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A Note in the Genetics of Koilonychia

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Koilonychia (Heller, 1900), sometimes called the spoon nail (Crocker, 1896), is one of the rare abnormalities of nails in man. Nails, not only of fingers but sometimes of toes, are concave, warped, and turned upward at the distal end, and have the lateral margins loosely attached to the skin. Most of the recorded cases are sporadic, and the abnormality develops in relation with some environment factor. Familial congenital cases have also been reported by some investigators (Wilson, 1905; Ormsby, 1926; Cipollaro, 1930; Riecke, Eddow c. f. Cipollaro and others). Gates (1946) assumes dominant inheritance in these pedigrees. However, the genetics of this abnormality has not yet been fully elucidated. The present authors found a large Japanese kindred which includes 36 affected members extending in 5 generations. This kindred seems to show that this congenital abnormality is due to a simple dominant autosomal gene.

Clinical Aspect of the Propositus

Propositus (III-26), a house-wife, 40, living in Wakayama City, complained of languor in limbs, and was referred to Wakayama Public Health Center for consultation.

Physical Examination. The patient has a moderately developed and well nourished physique, the conjunctiva shows slight anemicity; pupil reaction and accomodation normal; no pathological change in mouth cavity; heart, lunghs, abdominal and pelvic viscera normal, nervous and all other physiological functions also normal. In fact, a thorough physical examination of her revealed nothing pathological. No helminth eggs were present in her faeces. The abnormality of nails is congenital, but causes no inconvenience in daily life. The nails are flattened, curved upward at distal end and loosely attached to the skin on lateral margins. The deformity is particularly conspicious in thumb nails, which are spoon-like in shape with the lateral and terminal margins turned upward; this peculiarity becomes gradually less conspicious toward the little fingers. Pressure on the nails causes no pain. The nails are pale in color and have pearl-like luster, the lunula cannot be recognized. The nails are rather thin and soft in texture, and easily injured; but have no fissure, crack, ridge, striation, pit or any sign of leuconychia.

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Family History

According to the propositus, this congenital abnormality is found in many members of her kindred: her mother has similar nails, while her deceased father and husband had normal nails. Her only son has spoon nails in all his fingers.

Family Study

Thirty-six members are affected among the whole 114 extending in 5 generations (Fig. 1).

The sibs of the propositus and their children.

There are 4 members (3 males and 1 female) including the propositus; III-24 (M) died when 6 years old by a traffic accident, had normal nails. III-25 (M) has spoon nails in all fingers and toes. He has 2 sons between the first wife who was a normal cousin; the elder son (IV-45) is affected, the abnormality is found in all nails except of the fourth on both feet. The younger son is normal, and married again after the death of his first wife, and has a normal son. III-26 (F) is the propositus who has an affected son, already mentioned. III-27 (M) is affected, and the abnormality is observed in all nails except on the first toes. Between a normal wife, he has two sons of whom the first (IV-48) has the abnormality in all nails, while the second is normal.

The mother of the propositus and her sibs.

The mother of the propositus (II-13), 65 years old, is alive and healthy. The nails of her left hand show the typical spoon shape, namely, the nails of the middle and index fingers of the right hand are concave, while the others are flat. The toe nails appear normal. She is the fifth of 9 sibs (3 males and 6 females). The eldest (II-6) (M) who died when 4 years old by an accident, was normal. II-7 (F) and her eldest son (III-11) are dead. Her second normal son remembers that they both had abnormal nails. II-9 (F) is said to have been normal and married her cousin who had spoon nails (II-10). According to II-13 and III-17, the couple have 7 children (3 males and 4 females), of whom only the third female child is abnormal; all the remaining six are normal and have 9 normal children (6 males and 3 females) and 5 normal grandchildren (2 males and 3 females). III-17 (F), 37 years old, is most severely affected, all her nails being of the typical spoon shape (Figs. 2 and 3). She married her cousin (III-21) who is also affected, and has 4 children (2 males and 2 females), of whom the first (IV-35) (M), 16 years old, and the second (IV-36 (F), 14 years old, show the abnormality in both hands. The third (M) is normal, but the fourth (IV-38) (F) has abnormal nails much like her mother's. The fourth sib of the mother of the propositus (II-11) was affected, and married her normal cousin who is a younger brother of II-10. The couple have four sons; the first (III-20) (M) was affected, and died when 33 years old; he left 4 children (2 sons and 2 daughters); the first (II-31) (M) shows the abnormality in all his nails, and his 2 daughters (V-8

