



# Pachyonychia Congenita Project

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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.

# HEREDITARY MULTIPLE SEBACEOUS CYSTS

F. E. STEPHENS\*

**O**CCASIONAL reports of familial multiple sebaceous cysts (wens) appear in the literature. Some workers have considered the tendency to produce these cysts to be hereditary. Dominant, recessive, and sex-linked inheritance have been reported. Several workers have reported instances where sebaceous cysts have become malignant, or have been associated in some way with malignancy. We have studied recently a kindred at the Laboratory of Human Genetics at the University of Utah (Kindred 77) which shows a high frequency of sebaceous cysts. Our purpose was to discover the type of inheritance involved, and to determine whether or not the condition is related in any way to malignancy.

## Description of the Trait

In Kindred 77 sebaceous cysts were found to occur in multiple form, varying in number from a few to several dozen, and ranging from the size of a pea to that of a walnut. While they were most frequently found near the hair line on the head and neck, they often occurred elsewhere on the body, such as the bend of the arm opposite the elbow. One case was reported of a cyst inside the nose. The age of onset varies greatly from the age of 10 to 40 years. When they first appear, they consist of a soft viscous-like material which later becomes hard and gristle-like. They can be removed and do not reoccur if the membraneous sack surrounding them is also removed.

## History of Kindred

The occurrence of sebaceous cysts in Kindred 77 is shown in the pedigree

chart in Figure 17. Kindred 77 consists of the descendants of I-1, who was born in England in 1807 and later emigrated to Utah. Two of his 10 children, II-7 and II-9, are known to have had sebaceous cysts. This characteristic has been found in five generations of this kindred. While in most cases the trait does not occur in the offspring unless it occurs in at least one of the parents, there are a few exceptions. Four individuals, III-1, III-6, IV-8, and IV-67, apparently do not have the trait, but have transmitted it to some of their children. It is possible that some of these were actually affected and for some reason it was not possible to secure the correct information. So far as is known, no cases of sebaceous cysts in this kindred have become malignant.

