

Final Pedigrees of Y Chromosome Inheritance

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THE METHOD OF INHERITANCE of hairy ear rims has been a matter of some controversy since Gates (1946) republished an Italian pedigree of this condition which had been reported by Tommasi in 1907. The case of two Italian brothers with hairy ears of the same extreme type was described by Cainer in 1898. These two pedigrees have been fully discussed in a recent monograph on the inheritance of hairy ear rims (Gates and Bhaduri, 1961), based on work done in Africa in 1955 (Gates, 1957) and India in 1959 (Gates, 1960).

That monograph also contained a discussion of the evidence to date for hairy ear rims in its anthropologic setting and included 21 new pedigrees. One of these was extensively and carefully collected by Bhaduri, and contained some 250 individuals in five generations and 30 sibships. The whole of this and other evidence appears to make it conclusive that the gene for hairy ear rims is in the Y-chromosome, as originally concluded from the Tommasi pedigree.

In any holandric pedigree there are two conditions, and two only, to be fulfilled: (1) that all the sons of an affected father will receive the gene and (2) that the daughters of an affected father do not receive or transmit the gene. The only departures from this method of inheritance will be (a) from possible rare cases of crossing-over from Y to X and (b) from cases (probably much less rare) of failure of penetrance. The latter condition may occur even with such strongly dominant genes as that for hexadactyly. Of course, all males having this gene will be hemizygous for it.

In the Bhaduri pedigree, as originally published (Gates and Bhaduri, 1961), there was one exception to rule two above. This "exception" was IV-46 in pedigree 21, who was a son of III-25 (a normal female) and was recorded as having slightly hairy ears, only four or five hairs in each ear. The explanation of this exception appears to be that the mother had received an X-chromosome in which the gene had crossed over from Y to X. Illegitimacy, although highly improbable, is not excluded from the pedigree.

The rule that a father always transmits to all his sons is occasionally broken by a failure of penetrance. Such a failure is found in Gates and Bhaduri, pedigree 11, in which III-3 (age 32) lacks the trait which is present in his three younger brothers as well as in the father. Also, in pedigree 15 the two sons of I-1, aged 27 and 25 years, should have developed the trait. In pedigree 18, II-5 and II-7, aged 35 and 30 years, respectively, should have done likewise, as well as III-2 at the age of 23. These could not be examined personally and it is quite possible that a few hairs not noticed by the proband may have been present in some cases.

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When only three or four long hairs are present in the ear rim, this is evidently to be counted as presence of the trait. Examination of many such cases confirms this view. In hasty or incomplete examination these few inconspicuous hairs may easily be overlooked, thus increasing the apparent frequency of failure of penetrance.

Seven pedigrees of this condition were recorded by Sarkar, Banerjee, Bhat-tacharjee and Stern (1961), who made the statement that "only one pedigree is compatible with, but does not prove the correctness of, the assumption of complete Y-linkage of a fully penetrant gene." It has never before been suggested that lack of penetrance alters the method of inheritance of a gene, and this is presumably not suggested by these authors. It is clear that all seven pedigrees of Sarkar *et al.* go to confirm the Y-inheritance.

In the seven pedigrees of Sarkar *et al.* there is also no case in which the daughter of a man with hypertrichosis pinnae has a son with hairy ears, unless she has married a man with the trait. These pedigrees are, therefore, all in complete accord with Y-chromosome inheritance and occasional failure of penetrance. In pedigree 7 of Sarkar *et al.* there are several apparent cases of failure of penetrance and one of skipping a generation, but Sarkar has informed Gates that the latter has not been certainly ascertained. Some of these cases may have resulted from overlooking a few hairs. Moreover, they are not in accord with any other method of inheritance. Dronamraju (1960) has also contributed a large pedigree of inheritance of this condition in the Y-chromosome. There is therefore no basis for the suggestion that this condition is inherited in different ways in different families. The only possible exception to this rule is in the rare cases (discussed by Gates and Bhaduri, 1961) of possible crossing-over of the hairy ear gene from the Y to the X chromosome. Stern (1957) did not discuss this possibility and wrote about "complete Y linkage," ignoring those rare cases where the natural interpretation is that of crossing-over from Y to X in certain pedigrees and from X to Y in certain other pedigrees of different abnormalities.

