

THE EFFECT OF ASCERTAINMENT BY TRUNCATE SELECTION ON SEGREGATION RATIOS

El efecto de la determinación mediante selección truncada
sobre los cocientes de segregación

L'effet de la détermination à travers la sélection tronquée
sur les quotients de ségrégation

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The present paper is concerned with the problem of early detection of the possible heritable nature of abnormalities in domestic animals. Although the principles are applicable to dominant as well as recessive autosomal traits reference is made especially to the latter because of their relative importance in animal breeding and production.

Assuming that evidence once has been obtained for simple recessive inheritance of an undesirable trait the next step is an evaluation of the importance of its presence among the animals used for breeding purposes. This depends primarily on the breeding policy and the efficiency of identification, registration and elimination of heterozygous individuals. However, such problems are considered in various papers and textbooks, see for example HUTT (1961, 1964) and LERNER and DONALD (1966). Here it may suffice to mention that identification of heterozygous individuals usually involves testmatings relative to the same *a priori* segregation ratios as those considered in the following. However, the application of these *a priori* ratios in the present context has a quite different purpose as mentioned in the first paragraph, namely to facilitate early detection of abnormalities of possible genetic origin.

Application of methods for early detection of the mode of inheritance of undesirable traits is particularly important in cattle because the considerable generation interval makes the procedure of testmatings time consuming, and even if similar matings have taken place unpurposely they might not always be sufficiently registered to justify assumption of complete ascertainment. Moreover, the evidence indicates that the number of undesirable traits of established or of

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probable genetic origin already approaches two hundred and fifty (cf. LAUVERGNE, 1968) and additional traits clearly are expected as new mutations arise.

However the method described in the following is not only applicable to cattle but also to the other domestic animals, uniparous as well as multiparous species. The method is an expansion of a method developed by BERNSTEIN (1929) and in the present paper it is used in connection with the Mendelian segregation ratios corresponding to *a priori* expectations of 1:1, 3:1 and 7:1. These segregation ratios are expected if (1) the genotypes of the parents are known and (2) if the ascertainment of family material is by complete selection. Meanwhile the latter requirement is usually only fulfilled in material obtained from (or corresponding to) planned matings in which all data are considered. If a certain group or class of data is cut off (= truncated) or excluded from observation then different segregation ratios are expected.

The three mating types $Aa \times aa$, $Aa \times Aa$ and $Aa \delta \times \{50\% AA \text{ } \varphi \text{ } \varphi + 50\% Aa \text{ } \varphi \text{ } \varphi\}$ are expected to yield dominant: recessive offspring in the ratios 1:1, 3:1 and 7:1, respectively. However, if the heterozygosity of a parent is deduced exclusively on the basis of the occurrence of at least one recessive individual (genotype aa) among its offspring the situation is quite different simply because matings of heterozygous parents yielding exclusively dominant offspring (genotype AA or Aa) cannot be included in the data because such families are not detectable. Therefore, the expected proportion of recessive offspring from heterozygous parents ascertained their offspring through will exceed *a priori* expectations. The deviation from *a priori* expectation depends on the number of individuals within each sibship, but since the bias is systematic and has an effect in one direction its influence on the segregation ratios can be measured.

The proportions of recessive offspring corresponding to the 1:1, 3:1 and 7:1 *a priori* segregation ratios are $q = 1/2$, $1/4$ and $1/8$, respectively. The corresponding proportions of dominant offspring are $p = 1/2$, $3/4$ and $7/8$, respectively. The *a priori* fractions of $q = 1/2$ and $q = 1/4$ usually refer to the fraction of recessive (or affected) individuals within a sibship of a given size (n) whereas the *a priori* fraction of $q = 1/8$ in this context refers to the fraction of recessive (or affected) individuals within that part of a sibship which is located within a herd. Thus in this instance n refers to the number of relevant sibs within a herd, and the total sibship includes sibs placed in different herds. This total sibship consists of paternal halfsibs the mothers of which likewise are paternal halfsibs.

As explained previously the proportion (q') of recessive offspring to be expected within detectable sibships or herds exceeds the *a priori* expectation (q) and it is possible to compute the *corrected* expectation q' . Since the fraction of sibships or herds with at least one recessive individual is $1 - p^n$ the expected proportion q'

of recessive individuals within this fraction is $q' = \frac{q}{1 - p^n}$. Consequently, the expected number of recessive individuals in detectable sibships or herds is $q' \cdot n = \frac{q \cdot n}{1 - p^n}$, and the expected number of dominant individuals is $1 - q' \cdot n = 1 - \frac{q \cdot n}{1 - p^n}$.

