

INHERITANCE OF HUMAN FACIAL FEATURES

A Pedigree Study Involving Length of Face, Prominent Ears and Chin Cleft

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MODERN studies of irregularities in human facial features and attempts to correct disfiguring departures from the norm had their origin shortly after the turn of the century. It is an interesting coincidence that just at this same time Mendel's laws were rediscovered and corroborated by studies of a great many plant and animal forms including man himself. At just the same time that the groundwork of modern genetics was being laid, the foundations of the new science of "dental orthopedia" prepared the way for two textbooks^{1, 2} for what today is one of the important branches of dental practice. Most variations in the teeth and jaws were then supposed to be the direct result of environmental influences and many of these influences were thought to lead to profound changes in external features.

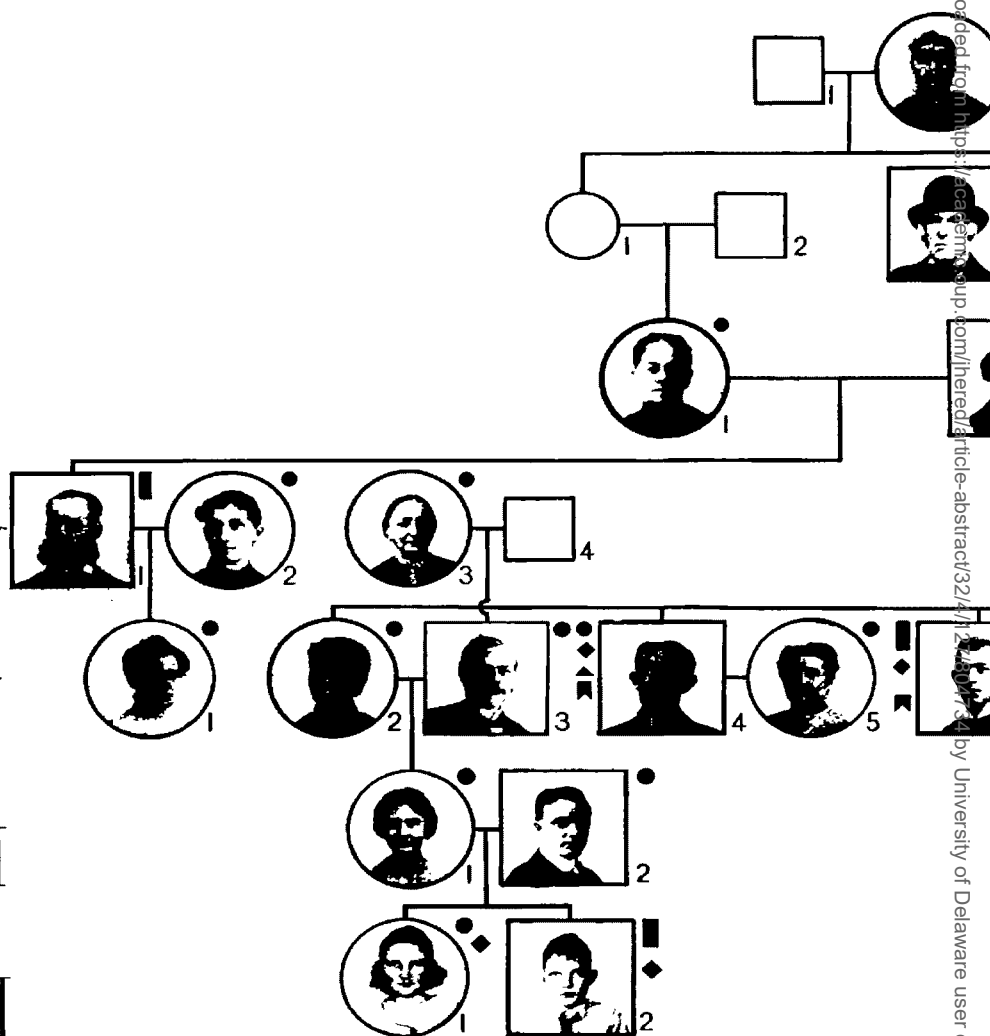
The possibility that such variations could be in any way inherited, seems never to have greatly impressed the early students of orthodontia although inheritance of many other characters was accepted at the time as obvious fact. The striking facility with which many abnormalities could be corrected by environmental manipulation led to the concentration of all efforts in this direction. Curiously enough the inheritance of some variations such as toothlessness, teeth without enamel, abnormally large incisors or canines, or increases or reductions in the number of teeth, has been studied almost exclusively as a matter of scientific interest rather than because of any ultimate importance to the dental profession. They are variations which attract attention because of their conspicuousness and their obvious tendency to run in families. Neither the dentists nor

the pure scientists paid the slightest attention to the inheritance of less-marked features of the lower face and jaws.