Sarkar *et al.* (1961) made the useful suggestion that different degrees of the trait should be recorded. This was done only in a preliminary way in pedigree 21 of Gates and Bhaduri (1961). Sarkar *et al.* proposed a scale of hairiness based upon amount, without emphasis upon location, as follows: 1, very scanty; 2, scanty (photograph of a Bengalee supplied); 3, medium (photograph of a Singhalese supplied); 4, marked (photograph of Goan in Gates, 1957, cited); 5, very marked or bushy (photograph of Tommasi's Italian case supplied).

We have decided to make a scale of degrees of hypertrichosis pinnae beginning with the most extreme (Italian) condition and having a morphologic basis emphasizing the location of hairs. This scale is as follows:

Stage 1. The Italian condition with long, stiff hairs and hair on the back and front as well as the rim of the ear. Photographs of Tommasi's case were given by Sarkar *et al.* (1961, Fig. 1) and by Gates and Bhaduri (1961, Figs. 2, 4, 5)

Stage 2. Long hairs all around the rim. Photograph of a Goan by Gates (1957, Figs. 1, 2).

Stage 3. Long hairs absent from the top (horizontal) portion of the rim.

Stage 4. A zone of hairs in the middle of the vertical rim. Photographs, Sarkar *et al.* (1961, Figs. 2 and 3) and Gates and Bhaduri (1961, Fig. 6).

Stage 5. A tuft of hairs at the base of the rim. Photograph, Gates and Bhaduri (1961, Fig. 1).

Stage 6. A trace (only three or four long hairs). Observations have shown that a father with typical hairy ears may have a son with only a trace. In some cases this may be regarded as a partial failure of penetrance.

It will be seen that the scale of Sarkar *et al.*, based simply on the amount of hair, is impractical, whereas the new scale based on regional distribution of hair can always be used in classifying hairy-ear types. It appears that stages 4 and 5 are clearly distinguishable. As previously pointed out, the tuft of hairs in stage 5 has been regarded by Thurston (1909) as resembling that of the bonnet monkey of South India.

In the six pedigrees recorded in this paper the degree of hairiness of the ears is noted as far as possible by a figure (1 to 6) on the right-hand side of the symbol for each individual. It will be seen that even in the same sibship the variation may be wide, although often it is rather narrow. Only after further observations will it be possible to determine whether the conception of cistrons or sub-genes is applicable here, or whether this is merely non-inherited variation. The conception of multiple sub-genes might even apply, but this seems unlikely.

Concerning the frequency of hairy ear rims, Sarkar *et al.* found 24 cases in 226 adult males (10.6 per cent) from Bengal and Orissa. We found among 103 adult Bengalis, measured in Calcutta, 26 with hairs in the ear rims (25.2 per cent). The higher frequency is probably due to the recognition that even a few long hairs represent the presence of the gene. Of the 26 cases, 11 showed a zone (Stage 4), 13 a trace (Stage 6) and one each a few hairs or a medium number. This mixed nomenclature is due to the fact that the scale of observations was being developed while the measurements were being taken. The results show that our percentage of frequency is over twice as high because half our cases showed only a trace, i.e., a few (3 or 4) long hairs. It may be tentatively concluded that in the Bengal population about 10 per cent of adult males show the marked condition, while this frequency is doubled if the presence of very few hairs is recognized as significant.

Observations were also made of hairs in the meatus of these Bengalis. They were practically all between the ages of 20 and 40 but ten of them (9.7 per cent) had short hairs in the meatus. In seven of these (three Brahmmins, three Kayasthas and one Vaishya) hairs were present in both the rim and the meatus. The Brahmmins with hairy rim numbered 11 (23.9 per cent) and the Kayasthas 7 (25.9 per cent), showing practically no difference in frequency. Observations were also made of body hair.

