GENETICAL SOCIETY OF GREAT BRITAIN

ABSTRACTS of Papers read at the HUNDRED AND FORTIETH MEETING of the Society held on 9th and 10th NOVEMBER 1962, at the MIDDLESEX HOSPITAL MEDICAL SCHOOL, LONDON, W.1.

AN AUTOSOMAL RING CHROMOSOME IN A HUMAN FEMALE WITH CONGENITAL MALFORMATIONS

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Although numerous structural aberrations (predominantly translocations) have been described in the human karyotype, very few examples of ring chromosomes have been encountered. The present report describes a mentally retarded female infant with cleft palate, cranial deformity and congenital dislocation of the hip, in whom a high proportion of cells derived from both blood and skin contained a very small ring chromosome in place of a no. 18 (Denver). This ring chromosome, which presumably originated after an appreciable deletion had occurred in a normal no. 18, showed a high degree of stability—a feature probably occasioned by its small size.

PROBABLE LINKAGE BETWEEN ONE LOCUS FOR CONGENITAL ZONULAR CATARACT AND THE *DUFFY* BLOOD GROUP LOCUS IN MAN

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A large family described by Nettleship (1909) which manifests one type of congenital zonular cataract has been studied in a search for linkage. There is a strong indication of close linkage with the *Duffy* blood group locus : the final probability ratios, calculated for the whole pedigree by an electronic computer, are somewhat higher than those which would arise from a simple count of no recombinants out of thirteen opportunities. Other relevant linkage data will be briefly discussed.

STRONG HISTOCOMPATIBILITY EFFECTS ASSOCIATED WITH THE B BLOOD GROUP SYSTEM OF CHICKENS

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Several recent lines of evidence lead to the conclusion that the B blood group system of chickens is also associated with strong histocompatibility effects. It would thus appear that in this case the genes cause the production of the same or similar antigens on the tissue cells as on the erythrocytes.

In the present brief series of experiments a single gene difference of the B system

occurring in an F_1 of two related inbred lines of White Leghorns was shown to influence rapid skin graft rejection in adults (first set rejection 5-6 days ; second set 3-4 days), and to produce powerful graft-versus-host reactions in embryos (focal counts on the chorio-allantoic membrane averaging 140). These effects were superimposed upon, but clearly distinct from, the other weaker histocompatibility differentials segregating (first set rejection times 12-1.30+days ; CAM focal counts 0-36).

There was, however, a tentative indication of the existence of segregation of another strong histocompatability locus : *B* may thus not be unique in its strength.

TRANSFERRIN POLYMORPHISM IN WELSH MOUNTAIN SHEEP

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The inheritance of the following three biochemical polymorphisms in the blood of Welsh Mountain sheep has been studied : serum transferrin type, hæmoglobin type and potassium concentration in whole blood. Transferrins differ from the other two systems in showing marked diversity. Whereas there are two electrophoretically different hæmoglobin types giving rise to three genotypes : *HbAA*, *Ab* and *BB* and two blood potassium levels : high potassium (HK) and low potassium (LK), six electrophoretically distinct transferrin bands with all 21 possible combinations have been identified. The inheritance of these genotypes was confirmed in 282 matings. Starch-gel electrophoresis using a modified Poulik's Tris discontinuous buffer system was used.

Gene frequencies in the six allele system in order of decreasing electrophoretic mobility were : Tf_A 0·261 ; Tf_B 0·239 ; Tf_C 0·190 ; Tf_D 0·241 ; Tf_E 0·042 ; Tf_F 0·026.

1030 animals from four age groups showed a marked consistency in the frequency of the different genotypes and a significant departure from Hardy-Weinburg equilibrium ($\chi^2_{(20)} = 40{\cdot}95$; $P<0{\cdot}005$), principally due to a marked excess of " CC " homozygous animals and a shortage of " C+ " heterozygotes.

The results are discussed in terms of (i) selective fertilisation ; (ii) maternal-fœtal incompatibility ; (iii) accuracy of biochemical typing.

