CYTOGENETIC CONSIDERATIONS
IN ANIMAL BREEDING*

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SUMMARY

Some fundamental aspects of cytogenetics, a brief review of work done with farm animals, and an assessment of the known effects of chromosomal aberrations on animal population are summarized in this paper.

The basic chromosome number and other detailed features of the karyotype have been reliably established for all the farm animals within the past 10 years. Intra-species variants, i.e. chromosomal polymorphisms have been found in cattle, pigs, and goats.

Two primary types of aberrations are recognized. The first, an alteration of numbers of whole chromosomes is known as heteroploidy. The second type, rearrangement of chromosomal segments, results following physical breaks of chromosomes.

Factors affecting the frequency with which aberrations occur include both genetic and non-genetic elements. Among the most important in the latter category are certain virus groups, some drugs and other chemical compounds, ionizing radiations, age of gametes, particularly ova, and in some animals, maternal age.

Presence of chromosomal aberrations are apparently responsible for a large proportion of the embryonic deaths in mammals and birds. In man, perhaps as many as one-third of spontaneously expelled abortuses are afflicted. Early indications from work with pigs indicates that the situation may be similar. Very little cytogenetic work has been done with congenitally malformed livestock, although a number of aberrations have been reported. In man, somewhat less than 1 p. 100 of new born babies carry chromosomal abnormalities.

Some aberrations, although they cause only slight if any somatic alteration, can be the source of reduced fertility or sterility in their bearers. Isolated cases of this phenomenon have been reported but a quantitative assessment of its importance has not yet been made.

The presence of centric fusions as chromosomal polymorphisms in cattle, pigs, and goats has been reported, and work is being conducted to ascertain how they effect fitness of the populations in which they occur.

It is concluded that the occurrence of chromosomal aberrations causes a serious reduction of potential productivity. The extent of this loss should be accurately determined and means found to reduce it.

I. — INTRODUCTION

Interest in the chromosomal complements of farm animals has been high since early in the 20th century when it was first established that the chromosomes were the vehicles on which the genes were carried. Particularly European and Japanese workers pursued the question of the normal chromosome numbers of the domesticated species. It is a tribute to the astuteness and perseverance of the pioneers that although techniques were laborious and inadequate, they managed, sometimes with uncanny accuracy to describe the karyological make-up of a number of vertebrates. (For a historical review of ruminants see HULOT and LAUVERGNE, 1967.)

Three technical developments of the 1950's enabled cytological studies of mammals to be made much more readily, accurately, and on larger samples of specimens. These were (1) HSU and POMERAT's (1953) discovery of the salutary effect of the treatment of cells in hypotonic solution prior to fixation; (2) the use of mitosis arresting agents, most notably colchicine, to arrest cell division at a stage most suitable for observation of chromosomes; (3) the development of simplified techniques for cell and tissue culture, particularly the startling finding of MOORHEAD et al. (1960) that certain substances would induce mitosis of lymphocytes.

Although various modifications of these new found methods have been widely used to study variations in the chromosomal complement of man, they have only quite recently been used by workers interested in increasing productivity of farm animals.

II. — CHROMOSOMES OF LIVESTOCK

The normal karyotype of all domesticated mammals and most of the birds has, by now been well established (HSU and BENIRSCHKE, 1970). In Table 1 this information is summarized for the more common ungulates. There are a number of interesting features characterized by the data on this table. First note the extreme evolutionary stability in the number of chromosomal arms in a wide spectrum of species of *Bovidae*. This point has been well made by the studies of a number of taxa by WURSTER and BENIRSCHKE (1968). In contrast is the wide variability exhibited by karyotypes of the various equine species. This fact would be even more notable if I had listed data for some of the zebras (see HSU and BENIRSCHKE, 1970). Secondly, it is interesting to note that although relatively few animals of any of these species have been examined, already chromosomal polymorphisms have been found. The centric fusion in cattle is in reference to the exhaustive study made by GUSTAVSSON (1969) of *Swedish Red and White Cattle*. A similar type of polymorphism is being studied by McFEE and others (1966) in a group of wild pigs descended from an importation to the U.S. from Germany. It appears that Zebu cattle possess an acrocentric Y chromosome while the Y of European cattle is submetacentric (KIEFFER and

CARTWRIGHT, 1968). Undoubtedly as more animals are looked at from a wider geographical spectrum more variants will be found. They are interesting in their own right and because they shed some additional light on the evolutionary history of our livestock.