The corresponding values for $n = 2, 3, \dots, 16$ are indicated in Table 1 for each of the three examples of *a priori* expected segregation ratios. One should notice

that families with a single offspring (obviously a recessive) are not considered because they do not yield any information on the segregation ratios, and hence they are excluded from analysis.

The application of the figures in Table 1 enables one to apply a chi-square test for testing a hypothesis of simple autosomal inheritance based on material obtained by truncate selection. For example, when ANDRESEN *et al.* (1970) investigated the genetics of the lethal factor A 46 in cattle they observed a segregation

TABLE 1

THE CORRECTED NUMBER OF DOMINANT ($d = 1 - q' \cdot n$) AND RECESSIVE ($r = q' \cdot n$) INDIVIDUALS WITHIN SIBSHIPS (OR HERDS) OF SIZE $n = 2, 3, \dots, 16$, EACH WITH AT LEAST ONE RECESSIVE INDIVIDUAL. FIGURES ARE INDICATED FOR THREE DIFFERENT «A PRIORI» EXPECTED SEGREGATION RATIOS

Family size n	1:1		3:1		7:1	
	d	r	d	r	d	r
2	0.667	1.333	0.857	1.143	0.933	1.067
3	1.286	1.714	1.703	1.297	1.864	1.136
4	1.867	2.133	2.537	1.463	2.792	1.208
5	2.419	2.581	3.360	1.640	3.717	1.283
6	2.952	3.048	4.175	1.825	4.640	1.360
7	3.472	3.528	4.980	2.020	5.559	1.441
8	3.984	4.016	5.778	2.222	6.477	1.523
9	4.491	4.509	6.567	2.433	7.391	1.609
10	4.995	5.005	7.351	2.649	8.304	1.696
11	5.497	5.503	8.129	2.871	9.214	1.786
12	5.999	6.001	8.902	3.098	10.122	1.878
13	6.499	6.501	9.671	3.329	11.027	1.973
14	7.000	7.000	10.437	3.563	11.931	2.069
15	7.500	7.500	11.199	3.801	12.833	2.167
16	8.000	8.000	11.960	4.040	13.732	2.268

ratio of 40:13 resembling the Mendelian 3:1 ratio, although the material was relevant to a 7:1 *a priori* expectation of normal: affected individuals. However, the observed ratio was in good agreement with a *corrected* 7:1 expectation. It should finally be emphasized that it is necessary to consider several precautions when applying this method as discussed by ANDRESEN (1974).

SUMMARY

Animal material for genetic investigations of recessive traits may be based solely on families in which the trait occurs only among the offspring. Since matings of heterozygous individuals having exclusively dominant offspring are not included in the material the segregation ratios 1:1, 3:1, or 7:1 are not expected among the offspring. A method of correcting such ratios is described and the results are tabulated. After correcting the data a chi-square test may be applied for testing a hypothesis of simple autosomal inheritance.

RESUMEN

El material animal para las investigaciones genéticas de los caracteres recesivos puede basarse solamente en familias en las que aquéllos aparezcan en la prole. Como los cruzamientos de individuos heterocigotes que dan prole exclusivamente dominante no se incluyen en el material, los cocientes de segregación 1:1, 3:1 ó 7:1 no pueden esperarse en aquélla. Un método para corregir tales cocientes se describe en el presente trabajo, tabulándose los resultados. Después de la corrección de los datos, una prueba *chi-square* puede aplicarse a fin de determinar la hipótesis de la herencia simple autosómica.

RESUME

Le matériel animal pour les recherches génétiques des caractères récessifs, peut se baser seulement sur les familles où ceux-ci apparaissent chez leurs descendants. Comme les croisements d'individus hétérozygotes ayant une descendance exclusivement dominante, ne sont pas inclus dans le matériel, les quotients de ségrégation 1:1, 3:1, 7:1, ne peuvent être espérés chez celle-là. Une méthode pour corriger de tels quotients est décrite dans ce travail, étant tabulés les résultats. Après la correction de données, on y peut appliquer une preuve *chi-square*, afin de déterminer les hypothèses de l'hérédité simple autosomique.

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