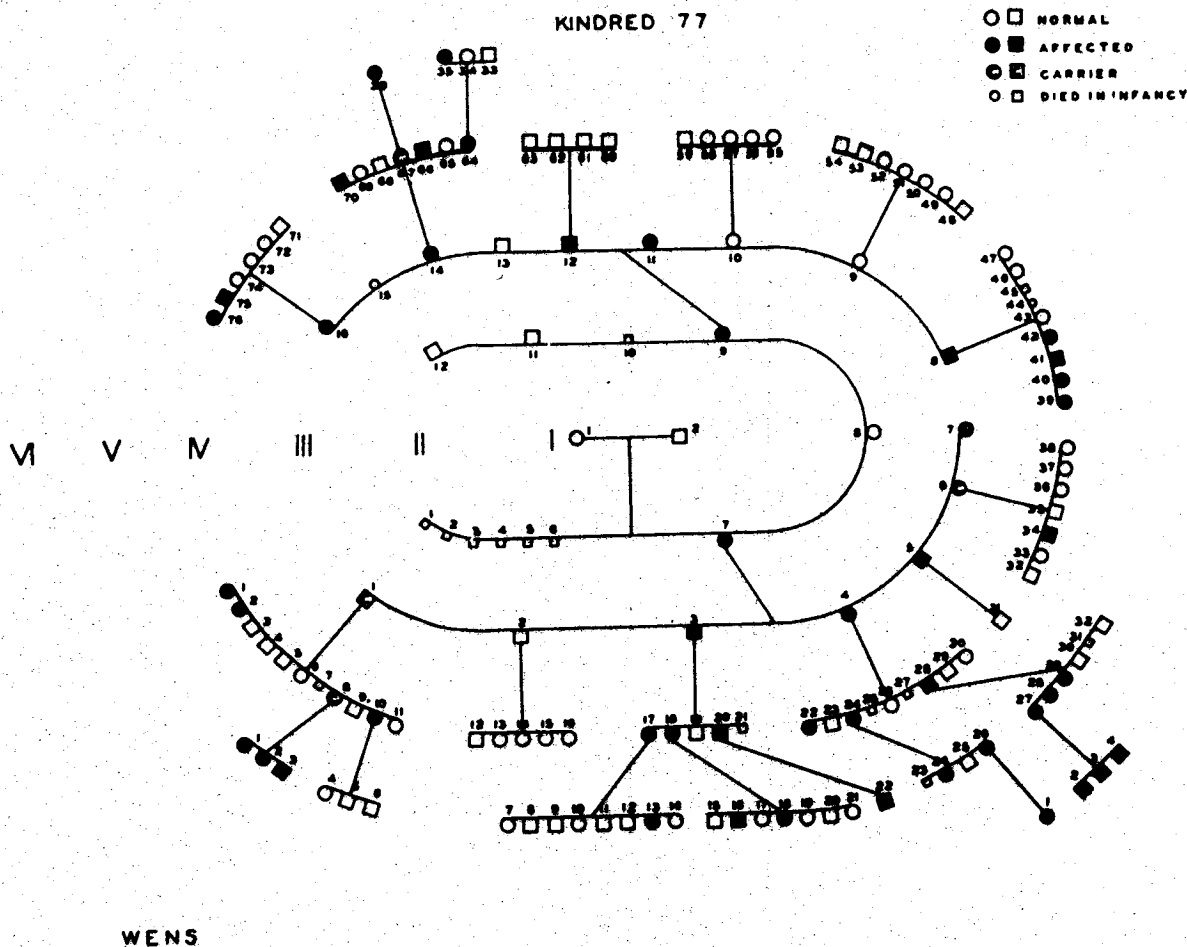
## Literature

Paget (cited by Gates<sup>3</sup>) considered cysts were more commonly inherited than cancer. Paget<sup>8</sup>, Klausner<sup>5</sup>, Prakken<sup>9</sup> and Weber<sup>11</sup> (cited by Ingram<sup>4</sup>) all reported families showing familial multiple sebaceous cysts. Ingram and Oldfield<sup>4</sup> reported a family having five affected individuals in three generations. They concluded that "the factor for multiple sebaceous cysts appears to be inherited in a Mendelian fashion and resembles a dominant more than a recessive."

Monro<sup>7</sup> described a family in which 20 persons had sebaceous cysts. He explained this on a basis of a single Mendelian dominant, subject to modifications. He also concluded that there might be some relationship between sebaceous cysts and malignancy. Levit<sup>6</sup> reported a kindred showing 10 affected cases in three generations. He concludes that the trait is due to recessive inheritance. Sedgwick<sup>10</sup> (cited by Gates<sup>3</sup>) reported on two families showing sebaceous cysts. He considers the trait to be sex-linked.

Caylor<sup>1</sup> reported that "approximately 3.44 percent of the sebaceous cysts in this (his)

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### PEDIGREE OF KINDRED 77

Figure 17

The occurrence of sebaceous cysts has been found in five generations of this Kindred. It is concluded that it is inherited as a simple Mendelian dominant.

series eventually became malignant" and Gardner<sup>2</sup> reported sebaceous cysts to be associated with multiple polyposis in a syndrome, but found no cases where the cysts became malignant. Welch<sup>12</sup> reported a case where two different sebaceous cysts in the same individual became malignant.

#### Inheritance

While the inheritance of sebaceous cysts has been reported as being dominant, recessive, and sex-linked, most workers seem to agree on some type of dominant inheritance. It will be seen from the pedigree history of Kindred 77 that with four exceptions, the trait does not occur in a child unless it has also occurred in one of the parents. It also occurs approximately equally often in males and females. If we include the four exceptions mentioned above as carriers of the defective gene and classify the offspring of affected individuals into affected and normal, we get 47 affected to 56 normal which is a good approach to a 1:1 ratio which would be expected if we were dealing with a simple dominant trait. From the

data which is available it seems that multiple sebaceous cysts in Kindred 77 are inherited as a simple Mendelian dominant with possible incomplete penetrance.

#### Summary

Forty-eight cases of sebaceous cysts or wens were found in four generations of Kindred 77. The trait occurs approximately equally often in males and females. With four exceptions it never occurred in a child unless it also occurred in at least one of the parents. It is concluded that it is inherited as a simple Mendelian dominant with possibly incomplete penetrance. There seems to be no relationship between sebaceous cysts and cancer in this kindred.

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### GENETICS OF COAT COLOR IN HORSES

A Brazilian geneticist has recently published a comprehensive and critical review of the literature on the genetics of coat color in horses,\* the most complete survey of the subject which has been made since the important publication by Odriozola in 1951. Where the views of Odriozola are at variance with those

of North American geneticists, the author shows preference in general for the latter, but with minor deviations of his own. This publication will be of much interest to readers of the *Journal of Heredity*, where the more important studies in this field have been published or reviewed.

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\*Genetica da Pelagem do Cavalo. Professor Raul Briquet, Junior. Instituto de Zootecnia, Rio de Janeiro, Brazil. 115 pp., December, 1959.