The fact that certain characteristics of the teeth and gums were inherited in a simple Mendelian manner, with a regularity which would make possible the prediction of the recurrence of the character in subsequent generations would seem to be a matter of vital importance to dentists. This has not been the case because these disfiguring or actually pathological defects are not the variations which attract most of the professional attention of orthodontists. Relatively minor defects are, on account of their greater frequency, the major preoccupation of the dental surgeon. If the disharmonies of the underlying osseous tissue of the jaw and the conformation of the overlying skull, which long have been most puzzling, should likewise prove to have an hereditary as well as an environmental background, an entirely new approach to the correction of such defects would be necessary. Since rational treatment of any abnormality necessitates a correct diagnosis and the determination and removal of underlying causes, the ability to distinguish developmental influences of an inherent nature, if they do actually exist, would not only be most advantageous, it would be imperative that such knowledge be applied in diagnosis and treatment.

While speculating as to the probable inheritance of such variations the attention of one of the authors (M. R. L.) was attracted to the striking contrast in facial conformation manifest in a brother and sister then under treatment. Through the interest and cooperation of their living relatives it has been possible to assemble a pictorial pedigree (Figure

I
II
III
IV
V
VI
VII



A SEVEN-GENERATION

A collection of photographs of a family showing several clearly heritable characters, such as shape of face, cleft chin, length of nose, and position of ears. The method of inheritance of such characters it does constitute a valuable aid in the study of heredity.

