Institute for Medical Genetics, University of Uppsala

On the Inheritance of Poly- and Syndactylies in Man

Lars Beckman, Lena Widlund

1. Introduction

Several different inherited anomalies of the hands and feet are known in man. In fact such anomalies were among the first human traits to be studied in the earliest period of Mendelian research in the beginning of this century. Thus it was observed very early that extra fingers and toes were inherited and that these traits usually seemed to follow a dominant, autosomal pattern of inheritance. The extra finger may be attached to the little finger or to the thumb. Sverdrup (1922) has described a large Norwegian family with supernumerary little fingers. As many as 37 individuals possessed the trait which could be traced in six generations. The trait was never transmitted through unaffected individuals and thus in this pedigree it behaved like a simple, autosomal dominant. In other pedigrees e. g. those reported by Jackson (1937) and Callan (1942) polydactyly was in several cases found to be transmitted by normal individuals, thus indicating a reduced expressivity of the trait. In a large Swedish pedigree Nylander (1931) found dominant inheritance of extra thumbs, which could be traced in five generations. In a negro family with 29 polydactylous individuals (Callan, 1942), one of the affected individuals had an extra thumb, while all the others had extra little fingers. In view of the relative rarity of the two types of polydactyly, the joint occurrence in one pedigree suggests that they are not strictly independent of each other from a genetical point of view.

Polydactyly may occur bilaterally or unilaterally on both hands and feet. In some individuals both hands and feet are affected (cfr. Jackson, 1937 and Callan, 1942). In some pedigrees it seems like polydactyly is caused by a recessive gene (Snyder, 1929 and Oliver, 1940).

In one family reported by Hefner (1940) extra thumbs were found to be associated with extra phalanges in the thumbs. In some individuals of this pedigree there were also duplications of the great toes. A very strange family was described by Lehmann (1952), in which syndactyly, a tendency to polydactyly and brachydactyly were found. Occurrence of poly- and syndactyly in the same family group was reported also by Vogel (1930). Other data on the inheritance of syndactyly suggest that this trait is inherited in a simple dominant way. In some cases syndactyly is confined only to the skin and superficial fascia (webbing), while in other cases there are also skeletal synostoses.

Ricevuto il 4. XII. 61

A remarkable pedigree with webbed toes was published by Schofield (1921), in which there were 13 affected males and 11 unaffected females. This pedigree seems to demonstrate a complete Y-linkage of the trait. In a work dealing with problems of complete Y-linkage in man, Stern (1957) has discussed the Schofield pedigree, which he classifies as a rather uncertain case of Y-linkage. Stern points out that the degree of penetrance in respect to webbed toes is different in males and females, webbing being mostly supressed in the female sex. In agreement with this Beckman, Gustavson, and Akesson (1961) found that out of 13 mongoloid idiots with syndactyly, 11 were males. The sexual difference of penetrance may have produced the result observed in the Schofield pedigree.

Walker (1961) studying polydactyly in a Batutsi family, claimed that the mode of inheritance in this family could be explained by postulating two dominant genes, one of which controls the penetrance of the polydactyly gene. Fraser Roberts (1959) believes that any one of a number of genes may produce somewhat similar, or even indistinguishable types of syndactyly in different families.

There exist various causistic reports on poly- and syndactyly, but larger investigations including reliable estimates of frequencies are rare. Therefore we decided to undertake a survey of poly- and syndactylies in the Swedish population.

2. This investigation

The data were collected from two different hospitals: the hospital of Sundsvall in North Sweden and the hospital of Jönköping in South Sweden. The records for all children born in these hospitals during the period of 1940-1960 were examined. In most cases there were complete data on poly- and syndactylies, including uni- or bilateral occurrence and exact location of the anomalies. The affected individuals found in this survey were then used as probands for further pedigree studies.

A) Frequencies

The frequencies of poly- and syndactylies of hands and feet showed no significant variations between samples from the two different geographical areas as can be seen in Table 1. In the total material the frequency of polydactyly of hands and feet were 0.37% and 0.08% respectively, while the frequencies of syndactylies of hands and feet were 0.14% and 0.22%. The frequencies observed in the present study are close to those reported by Böök (1951) for a South Swedish population.