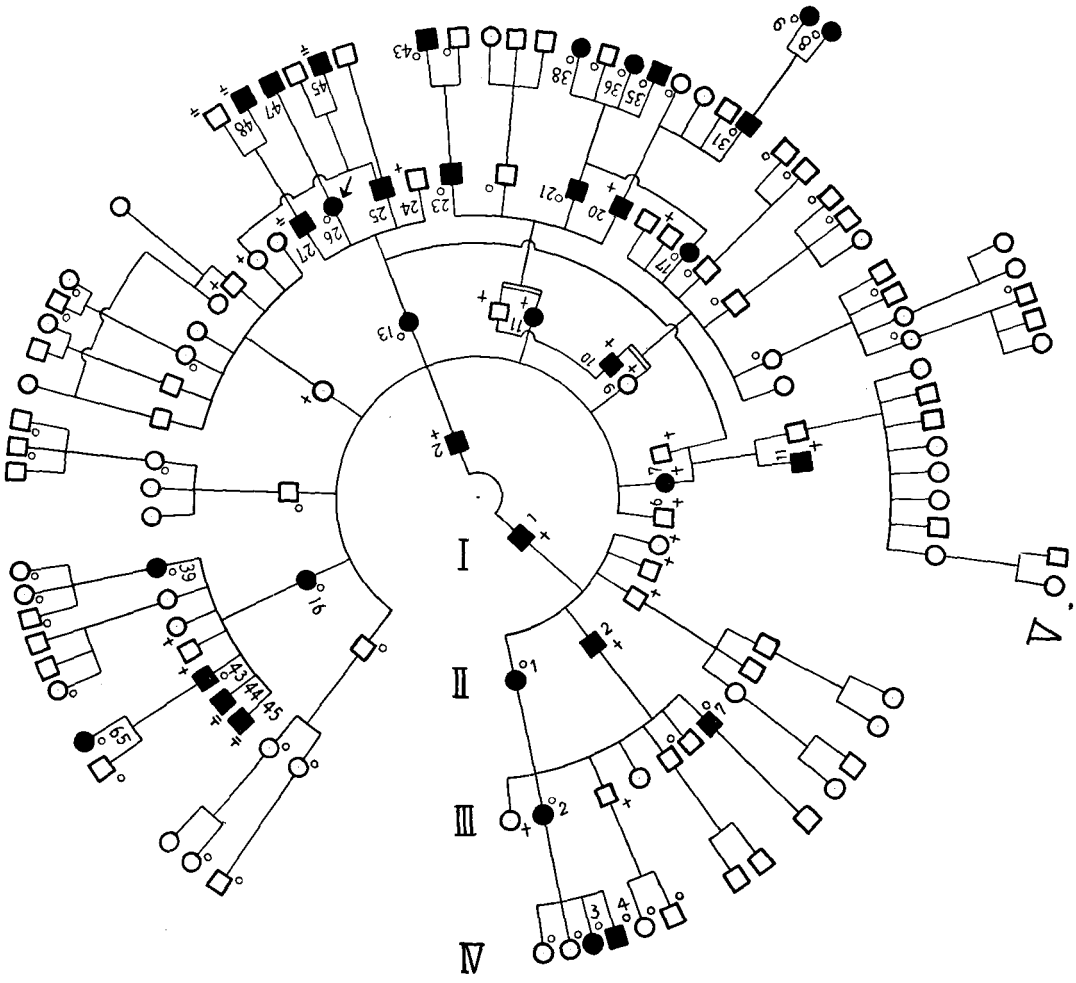

Fig. 1 The Pedigree of Koilonychia
†: dead
O: examined
±: confirmed by mail

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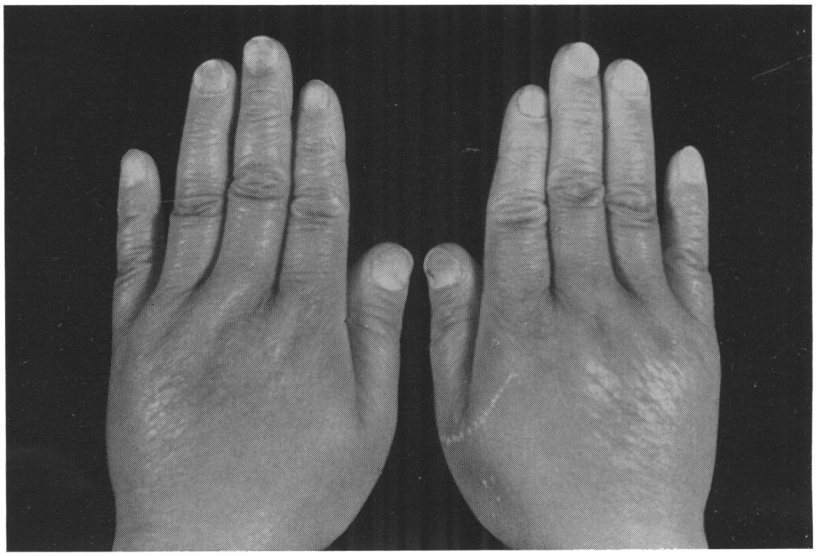