FITNESS AND ENVIRONMENTAL VARIATION IN POPULATIONS OF *DROSOPHILA PSEUDOOBSCURA*

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Populations of common origin and polymorphic for two gene arrangements of the third chromosome have been kept for long periods under conditions either of diurnal temperature constancy (K) or controlled thermal oscillation (V). Tests of these populations have shown that (*a*) the Darwinian fitness (viability) of V populations is greater than that of K populations ; (*b*) the genetic variance of V is greater than that of K populations ; (*c*) the chromosomal polymorphism is not balanced under these conditions.

The evolutionary significance of these and related findings will be discussed.

EVOLUTION IN CLOSELY ADJACENT PLANT POPULATIONS

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Natural populations of *Agrostis* sp. only a few feet apart may differ considerably from each other when their habitats are contrasting. Natural selection must be able to maintain such differences despite the occurrence of gene flow between such populations. A comparison of adult populations with populations raised from their naturally produced seed confirms this, and demonstrates that selection is acting in both a directional and a stabilising manner.

Other factors operating in such situations will be discussed.

AN ABNORMAL ACROCENTRIC CHROMOSOME ASSOCIATED WITH MONGOLISM

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A case of mongolism with clinically mild manifestations has been studied.

Cytological studies show a chromosome number of 47 including an abnormal small chromosome. This abnormal chromosome resembles the Philadelphia chromosome associated with chronic myeloid leukæmia.

Possible mechanisms for the production of this abnormal chromosome are discussed.

THE EFFECTS OF X-RAYS ON CROSSING-OVER IN *DROSOPHILA MELANOGASTER*

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The effect of X-rays in doses from 1000 to 8000 rads on crossing-over between the X-chromosomes of the female has been studied, using the markers *sc*, *ec*, *cv*, *ct*, *v*, *g*, *f* and *car*.

X-rays produced little or no effect on crossing-over in eggs laid during the first five days after treatment but produced a strong depression of crossing-over from day six onwards, which was maximal on days seven and eight.

Analysis of the individual segments revealed that the most sensitive regions to this depression were those furthest from the centromere. The segments nearest to the centromere actually showed a significant increase in crossing-over with a *maximum* effect at 4000 rads.

MATERNAL INHERITANCE OF A STERNOPLEURAL CHAETA NUMBER DIFFERENCE IN *DROSOPHILA MELANOGASTER*

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In testing heritability in a line of *D. melanogaster* that had been established under stabilising selection, a regression coefficient offspring mean on midparent of approximately 0·7 was obtained. The test had been set up with assortative mating, using as far as possible equal numbers of cultures from each class of chaeta-number found in the line.

So high a regression suggested a maternal component in the inheritance of the character. The progeny of the extreme high and low cultures of the heritability test were set up further with the four classes of mating H×H, H×L, L×H, and L×L and gave offspring with means characteristic of their mothers. The progeny of these four classes of cross were further tested with the eight matings possible between the four kinds of female and the two extreme kinds of male.

The results demonstrate no significant effect of fathers on phenotype of offspring, but a highly significant effect of both maternal grandmothers and maternal grandfathers.

The difference in chaeta number is large (over 3·5 chaetæ per fly). It is interesting to find that so big a chaeta number difference can be determined entirely by the mother's nuclear genotype.

THE *HAIRLESS* MUTANT OF *DROSOPHILA MELANOGASTER*: THE DEVELOPMENTAL LIMITATION OF ITS EFFECT UPON MACROCHAETÆ

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Hairless, a dominant visible mutant, with recessive lethality, acts mainly by removing particular macrochaetæ. At the sites of removal, vestigial structures are sporadically present. Most similar mutants differ in so far as either all, or else none, of the affected sites bear vestiges.

Environmental and genetic differences between flies bearing the mutant gene reveal an unexpected regularity in the pattern of distribution of vestiges. The morphological sites may be ranked in order of relative proportion of vestiges at affected sites. The series produced remains constant despite differences in the genetic background, temperature of development and mutational origin of the *Hairless* allele used, although the absolute probabilities of effect at each site may vary over a wide range.

The ubiquitous series probably results from some regularity in the developmental system of *Drosophila*. Further evidence indicates an ordered asynchrony of bristle development as the source of such regularity. This being so, overall changes in manifestation of the mutant can be considered as reflecting primarily changes in the time-course of concentration of normal gene product.