Polydactyly of the hands is more common than on the feet ($X^2 = 12.45$, ld. f. $P<0.001$). In agreement with previous investigations (Stern, 1957 and Beckman, Gustavson and Akesson, 1961) there is a significantly higher incidence of syndactyly among males ($X^2 = 5.26$, ld. f. $0.025>P>0.01$). Further it may be noted that 3 males out of the 23 individuals with syndactyly possessed the trait on both hands and feet, and that 3 female individuals out of 29 examined had polydactyly on both hands

and feet. Two individuals showed both poly- and syndactyly. Thus it seems that there is a moderate correlation between the occurrence of the anomalies on hands and feet, and that there is a slight association between Poly- and syndactyly.

Tables 2 and 3 show the distributions of syndactylies according to the type of fusion involved. The most common type of fusion of the toes is that between the 2nd and the 3rd toes (in Tables 2 and 3 some individuals other than those obtained

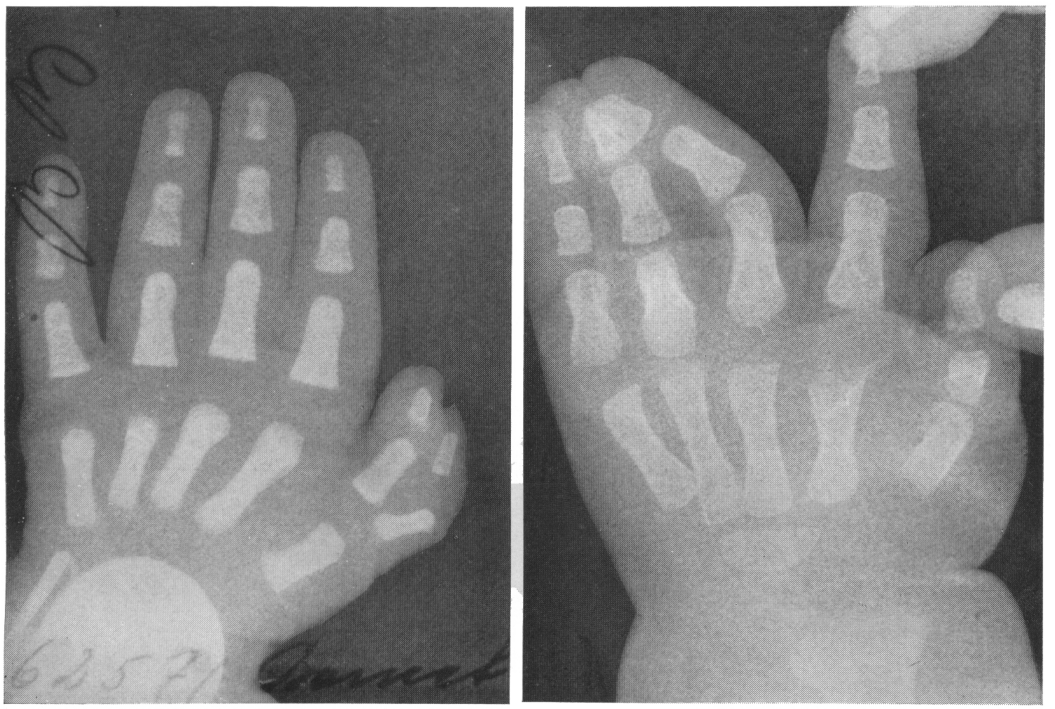


Fig. 1 X-ray photographs showing *a*) the hand of a newborn child wild with an extra thumb *b*) the hand of a child with fusions of fingers III-V, in this case there was bilaterally a skeletal synostosis only between the distal joints of fingers III and IV

from the hospital records of newborns have been included). In 5 out of 35 individuals examined there were multiple fusions of toes. The distribution in normal individuals has been compared with that of a sample of mongoloids (Beckman, Gustavson and Akesson, 1961). In 30 normal individuals with single fusions, 25 had syndactyly between toes 2 and 3, while in 13 mongoloids only 5 had syndactyly between the same toes. Thus there is a significant difference between the distributions. ($0.01 > P > 0.005$).