It may be pointed out that in a recent expedition by the three of us to the Lepchas, Bhutias, Tibetan refugees and Totos, which are all Mongoloid in

HYPERTRICHOSIS PINNAE

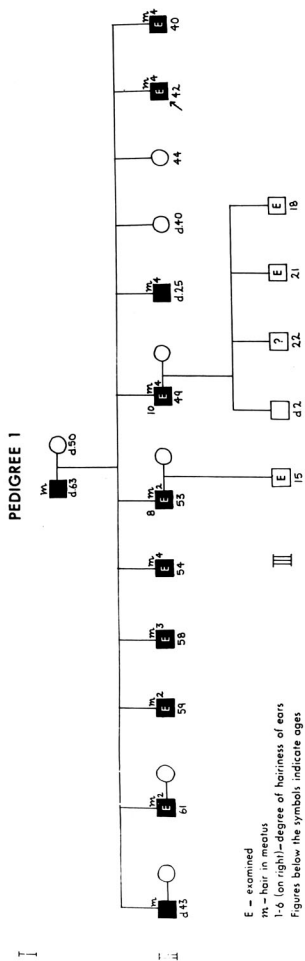


FIG. 1.

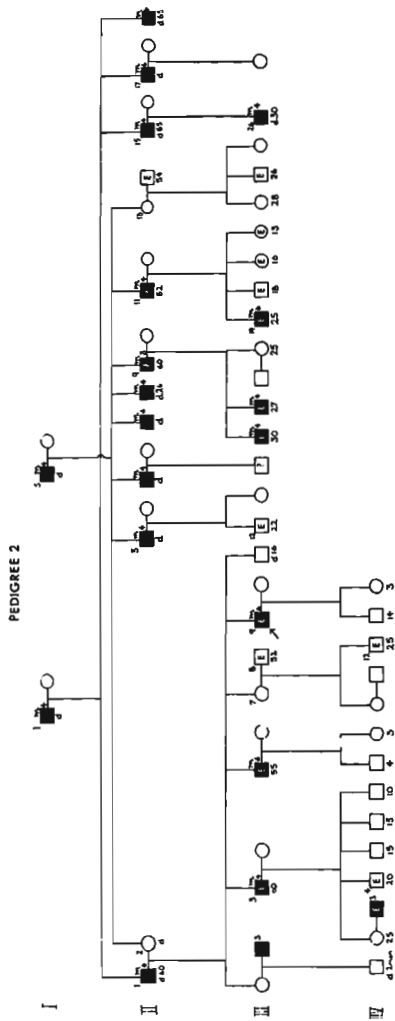


FIG. 2.

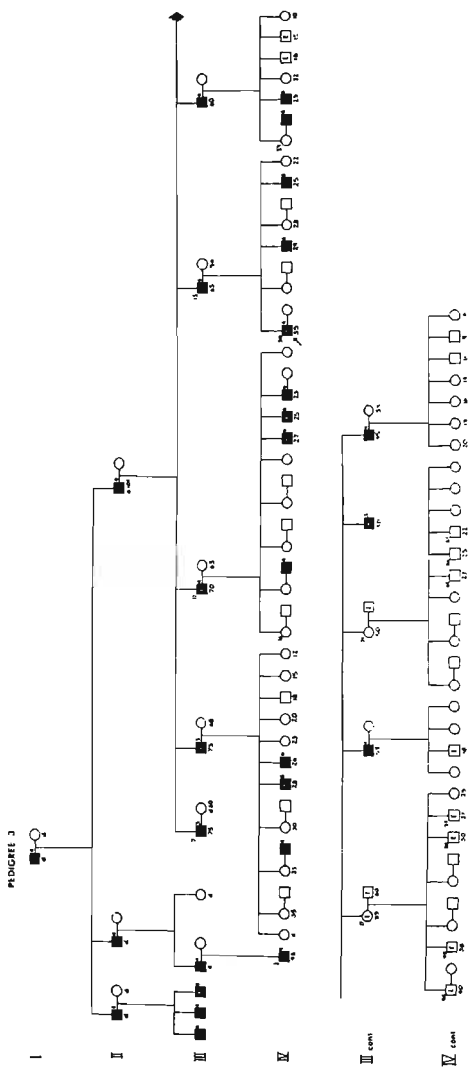


FIG. 3.