Fig. 2. Hands of III – 17

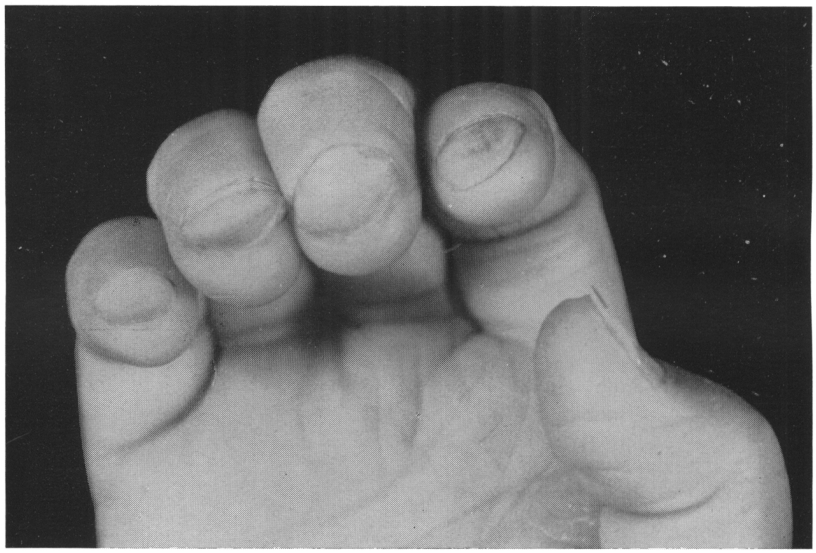

Fig. 3. Nails of III – 17

and 9) are also affected. The other 3 members (III-21, 22 and 23) are all nearly normal. III-21 shows the abnormality only in his left hand: the nails of the thumb, middle and index fingers are concave while those of the ring and little fingers are flat; the right hand and both feet bear normal nails. As recorded above, he married an affected cousin. III-22 (M) and his 3 children (2 males and 1 female) are all normal. III-23 (M) has spoon nails in both hands, and of his 2 sons, the younger (IV-43) (M) has the abnormal nails except on both ring fingers. The sixth sib of the mother of the propositus is dead, and was apparently normal. Her 7 childern (3 males and 4 females), and all grandchildren are normal. Her younger brother (II-15), as also his 3 daughters and 3 grandsons are normal. II-6 (F) has spoon nails on all fingers, except the thumbs. Between a normal spouse, there are 4 males and 3 females: the first (III-39) (F), 37 years old, shows the abnormality in nails of thumbs, middle and ring fingers of both hands, while the other nails are flat. She has 3 normal children (1 male and 2 females). The second (F) and her 3 children (2 males and 1 female) are normal. The third (F) and the fourth (M) who died in youth, had normal nails. The fifth (III-43) (M) hdodtuoaon nail on thumbs, middle and ring fingers of both hands, the other nails being flat. Of his 2 small children, the 2-year old daughter (IV-65), shows the abnormality in all nails of the right hand, while a 100-day old baby boy seems to be normal. Both the unmarried youngest sons of II-16, (III-44) and (III-45) (M), have deformed nails in both hands. The youngest sib (M) of the mother of the propositus is normal, and has 2 normal daughters and 3 normal grandchildren (1 male and 2 females).

The grandparents of the propositus and their sibs.

According to the memories of II-12 and II-14, the grandfather (I-2) of the propositus was affected, while his wife was normal, and his elder brother (I-1) had spoon nails. The latter had 5 children (3 males and 2 females) between a normal spouse; the first (II-1) (F), 73 years old, shows the anomaly in eight finger nails except of thumbs, the abnormality is also apparent in both big toes, while other toe nails are normal. Of her descendants, the first son who died at the age of 23, and the second son who died in the World War, were normal, while the second daughter (III-2) (F), 46 years old, shows the abnormality in all finger and toe nails except for normal small-toe nails. She has 4 children between a normal husband: the first and the second daughters are normal, while the third (IV-2) (F), 15 years old, and the fourth (IV-4) (M) have abnormal nails on all fingers. The second son (II-2) (M) of the grandfather of the propositus is dead. His abnormality is remembered by II-1. Of his 4 children (3 males and 1 female), the first and the second are alive, while the third died in the World War, and they are all normal. The fourth (III-7) (M), 32 years old, shows the abnormality having spoon-shaped nails on all fingers except on both little fingers. His son is normal. The third, fourth and fifth sibs of II-1, died 50, 25 and 12 years old respectively. According to II-1, they were all normal, so also their descendants: 3 children (2 males and 1 female) and 4 grandchildren of the third (2 males and 2 females).