The most common type of syndactyly of the hands is the fusion between fingers 3 and 4. Two out of 17 individuals showed multiple fusions of fingers.

Tab. 1. Numbers of individuals with poly- and syndactyly of hands and feet in two different samples

Sample	Sex	Polydactyly		Syndactyly		Number Examined
		Hands	Feet	Hands	Feet	
Sundsvall	Males	10	0	2	3	29083
	Females	6	1	1	1	
Jönköping	Males	4	2	5	7	35710
	Females	4	2	1	3	
Total	Males	14	2	7	10	64793
	Females	10	3	2	4	

Tab. 2. Different types of syndactylies of the feet

Sample		Syndactyly Between Digits				Multiple	Total
		I-II	II-III	III-IV	IV-V		
Normal Individuals	Males	0	13	2	1	3	19
	Females	1	12	0	1	2	16
	Total	1	25	2	2	5	35
Mongoloids		1	5	2	5	0	13

Tab. 3. Different types of syndactylies of the hands

Sample	Syndactyly Between Fingers				Multiple	Total
	I-II	II-III	III-IV	IV-V		
Males	0	0	6	3	2	11
Females	0	1	5	0	0	6
Total	0	1	11	3	2	17

Tab. 4. Different types of polydactylies of hands and feet

Sample		Preaxial Polydactyly			Postaxial Polydactyly			Total
		R + L	R	L	R + L	R	L	
Males	Hand	1	4	0	4	0	5	14
	Foot	2	0	0	4	1	4	11
Females	Hand	0	3	3	3	1	1	11
	Foot	1	0	1	5	2	1	10
Total	Hand	1	7	3	7	1	6	25
	Foot	3	0	1	9	3	5	27

R = Right Side; L = Left Side.

Out of the 23 cases with syndactyly for which certain data on laterality were known, 12 had bilateral occurrence of syndactyly, while 8 had right-sided and 3 left-sided manifestations.

The distributions of different types of polydactyly are shown in Table 4. Postaxial polydactyly of the feet (little toes) is more common than the preaxial type (great toes). Concerning the hands there is no significant difference between the frequencies of preaxial and postaxial polydactylies and unilateral occurrence of preaxial polydactyly is more common than the bilateral type.

B) Pedigree studies

It was possible to trace relatives to 35 individuals found in the hospital survey and to get more or less complete information concerning the familiar occurrence of the anomalies in question. The data were collected through interviews with the mothers of the proband children. It may be questioned whether the lack of personal examination has introduced any bias. The traits dealt with are generally very easy to observe, and it would seem that if the mother claims that her brother has got an extra thumb, there is little reason to assume that she is not telling the truth. On the other hand one might possibly suspect that some people for emotional reasons tend to hide anomalies in their families and thus that some cases may not be reported.

The 35 pedigrees as well as detailed descriptions of the types of anomalies are given in the appendix. Nineteen pedigrees (1-17, 20, 21) show only syndactyly, nine pedigrees (22, 23, 26, 27, 28, 30, 32, 33 and 34) show only polydactyly and finally seven families (18, 19, 24, 25, 29, 31 and 35) include both poly-and syndactyly.

There were 21 probands with syndactyly and only 4 of these had one parent affected with the same anomaly. These 4 cases all had syndactyly between toes 2 and 3, viz. the most common type. Nine of the 21 probands had affected relatives. The number of polydactylous individuals in sibships with one affected sib is clearly lower than expected, assuming a dominant transmission of syndactyly.

Some of the more remarkable pedigrees are commented on below: In pedigree 3 only the proband and a second cousin are affected, while all the other relatives are unaffected. In pedigree 5 the proband has got syndactyly of the fingers, but among the relatives there is mainly syndactyly of the feet. Four individuals in pedigree 18 show syndactyly between toes 2 and 3, but there is also one relative with a double thumb. The proband in pedigree 19 has got both poly- and syndactyly, one relative is syndactylous and the others are polydactylous. Thus in two families traced by syndactylous probands there is a mixture of poly- and syndactyly.