TABLE I

Characteristics of the karyotypes of some domesticated ungulates

TABLEAU I

Caractéristiques caryotypiques de quelques ongulés domestiques

Species	2N	Number of autosomal arms	Sex chromosomes		Known variants
			X	Y	
Cattle.	60	58	L Sm	S Sm S A	Centric fusion Y
Goats	60	58	MA	SM	Centric fusion
Sheep	54	58	LA	SM	
Pig	38	60	M Sm	SM	Centric fusion
Horse	64	88	L Sm	SA	
Ass	62	98	L Sm	SA	

L - Large   M - Metacentric
M - Medium   Sm - Submetacentric
S - Small   A - Acrocentric

III. — TYPES OF ABERRATIONS

Before proceeding with a discussion of the causes and effects of aberrations of chromosomes, it might be well briefly to review the types of things that can happen to chromosomes and to establish some terminology. A more complete discussion was previously published (FECHHEIMER, 1968). In general there are two types of alterations that can occur. The first is an alteration in the number of whole chromosomes present in a cell and the second is a structural rearrangement of chromosomal segments following the occurrence of physical breaks.

Alteration of the normal 2 N somatic number by whole multiples of a gametic set is known as euploidy. The two primary categories are haploidy and polyploidy, of which triploidy (3 N) and tetraploidy (4 N) are the most frequently seen types. Aneuploidy refers to a change in the genome of one or a few chromosomes. The most frequent types of hypodiploidy and hyperdiploidy are monosomy and trisomy respectively. These terms refer to karyotypes with one missing chromosome and one additional chromosome. In mammals all heteroploid conditions with the exception of some trisomics and monosomic X are lethal and result in the premature death of the animal, usually prior to birth.

Structural aberrations may be classified upon the number of breaks that necessarily preceeded the rearrangement of material. Thus one break results ultimately in the loss of a terminal segment. If the break is through the centromere, an isochromosome, i.e. one with two identical arms, may arise.

Two breaks may occur in the same chromosome. This can lead to an interstitial deletion, an inversion, or formation of a ring. Should one break occur in each of two chromosomes, the most likely recovery products would be translocation chromosomes each of which possessed a centromere. Products resulting from other types of refusions of broken ends are mostly unstable.

Quite obviously it is possible to have multiple aberrations, either in the same cell line or in separate cell lines of the same organism. It may seen unlikely, given the low frequency of occurrence of most aberrations, that one would find multiple aberrations, but such is not the case. The physical event giving rise to one error in many instances produces two new aberrant cells at the same time. An example of this would be nondisjunction of a chromosome at an early cleavage division. In this case two new lines, one monsomic and one trisomic for the chromosome in question would be generated simultaneously by the single event.

Finally, in discussing organisms which possess more than one cell line, it is useful to distinguish between mosaics and chimerae. The former is the of two or more cell lines derived from the same zygote. Chimerism is the presence presence of an additional cell line in an organism, derived from another zygotic product. Freemartins are therefore chimerae.