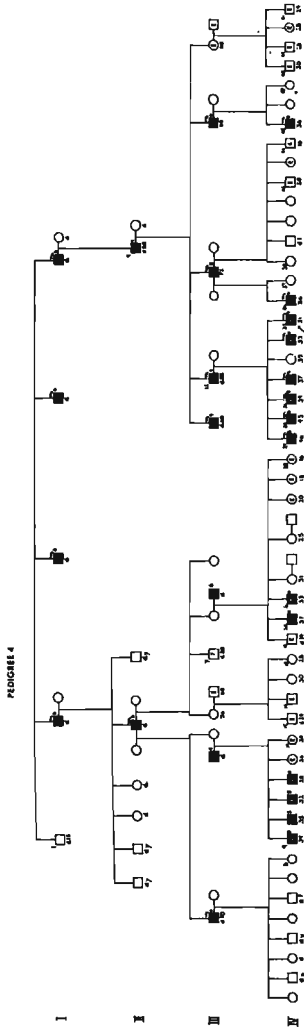


FIG. 4.

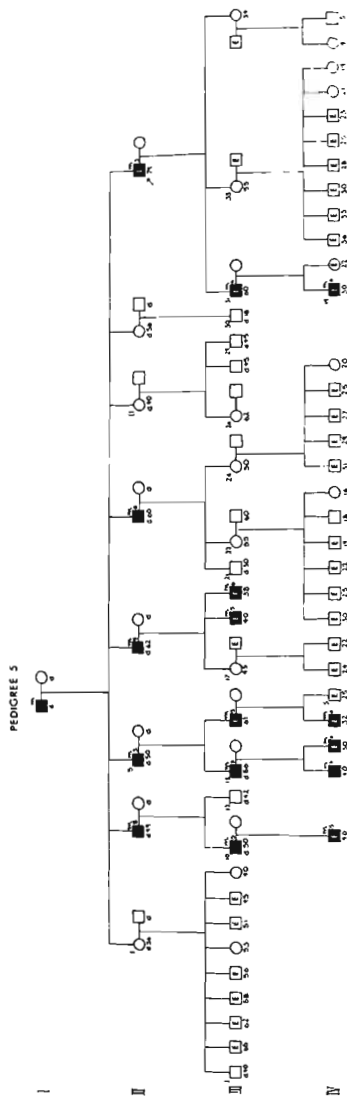


FIG. 5.

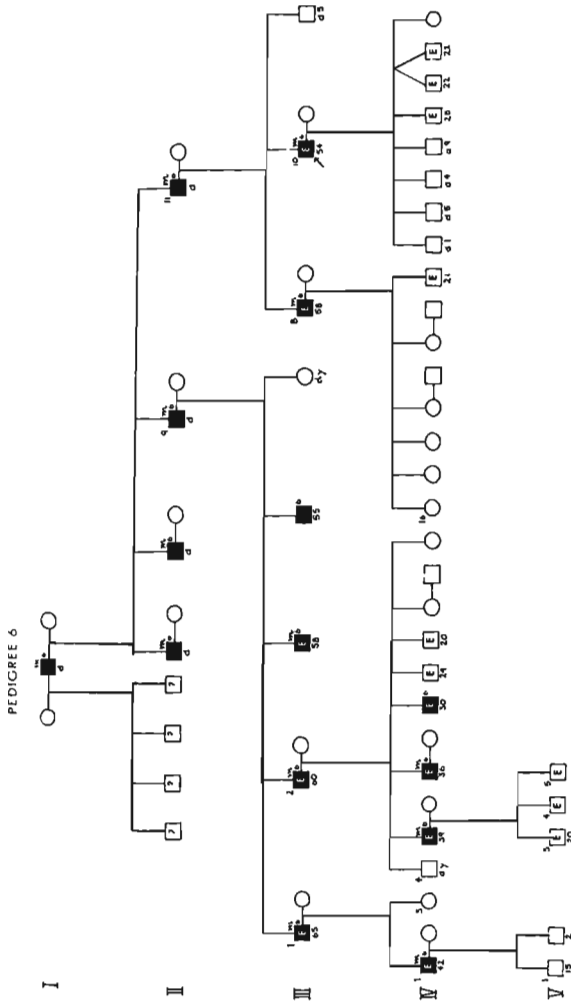


Fig. 6.