Four pedigrees (22-25) originate from probands with doubled thumbs. In no case were there any relatives affected with the same type of anomaly. In pedigree 24, however, one relative of the proband had syndactyly of the feet and another one was missing fingers 2, 3 and 4 bilaterally. A rather strange situation is seen in pedigree 25, where the proband has got an extra thumb, but four relatives show bilateral occurrence of syndactyly between the 2nd and 3rd toes.

In three cases (26-28) probands with extra little fingers had no affected parents or sibs. In another family, however (no. 29) the same trait was found in four generations in a way that clearly suggests dominant inheritance. In this family there was again a mixture of poly- and syndactyly, one individual being syndactylous.

Two probands with extra great toes bilaterally had only unaffected parents and sibs (30, 31). In addition to the extra great toes proband 31 had bilateral syndactyly on both hands and feet.

The inheritance of extra little toes shows the same characteristics as does syndactyly and extra little fingers, viz. in some families there are mainly unaffected relatives, and in others (e. g. pedigree 35) the trait follows a dominant pattern of inheritance. Family 35 shows rather interesting features. Extra little toes occur in four generations. In the third generation there are six affected sibs, one of which has got also extra little fingers. (See the Appendix). Among the offspring of this individual there are four affected children, three of which have both extra little fingers and little toes. Thus in this pedigree it seems like when extra little fingers emerge in one individual together with extra little toes, there is a tendency for both traits to continue in the next generation. In family 35 there is also one individual with syndactyly in addition to polydactyly.

In none of the pedigrees in this study were there any signs of Y-linkage, comparable to the conditions found in the Schofield pedigree.

3. Discussion

The results of the present study indicates that the modes of inheritance for poly- and syndactylies in man are fairly complicated and that they frequently do not fit in with simple Mendelian ratios. Concerning syndactyly and extra little toes and fingers there is a heterogeneity between families, some pedigrees being fully compatible with dominant transmission of the traits and others not. It is also rather likely that syndactyly may occur as a plain phenocopy. This assumption is consistent with the fact that syndactyly of the feet occurs in a high frequency (up to 10%) in mongoloid idiots. If the syndactylies found in mongoloids were genetically determined in the same way as in normal individuals, it would be necessary to postulate a high incidence of this trait in the parents of mongoloids. Further it is interesting to note that although poly- and syndactyly occur in about the same frequency in the total population, there is a frequency increase in mongoloids only for syndactyly.

The inheritance of extra thumbs and great toes seems to be still more complicated. Preaxial polydactyly mostly occurs unilaterally, which points towards a low degree of penetrance. This may explain in part the absence of affected relatives. On the other hand it doesn't explain why one may find only syndactylous relatives when tracing the ancestry of an individual with an extra thumb. It is remarkable that one may start with syndactylous probands and find polydactylous relatives and vice versa. This indicates that in some families the manifested anomalies are in a way unspecific, and that there is a hereditary pleiotropic disposition towards disturbances of the distal parts of the extremities.

In three families poly- and syndactylies were associated with reductions of phalanges or even absence of fingers. It may be that in families showing several types of anomalies, the anomalies are caused by moderate chromosomal aberrations such as minor deficiencies, while in other families polydactyly or syndactyly are caused by single gene differences.

4. Summary

Data on the occurrence of poly- and syndactylies among 64793 newborn children were collected from the hospital records of two different Swedish hospitals, one being located in the North and the other in South Sweden. Frequency estimates are given for the different types of anomalies and the families of 35 probands were studied.

Some pedigrees were compatible with dominant transmission of the traits, while in others the mode of inheritance is irregular. There was no evidence for Y-linked syndactyly. In several cases it was found that a proband with polydactyly had relatives with syndactyly and vice versa. Some pedigrees included also individuals with reductions of phalanges and fingers.

References

Beckman, L., Gustavson, K.-H. and Akesson, H.-O.: Studies of some morphological traits in mental defectives. Hereditas (in press) 1961.

Böök, J. A.: The incidence of congenital diseases and defects in a south swedish population. Acta genet. 2: 289, 1951.

Callan, R.: *Polydactyly in a negro family. Jour of Hered.*, 33: 229, 1942.

Hefner, R. A.: Hereditary polydactyly. Jour. of Hered., 31: 25, 1940.