IV. — VARIABLE EFFECT OF ABERRATIONS

The effect that any of these chromosomal aberrations will exert on an organism or on a population depend upon a number of interrelated factors, some of which are briefly discussed.

a) Quite obviously if a large chromosomal segment were lost (or perhaps duplicated) the expected effect would be greater than if a small segment were involved. This would be particularly so if the lost segment were composed primarily of heterochromatin and thus contained little genetic activity.

b) Some aberrations, e.g. inversion or translocation, when carried as heterozygotes, will exhibit no somatic manifestation on their bearers. However, at meiosis complications do arise and aberrant gametes will be produced. Secondary aberrations are, as a consequence to be expected in some of the progeny of parents carrying a structural aberration.

c) The later in embryogenesis that an aberration occurs, the more attenuated will be its influence on the organisms. If, for instance it occurs at the first cleavage division then all (or at least half) of the zygotes cells will eventually contain an aberrant genome. On the other hand if it first occurs in one cell in an already differentiated tissue, only a small proportion of the organisms cells will contain an altered genome and no noticable alteration of phenotype need be produced.

d) Some aberrations are self limiting in that they produce such profound effects that they severely alter the mitotic activity of the cell containing them. In this case the aberrant cell line will remain small and may not manifest itself at all. Very little is in fact known about competition and selection among genetically different cell lines.

e) Other phenomena of embryogenesis can serve to attenuate the phenotypic expression of an aberration.

(1) It was pointed out by BEATTY (1957) that if an event occurs during the first few cleavage divisions, the aberrant cell line might be entirely included in the extra-embryonic tissues, because only a few cells in the inner cell mass are destined to contribute to the embryo itself.

(2) The presence of dosage compensation mechanisms is another factor that alters the effect of an aberration. Inactivation of one X chromosome in each cell of developing mammals is such a mechanism that was first postulated by LYON (1962). When an aberrant X is present, it has been shown that it is preferentially inactivated so that most cell lines are left with the normal X as the fully functional one.

f) Just as with gene mutations, the same event even in otherwise genetically identical populations will produce variable effects depending upon the environment in which the population is found. The fact has been elegantly demonstrated by DOBZHANSKY and his group. They have observed that the frequency with which particular inversions are observed in populations of *Drosophila* is influenced by a number of environmental factors. Some inversions, detrimental to the population in one ecological niche, enhance its fitness in others.

V. — CAUSATIVE FACTORS OF ABERRATIONS

Explicit in the preceding discussion is the concept that mitosis and meiosis are not invariably regular processes that always give rise to the expected end products. Both are subject to a variety of irregularities, each of which has its own expected array of aberrant end products.

In Table 2 are listed a number of agents that have been implicated as etiological factors of chromosomal aberrations. It will be seen that, like any other characteristic of living organisms both genetic and non-genetic factors are involved. Let me very briefly relate the nature of some of the evidence implicating the factors listed. In regard to genetic control, the evidence is quite clear that incidence of heteroploidy differs in genetically distinct stocks of mice. Furthermore, selection for increased incidence yielded a positive response (see BEATTY, 1957 for review). In cattle inbred lines derived from a common source have been shown to differ in the frequency of cells with hypodiploid or hyperdiploid chromosome complements (ZARTMAN and FECHHEIMER, 1967). At least one gene is known in man that produces chromosome breaks and also brings about a relatively high rate of somatic pairing (GERMAN and ARCHIBALD, 1965). That meiosis is primarily a genetically controlled process is clear from studies of actions

TABLE 2

List of factors known to affect the incidence of chromosome aberrations

TABLEAU 2

Liste des facteurs connus pour influer sur l'apparition des aberrations chromosomiques

Agent	Effect of Agent	Reference (Review as key)
Genes	Chromosome breakage Nondisjunction Aneuploidy Polyploidy Control of meiotic events	GERMAN and ARCHIBALD (1965) HAUSCHKA et al. (1962) ZARTMAN and FECHHEIMER (1967) BEATTY (1957) SANDLER et al. (1968)
Primary aberrations	Chromosome breakage Aneuploidy	BURNHAM, 1962
Autoimmune disease	Nondisjunction	BURCH et al., 1966
Ionizing radiations	Chromosome breakage Polyploidy Nondisjunction	KIHLMAN, 1961 MILLARD, 1965 GRIFFIN and BUNKER, 1967
Drugs and other chemical. . .	Chromosome breakage	EVANS, 1970
Compounds	Polyploidy	KIHLMAN, 1966
Viruses	Chromosome breakage Nondisjunction Loss of chromosome	EVANS, 1970 BURDETTE and YOON, 1967 BURDETTE and YOON, 1967
Temperature shock	Polyploidy	BEATTY, 1957
Maternal age	Meiotic disjunction	PENROSE, 1966
Age of gametes ova	Polyploidy Aneuploidy	VICKERS, 1969 WITSCHI and LAGUENS, 1963
Sperm	Uncertain-indirect evidence	SALISBURY, 1965
Age and physiological state . .	Polyploidy of somatic cells Chromosome breakage Loss of sex chromosome Aneuploidy	SWARTZ, 1956 CROWLEY and CURTIS, 1963 COURT BROWN et al., 1963 BRUÈRE, 1967