Genetic Analysis of the Data

The mode of the inheritance.

The kindred includes 36 affected members extending in five generations (Fig. 1). The 17 marriages between an affected member and a normal member which have more than 2 children, produced 29 affected (16 males and 13 females) and 37 normal progeny (22 males and 15 females). This ratio 37 : 29 falls within the limit of error of the 1 : 1 ratio of the affected and normal offspring. The sex ratio of the affected members, 16 males : 13 females, also is about 1 : 1. Thus, the abnormality seems to be due to a single autosomal dominant gene (Table 1 and 2).

Table 1 – Normal and affected offspring from normal-affected matings

Size of sibship	No. of sibship	Male children		Female children	
		Normal	Affected	Normal	Affected
2	6	5	4	0	3
3	2	2	0	3	1
4	5	5	8	5	2
5	1	2	1	1	1
7	2	5	3	4	2
9	1	3	0	2	4
Total	17	22	16	15	13

Table 2 – Test of hypothesis of autosomal dominant inheritance

Children		Normal	Affected	Total	D. F.	χ^2	P
Male:	observed	22	16	38	0.947	1	
	expected	19	19	38			
Female:	observed	15	13	28	0.142	1	
	expected	14	14	28			
Total:	observed	37	29	66	0.969	1	$0.5 > P > 0.3$
	expected	33	33	66			

Table 3 – Test for homogeneity within sexes

Source	χ^2	D. F.	P
Sum	1.089	2	
Total	0.969	1	
Heterogeneity	0.020	1	$0.90 > P > 0.80$

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Penetrance and expressivity of the gene.

In the family, the abnormality appears in each of the five generations, and never skips a generation; moreover, it is found in none of the progeny of normal matings. Thus, penetrance of the gene in the family is apparently fairly high.

The characteristic spoon-like shape is not always found in all nails of the affected member. Among the 25 affected members carefully examined, fourteen had spoon-like nails on all fingers of both hands, 4 only on those of one hand, while 2 members showed the defect on all fingers except thumbs, or small fingers, and in 3 members the abnormality is seen on some other fingers. Moreover, the grade of concavity of the abnormal nails is not the same, some being deeply and others shallowly concave. In all these cases, however, the affected nails distinctly differ from normal ones, as the nails are more or less deformed, pale-blue in color, and somewhat softer in texture than the normal nails, and show no lunula at the root.

The abnormality in toe nails is more difficult to discriminate, as deformed nails developed by due to post-natal effects like shoe wearing, etc. are very common. Thus, the diagnosis of the abnormality for toe nails of the members of this kindred is more uncertain than that for hand nails.

Discussion

This rare abnormality of nails has been recorded sporadically by dermatologists. According to Cipollaro's review (1930) of the literature, about 35% of the cases have a genetic background or due to special occupations, using specific chemicals, such as copper vitriol, sulphuric acid and some strong alkaline. As Ormsby (1917) and Cipollaro point out, familiary occurrence of the abnormality is sometimes found, Wilson's report (1905) suggests that the abnormality may be due to a dominant gene, since he has found a family in which seven members in three generations had spoon nails: the grandmother was abnormal and had 4 affected (2 males and 2 females) and 9 normal children, and her tenth female child had 2 affected sons among her 3 children; the abnormality was congenital and not accompanied by any anomaly or disease. In Eddow's pedigree (cited by Crocker, 1950) the abnormality appeared in 3 sibs and their father's sibs. Riecke records a female patient who had 4 affected children, Cipollaro's pedigree includes 3 affected members, mother and two children, the kindred comprised 36 cases among 114 members, and showed simple autosomal dominance of the abnormality. He points out, however, that the recessive inheritance is not entirely excluded, because about 35% of the recorded cases are sporadic, and have no assignable cause. In Heller's pedigree (1937) the defect is shown in 4 children of normal parents.

About the penetrance of the gene, our study indicates that it is fairly high. In both Wilson' and Wälsch's pedigrees, the defect extend in 3 generations, while in Eddow's pedigree, the parents of the propositus are both normal, though some of the father's sibs are affected.

