GENETICAL SOCIETY OF GREAT BRITAIN

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GENETICS OF PSEUDOXANTHOMA ELASTICUM

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Pseudoxanthoma elasticum (P.X.E.) is a rare inherited disease of elastic tissue which normally behaves as a recessive. Examination of case reports showed that of 22 families of two or more sibs, in which complete sibships were recorded, 21 show *either* males *or* females affected, but not both. This strongly suggests the possibility of partial sex linkage but some doubts are thrown upon this theory by cases resulting from consanguineous matings. These will be discussed.

Relatives of four families in which P.X.E. has occurred have been examined to see whether any manifestation can be detected in the heterozygote. In all four families some first-degree relatives have shown abnormally visible major choroidal vessels.

FEATURES OF THE "ADIPOSE" MOUSE

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Two genetically different forms of obesity associated with infertility and hyperglycæmia are now known in the mouse. Much attention has been devoted to the "obese" condition, but "adipose", only recently discovered by D. S. Falconer, has not yet been completely described. The reproductive system of 12-week-old animals with this syndrome has therefore been systematically examined. Typically, though not invariably, the ovaries lack luteal tissue, the uterus is undeveloped and vaginal cycles are always weak and irregular. On the other hand, structurally mature Graaffian follicles are produced and little histological pathology is evident in the male. The testes are small, but apparently normal spermatogenesis occurs and the accessory reproductive tract is indistinguishable from the wild type. The penis and scrotum, however, are poorly developed and males like females are completely sterile. The reproductive system is very similar to that in "obese" mice and indicative of gonadotrophin deficiency, but at least at 12 weeks of age is not as abnormal and it is of consequence that the obesity is also less.

It has been maintained that the diabetic condition of the "obese" mouse is dependent upon a hypersecretion of pituitary diabetogenic hormone. If, as seems likely, the same situation prevails in "adipose", it is anomalous that tail growth and the "tibia test" indicate no increased output of growth hormone, since it is widely believed that growth hormone and the diabetogenic factor are the same substance.

EVIDENCE FOR THE EFFECTIVENESS OF DISRUPTIVE SELECTION IN NATURE

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The African swallowtail butterfly, *Papilio dardanus*, is markedly polymorphic in the female and many of the forms are mimetic. The males are everywhere

monomorphic and tailed, whereas the majority of the female forms are tailless like their models. However, in race *meriones* from Madagascar the female is non-mimetic and is always male-like and tailed. In race *antinorii* from Abyssinia, about 80 per cent. of the females are also non-mimetic, male-like and tailed, but mimics do occur and are always tailed.

Measurement of wild material and race crosses carried out in this country indicate :

1. That taillessness is semi-dominant to tailed, and that the locus controlling the character is independent of that determining wing pattern.

2. That using a tailless race the dominance in females is more complete in race crosses with *antinorii* than with *meriones*. Within race *antinorii*, particularly in the mimetic forms, the tails are shorter than in males of all races and the females of *meriones*.

3. The difference between the tail length in the *antinorii* females and those of *meriones*, coupled with the differences between the male-like and mimetic forms, suggest that there is disruptive selection tending to reduce the tail length in the mimetic but not in the non-mimetic forms. This view is strengthened by the fact that the variance of tail length is greater in *antinorii* than in *meriones* as would be anticipated by analogy with Thoday's results in *Drosophila*. Moreover, in the tailed individuals of the F_2 of an *antinorii* to a tailless stock the variance tends to increase and the difference in the mean tail length between the forms to vanish.

The findings generally suggest that in *antinorii* selection is operating towards taillessness (*i.e.* more perfect mimicry) in the mimics in the absence of the major gene for taillessness.

THE LEWIS ANTIGENS IN SALIVA

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Studies have been made of the Lewis antigen content of saliva. No difficulty has been found in scoring specimens from non-secretors for the presence or absence of Le^a substance if they have been stored at −20° C. for three months or longer. With secretor saliva, however, anti-Le^a sera differ from each other in the extent to which they are inhibited ; by repeated testing with several antisera it has been possible to get a clear separation of specimens which contain Lewis antigens from those which do not inhibit anti-Le^a or anti-Le^b.

In 3·6 per cent. of 861 normal people no Lewis antigens have been detected in the saliva, and there are no significant differences between the frequencies in males and females, secretors and non-secretors, nor between those of the different ABO blood groups. Family studies have given results compatible with the hypothesis that the presence of Lewis antigens in saliva is inherited as a dominant character. The genetical interpretation is complicated by the fact that amongst those scored as having no Lewis antigens are a number of individuals, both secretors and non-secretors, whose fresh saliva inhibits anti-Le^a. The titre of the inhibiting substance gradually drops during storage at −20° C. until after 1-3 months there is no inhibition, unlike the titre of the usual Le^a substance which at −20° C. remains constant for at least a year.

An anti-Le^a serum is described which is not inhibited by non-secretor salivas of high Le^a titre nor by secretor salivas which do not contain Le^a. It is inhibited only by secretor salivas which contain Le^a substance, further evidence that the Le^a substance in the saliva of non-secretors is antigenically different from the Le^a substance in the saliva of secretors.

GENETIC STUDIES IN DUODENAL ULCER

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Recent studies have brought to light two specific genetic factors which seem to contribute to the inherited tendency to duodenal ulcer. These two genetic factors are, firstly, blood group O, and secondly, salivary ABH non-secretion, both of which have been shown to be associated with the disease in population surveys.

The present position of studies on these two associations will be reviewed.

PROTECTIVE FACTORS IN ERYTHROBLASTOSIS

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Only one in twenty Rhesus negative women carrying a Rhesus positive fœtus becomes sensitised, and it is therefore probable that protective mechanisms exist. Such mechanisms are known to include ABO incompatibility and the placental barrier. Further evidence in support of ABO incompatibility is presented, together with experimental observations on the passage of fœtal red cells across the placenta.

HUMAN INTERSEXES

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Sex determination is dependent upon genetic, gonadal and genital factors. Our knowledge of anomalous chromosomal patterns, atypical gonadal structure and ambiguous genitalia has greatly advanced in the past decade. However, many problems of intersexuality remain unsolved.

HEREDITARY MODIFICATIONS OF POXYVIRUSES

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Progress in the study of recombination among animal viruses has been hampered by the lack of suitable selective markers. The authors have had some success in demonstrating heritable characters of viruses of the pox group which may facilitate the study of recombination between some of these viruses.