SOME MATHEMATICAL STUDIES ON THE BEHAVIOUR OF METAGONS IN MATE-KILLER (MU) PARAMECIA

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Gibson & Beale (*Genet. Res.*, *3*, 24-50, 1962) have suggested that the mu particles of certain mate-killer paramecia depend on the presence of particulate elements—metagons—which are produced by the *M* genes. On loss of the latter, gradual dilution out of the metagons causes the delay of 8-15 fissions before descendant cells without mu particles appear.

The simple dilution hypothesis does not give a good fit to the available data on the proportion of cells lacking mu particles which are found at successive fissions, and various modifications of this simple hypothesis will be discussed. These include

occasional division of the metagons, initial clumping of the metagons in groups, gradual decay or destruction of metagons, and unequal distribution of the metagons to the two daughter cells at fission. Only the first and last of these four hypotheses give a good agreement with the observed data. Mathematical solutions of the first three of these cases can be derived from simple modifications of the generating function giving the frequency distribution for successive generations in the simple dilution case.

SOME ELECTRONIC COMPUTER PROGRAMMES OF GENETIC INTEREST

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A non-technical account will be given of a series of electronic computer programmes to deal with the most common and laborious computations arising in genetic research. They are of three types :—

1. Computational routines, which can be used repeatedly with fresh bodies of data.
2. Tables of special functions and precalculated sets of values, to facilitate, or even make unnecessary, computations on desk calculators.
3. Tables which will be of use in the design of experiments and the interpretation of the results obtained.

Priority has been given to elimination of the heavy labour of computing heritabilities and genetic correlations from animal breeding data, and the programmes written and projected will be described from the user's point of view.

PLASTID ABNORMALITIES INDUCED BY 2-THIOURACIL

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The pyrimidine analogue, 2-thiouracil, produces two classes of abnormality in the chloroplasts of hemp, a short-term one confined to tissues developing during treatment and probably attributable to a blockage of protein synthesis, and a sporadic long-term one propagated through particular cell lineages in otherwise normal tissues. The possibility that the latter type of anomaly results from the induction of mutations in a " plastogene " system will be discussed.

THE EFFECTS OF DIETARY DEFICIENCIES ON THE PENETRANCE OF MELANOTIC TUMOURS IN *DROSOPHILA MELANOGASTER*

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While the penetrance of *tu* genes has been shown to depend on nutritional variables, no detailed examination of these has been made using germ free culture on defined diets. Under such axenic conditions, it can be shown that tumour penetrance in a new *tu* strain is raised by reducing to sub-optimal amounts the dietary RNA, cholesterol or biotin. The RNA effect is not produced indirectly through DNA, but results from a shortage of adenylic acid and is dependent on the dietary adenylic-cytidylic balance. At threshold amounts of RNA, sub-optimal supplies of thiamine, pyridoxine and folic acid all increase tumour penetrance by interfering with the *de novo* synthesis of adenylic acid by the larvæ. Nutritional manipulation affects

tumour formation only during a very short phase after the second ecdysis (*c.* 45-50 hours from hatching) suggesting that this is the period when the *tu* gene is effective. High tumour penetrance resulting from biotin or cholesterol deficiencies is not greatly reduced by raising the RNA supply, so the three effective treatments must operate through different metabolic routes to produce the same phenotype.

A STRAIN COMPARISON OF TUMORIGENIC RESPONSES IN *DROSOPHILA MELANOGASTER*

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The penetrance of melanotic tumours is low in the tu^K and *tu bw* ; *st Su-tu* strains reared on fully supplemented chemically defined aseptic media. In both strains tumour penetrance responds to the same dietary deficiencies, but there are quantitative differences in sensitivity to each treatment (nucleotide imbalance, cholesterol and biotin deficiencies). Both strains show tumours when fed excess tryptophan, but only the latter responds to X-irradiation of the egg. Crosses between the strains behave differently according to treatment. Chromosome analyses of the response to the different effective treatments show that while both strains carry major *tu* genes in the second chromosome they also carry different modifying genes (including suppressors) on the other chromosomes. The difficulties involved in this type of analysis, and their implications, will be discussed. In particular, whether the biochemical picture refers to the *tu* gene or to its suppressor.