## GENETICAL SOCIETY OF GREAT BRITAIN

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#### ABH SECRETOR AND LEWIS CHARACTERS IN MAN

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Using material supplied by Drs I. Bianco, E. Silvestroni and M. Siniscalco, from 320 individuals, including members of 66 families, in Ferrara, Italy, further evidence has been obtained of the interaction of ABH secretor (i.e. Se, se alleles) and Lewis (i.e. L, l alleles). The ABH secretor status of each individual was determined by standard inhibition tests on saliva.

The presence of Le<sup>a</sup> substance in the saliva sample was also determined by inhibition tests. Using an arbitrary scale, each individual was given an inhibition score. In a few cases there was difficulty in classifying individuals as Lewis-positive (*LL* or *Ll*) or Lewis-negative (*ll*). The discrimination was improved by doing similar inhibition tests with anti-Le<sup>b</sup> on a selected sample and deriving a total score for the amount of Le<sup>a</sup> and Le<sup>b</sup> substances. The frequency of *ll* in the unrelated individuals of the whole sample was 10.61 per cent.

Family data supported the hypothesis that L (Lewis present) is dominant to  $\ell$  (Lewis absent). The presence of Lewis substances in the saliva is independent of the presence of ABH substances, but the amount of Lewis substances in saliva and their presence in detectable amounts on erythrocytes is influenced by the secretor-phenotype.

## FAMILIAL LOW PSEUDOCHOLINESTERASE LEVEL IN PLASMA

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Plasma pseudocholinesterase is measured principally for two practical reasons. The enzyme level is low in liver disease and its determination is used as a liver function test. The short-term action of the widely used relaxant succinyl-dicholine is due to its rapid destruction by pseudocholinesterase. Occasionally a prolonged apnœa—due to excessively prolonged paralysis of the respiratory muscles—is encountered by anæsthetists using succinyl-dicholine, and it can then be traced to a low pseudocholinesterase level. Sometimes this can be explained by liver disease. On occasions no such explanation can be found. This "idiopathic" enzyme deficiency could be shown to be familial. The range of normal enzyme level is wide. Nevertheless with the increasing number of family trees studied a pattern representing the homozygous and heterozygous states can be recognised.

#### GENETIC CHANGE IN DIPLOID YEAST

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The frequency of spontaneous change from heterozygosity to homozygosity at specific loci in diploid cells of *Saccharomyces cereviseæ* has been determined using a visible gene marker, namely red colouration. When colonies derived by plating

out white diploid cells of constitution  $\propto ad_1$  (red) me tr + /a + (white) + + ur are examined, up to 42 per cent. are sectored red and approximately 0.05 per cent. are pure red. All single-cell isolates from these colonies are diploid. Further analyses of single-cell isolates from each of the red and white portions of 55 sectored colonies revealed among the white cells two homozygous methionine-requiring strains and two uracil requirers, and among the red cells one uracil and one methionine requirer.

Such frequencies of genetic change at specific loci are in sharp contrast to those which occur in haploid cells; among 2117 colonies derived from haploid cells treated with ultra-violet irradiation and tested for auxotrophy by the "total isolation" technique, only one isolate was found to have mutated at the  $+/ad_1$  locus. In the same experiment nine methionine, six tryptophane and two uracil-requiring auxotrophs were recovered which have not yet been tested for allelism; several loci may therefore be represented by each frequency.

In view of this contrast it is tempting to deduce that a mechanism other than what may be termed "classical" gene mutation is responsible for the high frequency of change at specific loci in heterozygous diploid cells. Alternative explanations to account for these preliminary observations are discussed.

### SELF-FERTILITY IN THE FIELD BEAN (VICIA FABA L.)

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Estimates of natural cross-pollination in commercial crops of field beans vary from 20-70 per cent., although under normal conditions an average of 30 per cent. is most common. Despite this, few seeds are set when insects are excluded and even after artificial tripping of the flowers is performed complete seed set is rarely attained.

Spontaneous self-fertility (without flower tripping) in a population shows a continuous distribution when measured in terms of total number of seeds produced per plant.

A diallel cross performed using six parent plants to assess their relative cross-and self-compatibilities did not demonstrate consistent superiority of crossing over selfing but rather that certain plants crossed more successfully than others. The progeny from this diallel cross when grown in an insect-proof greenhouse exhibited considerable differences in spontaneous self-fertility. Crosses were generally more self-fertile than selfs and some parents gave progeny which were more self-fertile than others. The Covariance/Variance (Wr/Vr) graph which could be constructed from the data indicated considerable interaction ( $b = o \cdot 32 \pm o \cdot 13$ ) and this interaction disappeared on the removal of one parental array.

From the data presented, a system of self-incompatibility has been postulated which is largely based on polygenes, and which has developed by the breakdown of a once efficient system through the effect of unconscious selection for uniformity throughout the past history of the crop.