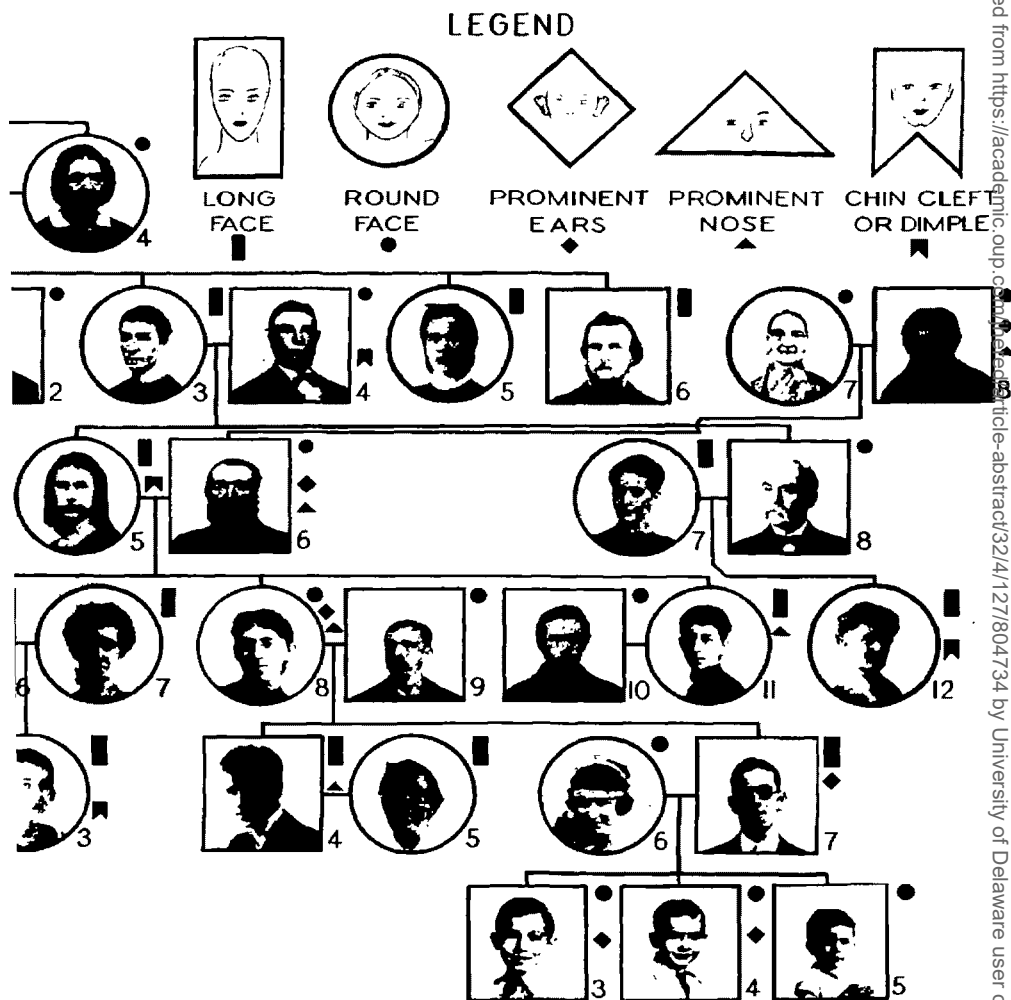


PHOTO-PEDIGREE

and alternative characters gives a unique opportunity to observe the inheritance of these characters. While this single pedigree can hardly be taken as finally determining the value of these characters, it adds a new piece to our knowledge.

10) which enables us to trace several not uncommon facial features for seven generations to ancestors settling in Rhode Island at about the time of Roger Williams. These were, prominent ears, a prominent nose, a cleft chin and an unusually long face. The first three of these features are of more biological than professional interest, but length of face is a character which is a definite component of the previously mentioned problem of conformation. The manner in which this character not only appears to run in the family but the way in which it segregates in a more or less regular fashion is worthy of more than passing notice. If the techniques and standards of measurement which the orthodontists have developed could be applied to more cases of this type it would seem not impossible that much could be learned as to the actual manner of inheritance of such traits. The theoretical and the very considerable practical utility of such information would be of definite value. Unfortunately it has not been possible to obtain actual measurements of many of the individuals of this family but the pictorial results are presented with the hope of stimulating interest in this direction. The only individuals included in the pedigree are forty-two members of the family of whom satisfactory photographs or daguerreotypes were obtainable.

Shape of Face

Contrasting types in facial shape were first observed in the two children, sister and brother VII-1 and VII-2 (Figure 10), the former possessing a broad or round face whereas the latter has a long face. The origin of the long face character can be traced back seven generations through the great-great-grandmother IV-5, to the original maternal ancestor I-2 from which are descended thirteen long faced individuals III-2, 3, 5 and 6; IV-1, and 5; V-6, 6, 11, and 12; VI-4 and 7; and the propositus VII-2. Although the classification of these individuals is arbitrary and the characters in some of the pictures have been obscured by the manner of hairdress or other facial adornment of the times, the classifi-

cation of each individual has been substantiated as being the type recognized by other members of the family.

Several interesting facts of genetic significance are evident:

1. Long face occurs rather *infrequently* and *from round-faced parents*.

2. It *skips generations* as in the case of individual IV-1 who could have obtained the trait from either his great-grandmother I-2 or his grandfather II-3 by way of his parents who were first cousins. Individual VII-2 likewise obtained it from his great-grandmother IV-5.

3. *Mating of round-faced hybrid carriers of long face* such as II-4 and IV-8 *to long-faced individuals* II-3 and IV-6 respectively *produce both long- and round-faced progeny*. They are not in equal numbers as might be expected if the trait were transmitted in a simple mendelian manner, but the number of progeny is small.

4. *Mating of known heterozygotes produces both kinds of progeny* although the numbers are too small to form a significant ratio.

5. Only one mating of *long face by long face* is at hand. Individuals V-6 and V-7 have one daughter VI-3 who is long faced. Additional matings of this type are of special interest in order to determine whether or not they breed true, as they should if long face is a simple recessive.

Each of these listed observations is a characteristic frequently found in a pedigree of a trait which segregates in a typical and simple mendelian manner. It would be folly, however, to draw the definite conclusion from these meager data that the manner of inheritance of this trait is of this simple nature, especially in view of the craniometric studies of Cameron³ and also studies by Castle⁴ of adult body size in the rabbit and by Green,⁵ Castle, Gates and Reed⁶ on the mouse. In the latter cases the inheritance of the character is believed to be the result of numerous hereditary influences acting in a cumulative manner. Cameron found that with respect to both facial width and height, the races of man fall

into several distinct groups but that there is no hard and fast zone of demarcation between them. Both measurements display wide ranges of variation within races which in some cases are overlapping. It is quite probable that if all of the individuals in the above pedigree could have been subjected to quantitative measurement there would be a similar overlapping of types. In fact there are several individuals which have been arbitrarily classified as round but which if accurately measured might well be classified as intermediate in facial length, and would constitute evidence of the continuous nature of this variation.