varying degrees, no cases of hairy ears were found. Similarly the Khasis in Shillong, Assam, were without this trait. The bodies of these tribes were also relatively hairless, the Tibetans extremely so. It is also not without interest that the late Dr. B. S. Guha, the well-known anthropologist, and his seven brothers as well as their father, who are Bengalis of the Kayastha caste, were all without hairs in the ear rims. Four of them were examined personally.

An Assamese Muslim in Shillong was observed to have a zone of hair in the ear rim and also much hair in the meatus. Both ears were alike, and his father was stated by him to be the same. This was the only case of hairy ears which we observed in Assam. Evidently the hairy ear gene came to India from the West, and not from the East.

PEDIGREES

Pedigree 1 represents an Anglo-Indian family residing at Kharagpur, West Bengal. The father was of mixed descent and must have inherited the gene from an Indian ancestor. The father and all ten sons developed hair in the ear rims, meatus and lower bridge of the nose. This can only be explained by Y chromosome inheritance rather than autosomal, as all ten sons are affected and no female member possesses these traits. The father and all sons also had considerable hair on the chest, back, forearms and calves. The range of variation in the distribution of hair in the ear rim is not marked, being only from stage 2 to stage 4. The children of II-8 and II-10 were examined, but all sons were without hairy ears and under the age of 22, except III-3, whose record is doubtful.

Pedigree 2 represents a family of the Karam caste from Contai subdivision of Midnapore, West Bengal, where Sarkar *et al.* (1961) collected their pedigrees. The proband (III-9) and his two brothers who survived to adult age have hairy ear rim and meatus, but not many body hairs except a few on the calves. The 25 year old son (IV-12) of the proband's sister (III-7) has no hair either on the rim or meatus, as is the case with his father (III-8). The proband's father (II-1) and all of his three brothers are also affected with more or less hair in the rim and meatus of the ear, and their father (I-1) had the same condition. The son (III-26) of II-15 inherited the same degree of hairiness in his ears. The absence of hairy ears in a 20 year old son (IV-4) of the proband's brother (III-3) may be due to its late appearance in this family.

All brothers and the father of the proband's mother (II-2) had not only hairy ears but also some hair on the chest and calf muscles. Body hair is practically absent in the proband and his brothers. The body hair in this part of the pedigree, therefore, seems to be associated with hairy ears and is not transmitted through the female offspring. That the 22 year-old son of II-3 and the 25 year-old son of II-11 has only a trace of it indicates a later expression of hairy ear rather than lack of penetrance, provided the ages have been correctly reported.

Pedigree 3 represents a Bengali family of the Rari Brahman sub-caste, and the proband (IV-30) is one of us (Chakravarti). The proband's father (III-13) and all seven of his brothers have hair in the ear rim. Most of them have it in a middle zone (stage 4) like the proband himself. None of the seven sons of the sisters (III-17 and III-21), with ages ranging from 22 to 40 years, have

hairy ears. This excludes the possibility of its inheritance through the female line, whereas in the male line the penetrance is complete, as is amply demonstrated by the three sibships of generation III and their male ancestors.

It is noteworthy that with the exception of IV-1 and the proband (IV-30), who are the two oldest male members of generation IV, all affected persons in this generation are younger than 30 years and have only the least degree of hairiness in the scale. The proband remembers that his hairy zone had not developed completely until after his 28th year. The small growth of hair in the ear rim in most persons of this generation can be explained by their younger age. III-7, who is 75 years old, stated that some of the hairs in his ear rim were being shed from time to time after he reached the age of 65. The affected members of this pedigree have a wide variation of hairiness on the chest and no hair in the meatus.