Jackson, V. W.: Four generations of polydactyly. Jour. of Hered., 28: 267, 1937.

Lehmann, W.: Ueber eine Familie mit multiplen Missbildungen an Händen und Füssen. A. Ge. Me. Ge., 2: 87, 1953.

Nylander, E.: Präaxiale Polydaktylie in fünf Generationen einer Schwedischen Sippe. Upsala Läkareförenings förhandlingar, 36: 275, 1931.

Oliver, C. P.: Recessive polydactylism. Jour. of Hered., 31: 365, 1940.

Roberts, J. A. Fraser: An introduction to medical genetics, p. 103, 1959.

Schofield, R.: Inheritance of webbed toes. Jour. of Hered., 12: 400, 1921.

Snyder, L. H.: A recessive factor for polydactylism in man. Jour. of Hered., 20: 73, 1929.

Stern, C.: The Problem of Complete Y-Linkage in Man. Am. J. Human Genet., 9: 147, 1957.

Svedrup, A.: Postaxial polydactylism in six generations of a Norvegian family. J. of Genetics, 12, no. 3, 1922.

Vogel, K.: Uber familiäres Auftreten von Polydaktylie und Syndaktylie, Fortschritte der Röntgenstralen, 20: 433, 1913.

Walker, J. T.: A pedigree of extra-digit-V polydactyly in a Batutsi family. Ann. Hum. Genet., 25: 65, 1961.

Appendix

(Detailed description of the types of poly- and syndactylies in the pedigrees)

1. N. 1, synd. bilat. toes 2-3. N. 2, synd. right toes 2-3.

2. N. 1, synd. bilat. toes 2-3. N. 2, synd. left toes 2-3.

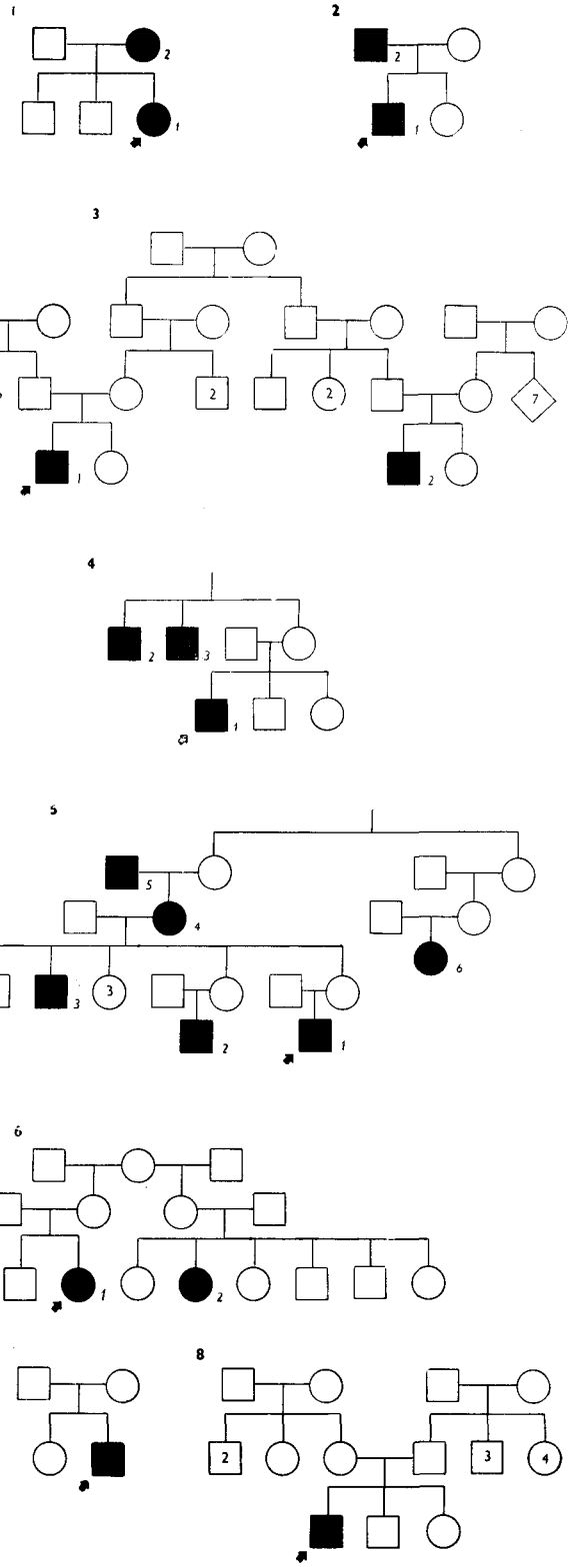
3. N. 1, synd. bilat. fingers 3-4-5 and synd. bilat toes 3-4. N. 2, synd. right fingers 2-3 and 4-5.

