

GENETICAL SOCIETY OF GREAT BRITAIN

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THE INFLUENCE OF THE ENVIRONMENT IN ANENCEPHALY

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Data on 4420 anencephalics born in Scotland between 1939 and 1956 are presented, various environmental features associated with differing incidence are discussed, and some limitations of the nature-nurture issue in relation to observational data are considered.

The basic findings are :—(1) There is a marked association with social class, the variation in incidence being fourfold. (2) There is a consistently lower incidence in illegitimate pregnancies. (3) The liability in first pregnancies is 40 per cent. higher than in second. (4) The incidence probably increases with maternal age, but may also be relatively high under the age of 20. (5) There is a marked regional variation, but urbanisation itself does not seem to be a factor. (6) The marked secular changes are largely related to a changing incidence in first births and in winter births. (7) There is a marked seasonal variation, the incidence being highest in mid-winter. (8) There is evidence that epidemic infections are not related to any considerable number of cases.

ACROCEPHALOSYNDACTYLY

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Acrocephalosyndactyly is a rare malformation which may conveniently be divided into two main sub-groups, (i) Apert's syndrome and (ii) other types. This paper is mainly concerned with (i) which is the commonest type.

The deformities in Apert's syndrome have been attributed to syphilis, rickets, chronic osteitis, hormonal dysequilibrium and even to maternal rubella. Recent authors have considered a hereditary origin probable.

Analysis of the pedigrees of 34 sporadic cases in the author's British series makes it very likely that sporadic cases of Apert's syndrome are due to new mutation. The discovery in Holland of an affected mother and child confirms this suspicion.

Of great interest to the geneticist is the marked effect of parental age. An increase in the mean ages of the parents at births of propositi can be attributed wholly to a raised paternal age ; so also can the associated shift in the birth order. In this respect the condition is very like achondroplasia.

Estimates of the incidence of Apert's syndrome at birth and in the general population are given.

PHAGE-MEDIATED TRANSDUCTION OF COLICINOGENY IN *SALMONELLA TYPHIMURIUM*

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Colicinogeny, *i.e.* ability to produce a colicine (an antibiotic active on certain coliform bacteria), is transferred with high frequency to cells of a non-colicinogenic

strain when these are mixed with cells of a suitable colicinogenic strain, the transfer probably resulting from cell conjugation. *Shigella sonnei* strain P9 produces two different colicines, which we term C_1 (probably colicine I) and C_2 (probably colicine E). When a non-colicinogenic *Salmonella typhimurium* strain (LT2) was mixed with P9, two different kinds of colicinogenic *Salmonella* were obtained, some clones producing colicine C_1 alone, others both C_1 and C_2 ; no *Salmonella* producing colicine C_2 alone have been detected in these experiments. Similar results were obtained when a doubly colicinogenic *Salmonella* strain thus produced was mixed with a (genetically marked) non-colicinogenic *Salmonella* strain.

In transduction experiments phage PLT22 grown on the doubly colicinogenic *Salmonella* was applied to a non-colicinogenic *Salmonella* strain; some colicinogenic clones were obtained, their frequency being about the same as in the transduction of other characters. Of the clones made colicinogenic by transduction all those so far tested produce only colicine C_2 , a class not encountered at all when colicinogeny was transferred between the same strains by mixing the cultures.

COMBINATION OF INFORMATION FROM 2×2 TABLES

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Provided attention is confined to only two blood types, the data for studying the relation between blood group and disease can be set out as a 2×2 table thus:—

		TRAIT :—Blood Group		
		<i>a</i>	<i>b</i>	
GENUS I : Patients	a	b	E	
GENUS II : Control Series	c	d	F	
	S	T	N	

Samples from two sub-populations or *genera* are sub-divided according to some *trait*. Data conforming to this pattern are common in genetics. Often there are several tables, the results of comparisons under different conditions such that the incidence of the trait would be expected to vary in both genera, but in such a way that there should be a more or less constant functional relation between the relative incidence figures in the different tables. Methods will be described for statistical analysis of such sets of data.

For the case of blood group and disease, Woolf (1955) suggested the use of the *relative incidence ratio*, $x = ad/bc$. For a set of tables, a weighted mean is computed of $y = \ln x$, with test of heterogeneity and significance. This *Empirical Estimate* is a "large sample" method, and breaks down entirely if any cell frequencies are zero. Simple computations will be described for obtaining the *Maximum Likelihood Estimate*, and for a rapid *Null Test and Estimate* which is often all that is needed.

These methods are valid for a wide class of postulated functional relationships, including for example comparative mutation rates. By use of the Null Test, the applicability of a selection of different "models" can be quickly assessed.

REFERENCE

WOOLF, B. 1955. *Annals of Human Genetics*, 19, 251.

**β -GLOBULIN POLYMORPHISM IN CATTLE AND
OTHER MAMMALS**

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Six β -globulin phenotypes have been found in the serum proteins of British breeds of cattle by starch gel electrophoresis. The six phenotypes are individual genotypes, being the homozygotes and heterozygotes of three alleles, β^A , β^D , and β^E . The gene frequencies differ markedly between breeds, β^E being least frequent in each breed.

Abnormal segregation ratios have been obtained with certain matings. Thus $\beta^A/\beta^A \times \beta^A/\beta^D$ and $\beta^D/\beta^D \times \beta^A/\beta^D$ result in a significant excess of offspring of the same genotype as the mother. The matings $\beta^A/\beta^E \times \beta^A/\beta^A$ and $\beta^D/\beta^E \times \beta^D/\beta^D$ result in a significant lack of offspring with β^E in their genotype.

β^E is clearly at a disadvantage, which may be partially offset by the apparent economic advantages of β^E heterozygotes. There is evidence of a cline in the frequency of β^E .

β -globulin polymorphism has been found in sheep and goats. The genetic mechanism is analagous to that in cattle. Two β -globulin alleles are postulated in goats and five in sheep.

Serum protein polymorphism has also been detected in the thread-proteins of cattle, in the slow-alpha proteins of cattle, in the pre-albumins of horses, and in the serum proteins of white mice.