Pedigree 4 describes a Bengali Kayastha family of Calcutta. The proband had a tuft of hair in the rim of the ear and considerable hair in the meatus. All five of his brothers had hair on the rim and in the meatus of the ear as well as body hair on the chest, forearm and calf muscles. Hair on the back and shoulders and profuse chest hair was observed in IV-9 and IV-31, whose hairy ear rim is like stage 3 of the scale. All males of earlier generations were said to have had hairy ears, and those of III-12 and II-9 could be seen in their photographs. There is no lack of penetrance in the male line except in I-1, III-7 and IV-19, all of whom died before the age of 30 years and might have had slight hairiness in the ears which could have been easily overlooked. The age of appearance of hairy ear is between 20 and 28 years in this pedigree. IV-15, IV-16, IV-48, IV-49 and IV-51 provide sufficient evidence that the character is never inherited through the mother, while it is always received from the father. The little amount of hair in IV-20 and IV-21 seems to have been clearly inherited from the father, who was reported to have, like them, no meatus hair, a characteristic of their mother's male relatives. The variation in degree of hairiness in ear rims of the father and sons may be the effect of constitutional factors affecting their expression.

Pedigree 5 is from the Tantubai (weavers) caste in Baranagore near Calcutta. The proband, an old engineer retired from the Railway Board and chairman of the local municipality, had plenty of hair in the rim, almost all the way around except the top margin. His body hair on back, chest, forearms and calf muscles was also profuse, and his eyebrows were thick. He remembers that he had full growth of hair in the ear rim at the age of about 50 years. His elder brother, II-5, had similar hair distribution in the ear rim and body. His older brothers and father possessed hairy ear rim and meatus, and all of their sons and son's sons who have reached 25 years of age display hairy ears. III-12 and III-21 may be cases of lack of penetrance, but more probably are cases of incorrect recollection. Aged sons of at least II-1, II-11, III-22, III-24 and III-33, who are all daughters of affected fathers, clearly show that daughters do not transmit the hairy ear gene. The seven sons of IP1 are all devoid of hairy ears at ages ranging from 40 to 65. Similarly, the six sons of III-33 are all without hairy ear rims at ages of 23 to 36 years. The association of body

hair and meatus hair with hairy ear rim and of growth of hairiness of the ear with age, are further demonstrated in this pedigree.

Pedigree 6 is again from a Bengal Kayastha family of Calcutta with hairy ears in four generations. The hairs in ear rim and meatus are both inherited from father to all his sons, but the quantity of hair on the ear rim is rather scarce, and meatus hair is more developed. The youngest person having such hair was 30 years old, and the oldest person not having it was 25 years old, provided the ages were correctly reported. Body hair was practically absent in all cases examined. It appears from this pedigree that the small quantity of hair in the ear rim may be connected with its late appearance and also with the small amount of body hair. The expression of hairy ear in all 14 sons of seven affected fathers strongly supports its holandric nature, although its development is very late.

Taken together, these six pedigrees leave no room to doubt that the hairy ear gene is inherited in the Y chromosome, as was shown by Gates and Bhaduri (1961).

SUMMARY

Six new pedigrees of hypertrichosis pinnae abundantly reinforce the conclusion that the gene is in the Y chromosome. Questions of lack of penetrance and fractionation of the gene are briefly discussed in the text. The pedigrees demonstrate that the daughters of affected males do not transmit the gene to their sons.

A scale is described for scoring degrees of hypertrichosis pinnae that has a morphological basis and emphasizes the location of hairs.

Within families, there is variation within relatively narrow limits in the degree of hairiness of the ears and also in the range of age of onset. An association of hair in the ear rim with hair in the meatus was noted in pedigrees 1, 2, 4, 5 and 6. An association between hairy ears and profuse body hair was observed in pedigrees 1, 2, 4, and 5. In pedigree 3 there was marked variation in the amount of chest hair in individuals with hairy ears. In pedigree 6 body hair was practically absent, hair in the rims was scarce (Stage 6) and late in development, and hair in the meatus was more developed.

It only remains to say that all pedigrees of hairy ears yet published are compatible with a gene in the Y-chromosome with occasional failure of penetrance, and most of them are incompatible with any other form of inheritance. The gene evidently came to India from the West.

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