of many mutant genes in bringing about variations that lead to production of aberrant gametes. Most of such work has so far been done with plants and lower animals (SANDLER et al., 1968).

Primary aberrations, both numerical and structural, cause complications at meiosis that result in new secondary aberrations in the gametic genomes. Frequently however the relationship cannot be discovered because the primary aberration will have been cryptic. An example of this would be paracentric inversions which can be discovered only by linkage studies or by viewing germ line cells at fist meiotic prophase. They cannot be seen in somatic cell preparations.

The relationship between autoimmune disease and aberration is not yet clear. It has been noted that parents of babies with monosomic or trisomic conditions have a higher incidence of autoimmune disease than paired controls (BURCH et al., 1966; VALLOTON and FORBES, 1967).

A multitude of specific external agents appropriately applied to cells will bring about aberrations. The entire spectrum of ionizing radiations, many chemical compounds (EVANS, 1970), certain virus groups (NICHOLS, 1966) and heat shock (BEATTY, 1957) produce the indicated effects. The action of some of these agents is fairly well understood while for others nothing at all is known except that results are repeatable.

The maternal age effect manifests itself in man where there is a precipitously increased frequency of trisomic children born to mothers over 40 years of age (PENROSE, 1966). While the same phenomenon was not observed in mice (GOODLIN, 1965), it may be that an unfortunate choice of lines was used for the study. EDWARDS (1970) has shown that chiasma frequency is lower in oocytes of older females, both human and mice. He argues that this fact could be a causative agent for increased trisomy.

When mating is delayed much beyond the time of ovulation, mammalian ova either lose their mechanism of defense against penetration by more than one sperm or do not undergo a second meiotic division. In both instances the zygote contains more than two haploid sets of chromosomes, i.e. is polyploid. Polyploidy resulting from delayed mating has been demonstrated in mice (VICKERS, 1969), rats (BUTCHER and FUGO, 1967) rabbits (SHAVER and CARR, 1967) and pigs (HANCOCK, 1959; HUNTER, 1967). It has not yet been demonstrated in a mono-tocous species but the relatively high occurrence of triploidy in man, where there is no estrus exhibited, might be considered indirect evidence.

SALISBURY (1968) has repeatedly observed a correlation between time of storage of sperm prior to use in A.I. and estimated death rate of the embryos resulting from its use. Such observations are certainly suggestive of the increased presence of aberrations in stored gametes.

Finally, it can be shown that aberrant cells accumulate in certain mammalian tissues at specific times in the life of the individual (SWARTZ, 1956) or can be induced to accumulate (or not to) by appropriate hormone treatment. The physiological function of polyploid cells in liver, pancreas and other tissues is not known. Their appearance can be hastened by sex hormone treatment and can be repressed by castration or thyroidectomy (SWARTZ et al., 1960; SWARTZ and FORD, 1960).

It is clear from the foregoing that enough is now known about the etiology of various types of aberrations that a researcher can produce, almost at will, any type of aberration that he wishes to study. Not nearly enough is known however to be able to discern how the spontaneous incidence of chromosomal aberrations might be reduced by the animal breeder or clinical practitioner for economic or humanitarian purposes.