4. N. 1, synd. left fingers 4-5, right toes 2-3 and left toes 1-2-3. N. 2 and 3, type unknown.

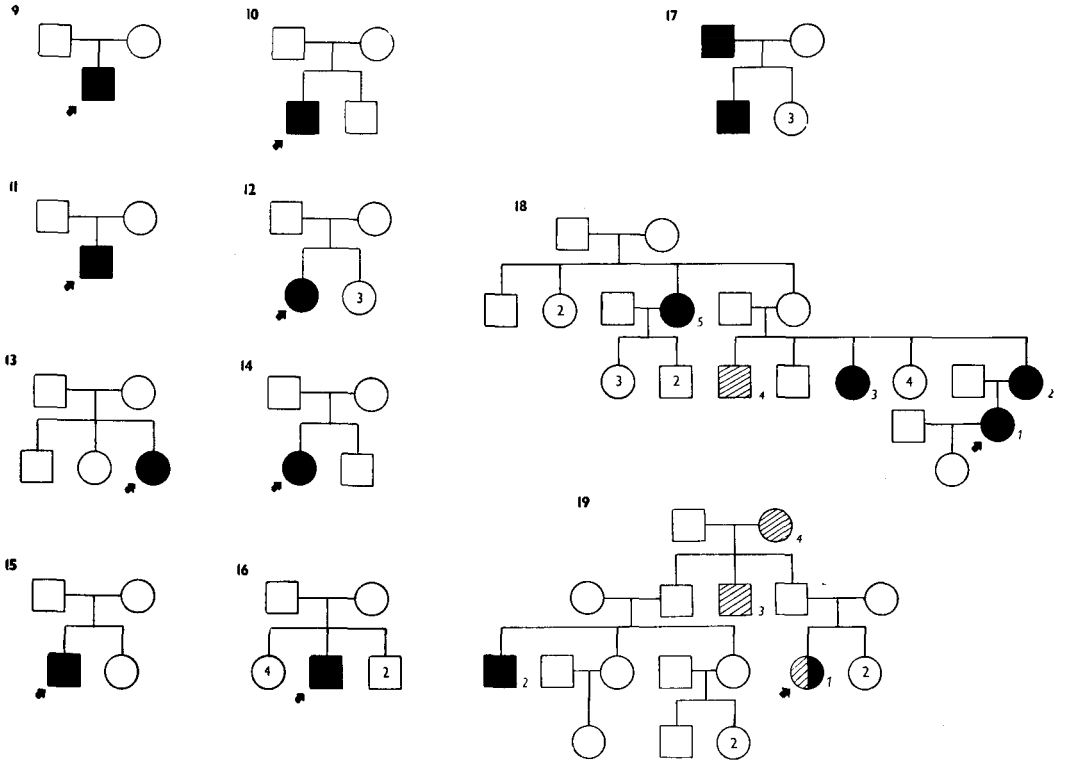
5. N. 1, synd. left fingers 3-4-5. N. 2, synd. left toes 2-3. N. 3, synd. bilat. toes 2-3. N. 4, synd. one hand. N. 5, synd. one foot. N. 6, synd. bilat. toes 2-3.