The expressivity of the abnormality differs in different cases.

According to the complications of the defect, about 30% of the cases have some associated disease, such as eczema, lichen planus, psoriasis and acanthosis nigricans, which are found mostly in sporadic cases. Ormsby's case had congenital alopecia and deformed teeth. In Heller's case Basedow's disease and hyperthyreoidism accompanies the koilonychia. These cases are rather exceptional, and the abnormality is seen without any complication in the majority of cases.

Summary

Our study on a kindred of spoon nails found among residents in Wakayama City, shows:

1. The kindred includes 36 affected members extending in five generations (Fig. 1). The seventeen marriages between affected and normal members of the kindred and which have more than two children, produced 29 affected, 16 males and 13 females, and 37 normal children 22 males and 15 females. This 37 : 29 ratio falls within the range of the 1 : 1 ratio of the affected and normal offspring. The sex ratio of the affected members, 16 males : 13 females, also is about 1 : 1. Thus, the abnormality seems to be due to a single autosomal dominant gene.

2. The penetrance of the gene in this family is apparently fairly high.

3. Regarding the expressivity of this abnormality, the concavity of nails is not always distinct especially in toes, while the expressivity of the gene in finger nails is more apparent.

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RIASSUNTO

Il nostro studio su di una famiglia di affetti da coilonichia, trovata fra abitanti di Wakayama City, dimostra che:

1. La famiglia comprende 36 membri affetti distribuiti in 5 generazioni (Fig. 1). I 17 matrimoni, con più di due figli, avvenuti fra membri affetti e normali della famiglia, hanno prodotto 29 bambini affetti (16 maschi e 13 femmine) e 37 normali (22 maschi e 15 femmine). Questa proporzione di 37:29 rientra nell'ambito della proporzione di 1:1 fra discendenti affetti e normali. La sex-ratio dei membri affetti (16 maschi: 13 femmine) è ugualmente di circa 1:1. La anormalità sembra, quindi, dovuta ad un solo gene autosomico dominante.

2. La penetranza del gene in questa famiglia è apparentemente molto elevata.

3. Per quanto riguarda l'espressività di questa anormalità, la concavità delle unghie non è sempre molto chiara, specialmente nelle unghie dei piedi, mentre nelle unghie delle mani l'espressività del gene è più apparente.

RÉSUMÉ

Notre étude sur une famille d'affectés de Koilonychie, trouvée parmi les habitants de Wakayama City, démontre que:

1. La famille comprend 36 membres affectés, distribués dans 5 générations (Fig. 1). Les 17 mariages entre membres affectés et normaux de cette famille, ayant eu plus de deux enfants, ont produit 29 enfants affectés (16 mâles et 13 femelles) et 37 normaux (22 mâles et 15 femelles). Cette proportion de 37:29 rentre dans la proportion de 1:1 entre descendants affectés et normaux. La sex-ratio des membres affectés (16 mâles: 13 femelles) est également de 1:1 environ. Cette abnormalité paraît, donc, être due à un seul gène dominant autosomique.

2. La pénetrance du gène dans cette famille semble être très élevée.

3. En ce qui concerne l'expressivité de cette abnormalité, la concavité des ongles n'est pas toujours très claire, surtout dans les doigts des pieds, tandis que dans les ongles des doigts des mains l'expressivité du gène est plus apparente.

ZUSAMMENFASSUNG

Unsere Untersuchung über eine Familie mit Koilonychie behaftet, unter Wakayama City einwöhnern gefunden, beweits:

1. Die Familie enthält 36 behaftete Mitglieder in 5 Generationen (Abb. 1).

Die 17 Ehen zwischen behafteten und normalen Mitgliedern der Familie mit mehr als zwei Kindern, erzeugten 29 behaftete (16 männlich und 13 weiblich) und 37 normale (22 männlich und 15 weiblich) Kindern. Dieses 37:29 Verhältnis wird im 1:1 Verhältnis (normale und behaftete Mitglieder der Nachkommenschafts) enthalten. Das Geschlecht-Verhältnis der behafteten Mitglieder (16 männlich: 13 weiblich) ist auch von 1:1, ungefäht. Daher, scheint es, dass die Ursache der Unregelmässigkeits ein dominantes autosomatisches Gen ist.

2. Die Penetranz des Gens in dieser Familie scheint sehr stark zu sein.

3. Was den Ausdruck der Unregelmässigkeits anbetrifft, so ist die Holheit der Nägel nicht immer deutlich, insbesondere in den Zehen, während der Ausdruck des Gens in den Fingernägel mehr sichtbar ist.