutation has occurred (Auerbach 1956). Moreover in studying the manisfestation of this abnormal gene in Pedigree A one finds a varying expressivity and specificity of the gene *i.e.* the degree of manifestation and the form of manifestation differ in the individuals affected. II_2, the propositus, (the individual with the anomaly first seen by the author and from whom the study stems) showed evidence of syndactyly, polydactyly and ectrodactyly. (Figs. 1-4) In his children and the affected brother, mainly ectrodactyly is seen though some degree of syndactyly is also present. (Figs. 5-10) The affected child of II_6 shows ectrodactyly in the feet and in one hand there is a type of polydactyly present.

These variations may mean that modifying factors are present and that they are brought about either by the action of the allele or by environmental factors. In congenital constriction bands or annular defects it has been postulated that dietetic factors are important (Pillay 1964). This anomaly arises from germplasm deficiency (Streeter 1930) but the poorer Malays appear to be particularly prone to develop this condition (Pillay 1964).

In pedigree B, ectrodactyly with syndactyly is seen to be present in one member of a family. One has to study the progeny of this individual to be sure that a mutation has not taken place. However isolated examples of ectrodactyly and syndactyly have been reported, *i.e.* this lad could be what is termed a phenocopy.

SUMMARY

Two pedigrees for ectrodactyly are recorded. Pedgree A is interesting in that a double mutation has occured for this condition. This abnormality is closely associated with other hand anomalies such as syndactyly and polydactyly and all these seem to be the result of an abnormal gene showing varying expression.

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