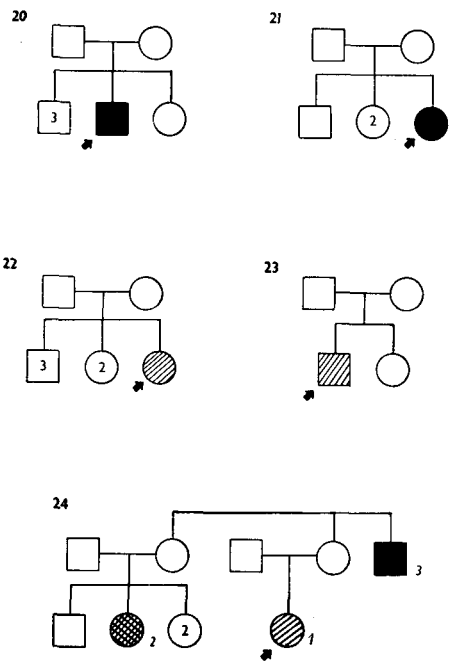
6. N. 1, synd. bilat. toes 2-3. N. 2, synd. right toes 2-3-4.

7. Synd. bilat. fingers 3-4.
8. Synd. right toes 2-3.



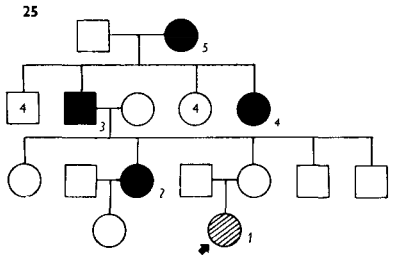
https://doi.org/10.1017/S1120962300020448 Published online by Cambridge University Press

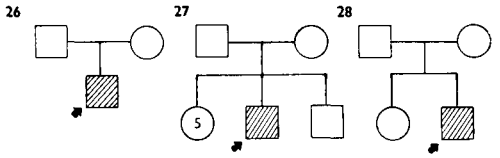


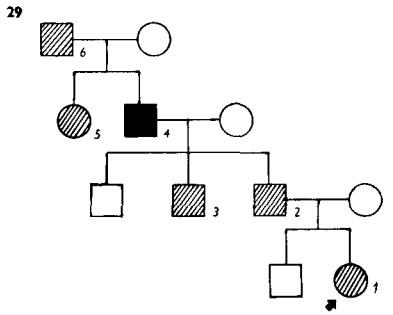


9. Synd. right fingers 4-5.
10. Synd. right toes 4-5.
11. Synd. bilat. fingers 3-4.
12. Synd. left fingers 3-4 and synd. bilat. toes 2-3 and 4-5. Missing phalanges on right finger 3 and left finger 2.
13. Synd. right toes 2-3.
14. Synd. bilat. fingers 3-4 and synd. bilat. toes 2-3.
15. Synd. left fingers 3-4.
16. Synd. left fingers 4-5.
17. N. 1 and 2, synd. bilat. toes 2-3.
18. N. 1, 2, 3, and 5, synd. bilat. toes 2-3. N. 4, extra thumb one side.
19. N. 1, synd. bilat. toes 4-5 and extra little toe right. N. 2, synd. left toes 2-3. N. 3 and 4, extra little fingers.
20. Synd. right toes 2-3 and 4-5.
21. Synd. right toes 2-3-4, left toes 3-4 missing and right fingers 2-3-4-5 with rudimentary phalanges.
22. Polyd. right thumb.
23. Polyd. right thumb.
24. N. 1, polyd. one thumb. N. 2, missing fingers 2-3-4 bilat. N. 3, synd. one foot.

https://doi.org/10.1017/S1120962300020448 Published online by Cambridge University Press