VI. — EFFECTS OF CHROMOSOME ABNORMALITIES

Just as with gene mutations, the effect of chromosome aberrations is harmful in the overwhelming majority of instances in that they reduce the fitness of their bearer in one or more of a variety of ways. Embryo and fetal deaths,

congenital malformation, intersexual phenotype, other forms sterility or reduced fertility are the costs of chromosome aberrations. Following is a brief review what is known regarding the incidence of chromosomal abnormalities and their effects in animal populations.

A. — *Chromosome aberrations and embryonic death*

In Table 3 are shown some representative estimates of the frequency of embryos afflicted with one or another type of chromosomal aberration. In man, well over 1,000 spontaneous abortions have been examined cytologically with the very surprising finding that about 1/5, i.e. 20 p. 100 possess an aberration that in all likelihood was the cause of death and abortion (Geneva Conference, 1966). The frequency of embryos with aberrations is higher in younger embryos (CARR, 1970) and it is now thought that among the very early abortions (those occurring in the first month of pregnancy) the incidence may be as high as 30-40 p. 100. If it is supposed that 20-25 p. 100 of successfully fertilized human eggs are subsequently aborted, then it can be calculated 4-5 p. 100 of all zygotes carry a lethal chromosome abnormality.

BEATTY and FISCHBERG (BEATTY, 1957 for review) looked at more than 3,000 mouse embryos and found about 2.5 p. 100 were heteroploid at 3 1/2 days. Similar findings were reported by VICKERS (1969). All of these die and are resorbed. In the rabbit (SHAVER and CARR, 1967) and pig (McFEELY, 1967)

TABLE 3

Incidence of chromosome aberrations in vertebrate embryos

TABLEAU 3

Fréquence des aberrations chromosomiques chez les embryons de vertébrés

Species	Product examined	Proportion with aberration	Source
Man	induced abortuses spontaneous abortuses	0.02 0.20	Report from Geneva conf. (1966)
Mouse.	3 ½ day embryos	0.025	BEATTY (1957)
Rat	11 day embryos	0.015	BUTCHER & FUGO (1967)
Rabbit	6 day blastocysts	0.10	SHAVER & CARR (1967) (1969)
Pig	11 day blastocysts	0.10	McFEELY (1967)
Chicken	16 hour embryos	0.01-0.12	MILLER et al. (1970)

although the scale of the experiments so far conducted has been small, it appears that about 10 p. 100 of preimplantation embryos are chromosomally defective and destined to die as a result.   From preliminary studies of cattle by McFEELY (1968) and HARPER (1970) it appears that the situation is similar.   The meaning of all this is that about one third of the prenatal pregnancy wastage of our farm animals is attributable to the presence of chromosome abnormalities.

We have found the chicken to be a very useful organism for more detailed studies of this phenomenon.   Different strains have different incidences of chromosomal aberrations in very early embryos, one broiler stock having an incidence of 12 p. 100 (MILLER et al., 1970).   Aberrant embryos in this stock include monosomy, trisomy, ZZW, haploidy, triploidy, tetraploidy and rearrangements. In addition simple and complex mosaics are common occurrences in this stock. In one experiment 15 of 34 aberrant embryos all derived from one hen.   This observation plus the clear difference in frequency among lines leads one to suppose there is a strong genetic component in the etiology.

The distribution of types of abnormalities seen in embryos of different species of animals is shown in Table 4.   It is seen that in aborted fetuses of man trisomy is the predominant abnormality and monosomy (mostly XO) and polyploidy are about half as frequent.   In the mouse, pig and rabbit polyploidy causes most

TABLE 4

Distribution of aberrations in embryos of five species

TABLEAU 4

Distribution des aberrations chez les embryons de cinq espèces

Category of aberration	Species				
	Man	Mouse	Pig	Rabbit	Chicken
Monosomy	32	23 aneuploid			3
Trisomy	63			1	
Haploid		6			4
Polyploid	34	91	7	7	6
Euploid mosaic . . .	12	9	1	3	30
Other	12	12	1	2	1
	153	141	9	13	44

Source: Man: Geneva Conference; Mouse: BEATTY, 1951; Pig: McFEELY, 1967; Rabbit: SHAVER and CARR, 1967; Chicken: BLOOM, 1969 and MILLER et al., 1970.

of the trouble while in chickens, euploid mosaics are by far the most important. Some of these differences reflect the type of material collected and the techniques used but there is no doubt that different mechanisms are involved in causing cytological errors in the different species. Very probably, an important job for those interested in livestock production is learning about the causes of and how to control the induction of spontaneously occurring polyploidy.