#### SYNDROME OF DEAFNESS AND GOITRE

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In recent years several types of familial goitre have been described; they are apparently due to inborn errors of metabolism and are inherited in a recessive manner. One such type is characterised by a failure of the thyroid to bind iodine normally and is also invariably accompanied by severe congenital nerve deafness. Clinical and genetical aspects of this syndrome in several families are discussed.

### MATERNAL EFFECT OF ma-I+ ON XANTHINE DEHYDRO-GENASE OF DROSOPHILA MELANOGASTER

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Previous work established that maroon-like (ma-l) and rosy (ry) eye-colour-mutants of Drosophila melanogaster are deficient in xanthine dehydrogenase, and therefore the substrates of this enzyme (hypoxanthine and 2-amino-4-hydroxy-pteridine) accumulate, while the products (uric acid and isoxanthopterin, respectively) are lacking. Recent studies have shown that ma-l/ma-l+ females exert a maternal effect, in that their ma-l progeny have a wild-type eye-colour. However, ry+/ry females do not exert a similar effect on their ry progeny. Biochemical studies have shown that in addition to increased amounts of red eye-pigments, maternally-affected ma-l flies have traces of isoxanthopterin and uric acid (the enzyme reaction products), as well as traces of the enzyme itself.

The maternal effect is probably due to a substance which ma-l+/ma-l females pass through the egg, and which is utilised by their ma-l progeny. On the other hand, ma-l+/ma-l; ry/ry females still maternally affect ma-l progeny, indicating that ry synthesizes this compound; its utilisation is probably blocked in this mutant and thus there is no maternal effect upon ry. This suggests that a sequential relationship exists between reactions blocked by these mutants.

## THE AXES OF LAMPBRUSH CHROMOSOMES AND OF THEIR LATERAL LOOPS

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The structures which project laterally from the axes of newt lampbrush chromosomes have diverse morphologies. Most of these structures are loops, though in some the loop form is obscured by fusion. There is an axis within each lateral loop, and this axis is coated with matrix. Digestion experiments with proteolytic enzymes and nucleases have shown that loop matrices consist of ribonucleoprotein, whereas loop axes, and also the connections between successive loops, contain deoxyribonucleic acid. The linear integrity of a lampbrush chromosome is maintained by an uninterrupted fibre, often centimetres long, of this latter substance.

# PHOTOPERIODIC CONTROL OF BREEDING SYSTEM IN ROTTBŒLLIA EXALTATA

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In the short-day grass, Rottballia exaltata L.f., each segment of the inflorescence bears three flowers, one sessile and hermaphrodite, one sessile and potentially male, and the third, stalked, also potentially male. In daylengths and night temperatures promoting early flowering, the two male flowers are sterile and the anthers of the hermaphrodite flower dehisce before exsertion, so that self-pollination is obligate. If plants are exposed for some weeks after germination to daylengths greater than the critical maximum for flowering and then subsequently induced to flower by transfer to short days, fertility is restored to some of the male flowers, and the anthers of the hermaphrodite flowers are exserted before dehiscence. Under these conditions plants are cross-pollinated. Breeding behaviour thus depends upon the photoperiodic experience of the individual plants, and this may be determined by the time

of the year when germination happens to take place. This environmentally regulated versatility of the breeding system in *Rottbællia* may be compared with that of *Silene pendula*, a long-day species in which the sexual function of individuals is determined by their photoperiodic experience in early life.

# FORWARD AND BACK MUTATION AT THE PYR3 LOCUS OF NEUROSPORA

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The citrulline requirement of mutant 33442 (cit) is suppressed by mutation of  $pyr_3$ + to  $pyr_3$ . Strains of the cit,  $pyr_3$ + genotype require only citrulline, while cit,  $pyr_3$  strains require only pyrimidine for growth. This makes it possible to select for forward as well as for back mutants at the  $pyr_3$  locus.

For forward mutation, a cit,  $pyr_3^+$  microconidial strain is plated on minimal supplemented with hydrolysed RNA (plus lysine and canavanine to eliminate leakage). A rate of  $3 \times 10^{-5}$  colonies per viable conidium was obtained after UV treatment (85 per cent. killing). Out of 19 colonies tested, 11 could grow on minimal medium, presumably due to back mutation at the cit locus. The rest were pyrimidine-dependent due to forward mutation in the  $pyr_3$  region. These were tested for the ability to complement each other in heterokaryons, i.e. to dispense with the pyrimidine requirement when grown together in pairs. This revealed two complementary groups, with four mutants in each. No recombinants were obtained among  $5.8 \times 10^4$  ascospores from inter-group crosses.

For back mutations, a cit,  $pyr_3$  strain is plated on minimal plus arginine (or citrulline). Treatment with UV (75 per cent. killing) yielded  $10^{-5}$  colonies per viable microconidium. Roughly half of these were citrulline-dependent, and presumably back mutations at  $pyr_3$  as shown for one case. The other half could grow on minimal. In one case analysed, this was due to mutation at an unlinked suppressor locus.