25. N. 1, polyd. left thumb. N. 2, 3, 4, and 5, synd. bilat. toes 2-3.

26. Polyd. left little finger.

27. Polyd. left little finger.

28. Polyd. little fingers.

29. N. 1, 2, 3 and 5, polyd. little fingers, N. 4, synd. type unknown. N. 6, polyd. little fingers and toes.

30. Polyd. bilat. great toes.

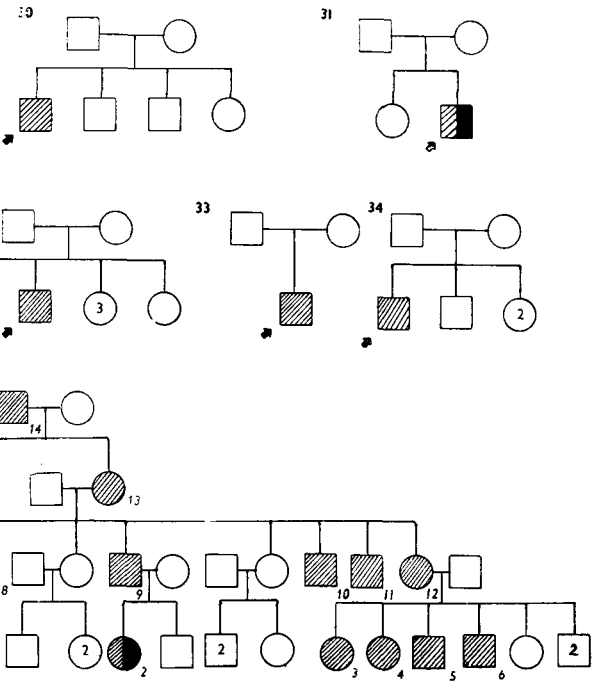
31. Polyd. bilat. great toes, synd. right fingers 2-3, left fingers 1-2-3 and synd. bilat toes 1-2-3.

32. N. 1 polyd. left little toe. N. 2 polyd. bilat. little toes.

33. Polyd. right little toe.

34. Polyd. right little toe.

35. 5, 8, 9, 10, N. 11, 13 and 14, polyd. bilat. little toes. N. 1 and 7 polyd. left little toe. N. 6 and 12, polyd. bilat. little fingers and toes. N. 2, polyd. right little toe and synd. right toes 4-5. N. 3, polyd. bilat. little toes and right little finger. N. 4, polyd. right little toe and left little finger.



https://doi.org/10.1017/S1120962300020448 Published online by Cambridge University Press

RIASSUNTO

I dati sulla frequenza della poli- e della sindattilia in 64.793 bambini appena nati sono stati raccolti dagli archivi di due diversi ospedali svedesi, uno al nord e l'altro al sud della Svezia. È riportata la frequenza delle stime per i diversi tipi di anomalie e sono state studiate le famiglie di 35 probandi.

In alcuni alberi genealogici la trasmissione dei caratteri era dominante, mentre in altri era irregolare. Non c'era nessuna prova per la sindattilia legata al cromosoma Y. In parecchi casi è stato trovato che un probando con polidattilia aveva parenti affetti da sindattilia e viceversa. Alcuni alberi genealogici comprendevano anche individui con falangi e dita ridotte.

RÉSUMÉ

On a collectionné des informations sur l'apparition de poly- et syndactylie parmi 64.793 nouveau-nés dans les régistrations de deux différents hôpitaux suédois, l'un du nord et l'autre du sud de la Suède. Des estimations de fréquence sont données pour différents genres d'anomalies et les familles de 35 probands ont été étudiées.

Quelques arbres généalogiques permettent la conclusion d'une transmission dominante du caractère, tandis que dans d'autres le mode de transmission est irrégulier. On n'a pas trouvé d'évidence de syndactylie Y-liée. Dans plusieurs cas on a trouvé dans le parenté d'un proband avec polydactylie d'autres individus souffrant de syndactylie et vice-versa. Quelques arbres généalogiques comprenaient des individus avec des réductions des phalanges et des doigts.

ZUSAMMENFASSUNG

Data über das Auftreten von Poly- und Syndaktylie unter 64.793 Neugeborenen wurde von Eintragungen in zwei verschiedenen schwedischen Spitälern gesammelt, eines im Norden und eines im Süden von Schweden. Schätzungen über die Häufigkeit der verschiedenen Anomalie Typen sind gegeben und die Familien von 35 Probanden wurden studiert.

Einige Stammtafeln stimmten mit der dominanten Transmission der Erbanlage überein, währenddem in andern die Vererbung unregelmässig ist. Für Y-angegliederte Syndaktylie fand man keinen Beweis. In einigen Fällen fand man, dass ein Proband mit Polydaktylie Verwandte mit Syndaktylie hatte und viceversa. Einige Erbtafeln schlossen auch Personen mit Reduktionen von Fingerknochen und Fingern ein.

https://doi.org/10.1017/S1120962300020448 Published online by Cambridge University Press