The most complete study of this sort of variation is to be found in the recently published researches of the late Doctor C. R. Stockard and his co-workers.⁷ Comparison of nine different indices of the skull and jaws obtained from several long snouted and several short snouted breeds of dogs and of the same indices obtained from the F_1 and F_2 generations of crosses between them reveals a dominance of the proportions of the long snouted dogs in the F_1 and a tendency for the segregation in the F_2 to cluster around the types of the parental races. However, the variation is clearly continuous, rather than discontinuous as is to be expected from characters determined by a single genetic difference. Furthermore, extreme parental types were not obtained in most cases, which is another characteristic of variations determined by numerous hereditary factors.

In the light of such experimental data it is too much to expect that facial conformation in man will actually lend itself to a simple mendelian interpretation. Nevertheless the repeated appearance of the long face in successive generations regardless of the phenotypes of the marriage partners concerned is difficult to conceive of as resulting either from environmental forces alone or as being due to the fortuitous recombination of any great number of hereditary influences. Since no two individuals develop in absolutely the same environments, phenotypic variation is always present and this may account for the observed small de-

partures from the strict long or round faced types. It is also quite possible that certain families, even certain racial groups are homozygous for several genes determining face shape. Thus we may get a simpler pattern of inheritance in some families than in others.

It is recognized that the pedigree method is not the most satisfactory method of complete analysis in dealing with a complex variation depending on a complete genetic constitution. Particularly is this true in man who is notoriously hybrid with respect to so many characteristics and in whom experimental control is out of the question. The results obtained by Stockard and his co-workers with variations so closely resembling the disharmonies found in man suggest a way that the problem of evaluating the hereditary influences in jaw development may be attacked. The problem is by no means hopeless if the recognized inadequacies of human material can be supplemented by experiments on laboratory animals in which it is possible to make controlled matings of genetically uniform material.

Chin Cleft or Dimple

A second character, the presence of a striking dimple or cleft in the chin, had its beginning in this family in individual III-4 where it was so well developed as to be a nuisance in shaving. It was consequently obscured in later life by the beard shown in the photograph. The fact that the character was directly transmitted to IV-5 and through her to V-4 and V-6, and to still another generation in VI-3, suggests dominance of the character. However, the cleft is also transmitted to V-12 from III-4 by way of IV-8. Neither IV-8 nor his wife (IV-7) possessed the character. Thus, the trait has skipped a generation making its inheritance as a recessive the more probable although neither interpretation here has adequate proof.

Prominent Ears and Nose

Attention should also be called to these two traits which quite obviously are running in the family. Due to the style of

hairdress the identification of the first character is in some cases doubtful and therefore interpretation of the manner of inheritance is not reliable. Similarly a prominence of nose has been recognized as a family trait although photographically it is not distinctly portrayed and therefore correct evaluation is impossible. It is, however, interesting to note that all of these latter characters are combined in individual V-4 showing how these facial features may unite to produce a very definite and distinct facial type.

Conclusion

A pictorial pedigree of seven generations and forty-two individuals manifests a segregation of the characters long versus round face, prominent nose and ears and cleft in the chin, in a manner which is in harmony with simple mendelian interpretations if it is assumed that minor

variations are phenotypic in their origin. Attention is called to the significance of such variation in the practical treatment of facial conformation by the orthodontist.

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FACTS-OF-LIFE FOR THE ORCHARD

Modern Fruit Production, by Gourley and Howlett, of Ohio State University, is really an excellent new text book of horticulture. The material presented is generally well digested and up-to-date. The authors have been chiefly interested in physiological work with the apple, so that the book is most satisfactory for students (and fruit growers) in regions of the United States where apple growing is important. The breeder will find the chapters on "the plant and its parts," "factors affecting flower formation," "fruit setting" and "origin and improvement of fruits" very helpful.

The chapter on fruit setting is in general excellent.

Those interested in small fruits may find the material somewhat less satisfactory than those interested in tree fruits. Thus in the chapter on the "fruit plant and its parts" the flower bud differentiation of the cranberry is not mentioned. A number of minor errors to be corrected in a later edition are noted. For students, and research workers in pomology as well, the authors have written a very fine and readable book.

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*The Macmillan Company, New York. 1941.