B. — *Chromosome aberrations in neonates*

Estimates of the frequency of neonatal animals with chromosome abnormalities are not available for any species other than man. Representative estimates of incidence of newborn babies with aberrations are listed in Table 5. As a general statement, the overall incidence can be taken as somewhere between 0.5 p. 100 and 1.0 p. 100. For technical reasons this can be thought of as the minimal frequency. The afflictions are divided rather evenly among three categories: sex chromosome aneuploidy, trisomy and rearrangements. Sex chromosome aneuploidy is higher in males owing to the fact that XXY is more viable and therefore has a greater probability of surviving to birth than does XO. Also XYY is not infrequent in neonatal samples properly ascertained. For the rest, there does not appear to be a difference between the sexes.

No similar surveys appear to have been conducted in animals. It is thought that the incidence would be lower because trisomy does not seem to be so frequent in animal populations. From the relatively few laboratories engaged in cytogenetic research with domestic animals, the whole array of expected sex chromosome aberrations are being turned up (see Table 6 for list). XX/XY chimerism has been seen in all the Ungulatas. XXX, XO, XXY and various mosaics containing these cell types have been reported by German, Scottish and New Zealand workers.

TABLE 5

Estimates of frequency of neonatal babies with chromosome aberrations

TABLEAU 5

Estimations des fréquences des nouveaux-nés porteurs d'aberrations chromosomiques

Aberration	Estimated frequency (%)	Source
Sex chromosome aneuploidy		
Males	0.32	LUBS and RUDDLE, 1970
Females	0.18	LUBS and RUDDLE, 1970
Trisomy	0.22	COURT BROWN et al., 1966
Rearrangements	0.14	LUBS and RUDDLE, 1970
Total	0.54-0.68	

What is needed now is a more systematic search for aberrant types and more complete study of those found so the various syndromes can be more precisely described and the epidemiological aspects defined.   In short, one wants to know

TABLE 6

Sex chromosome aberrations reported in farm animals and the Mouse

TABLEAU 6

Aberrations chromosomiques sexuels chez les animaux domestiques et chez la Souris

Sex chromosome Complement	Species	Source
XO	Mouse	RUSSELL et al., 1961 McLAREN, 1960 CATTANACH, 1962
	Pig	NES, 1968
XXX	Cattle	FINGER et al., 1969
	Cattle	SCOTT & GREGORY, 1965 RIECK et al., 1969
	Sheep	BRUÈRE et al., 1969
XXY	Pig	HARVEY, 1968 BREEUWSMA, 1968 GLUHOVSCHI et al., 1968
	Mouse	CATTANACH, 1961 McLAREN, 1960 RUSSELL & CHU, 1961
XYY	Mouse	CATTANACH & POLLARD, 1969
XX /XY Chimerism	Cattle Sheep Goats Pigs Mouse	MANY
Other mosaics & chimerics . .	Many seen in mouse	

what is causing them and what are the effects.   A comprehensive review of sex chromosome aneuploidy in animals was written by BEATTY (1964).

Along this line it should be noted that a number of aberrations can be experimentally produced in mammals now.   A number of structural aberrations were found in live born pigs derived from sperm that had been x-irradiated (ZARTMAN, FECHHEIMER and BAKER, 1969).   A total of 40 pigs was born of which 8 contained aberrations including translocations, inversions and deletions.   This sort of study is very promising both in terms of the genetic information that can be gained and by being able to produce at will aberrant animals for detailed anatomic and physiological study.

C. — *Chromosome aberrations and infertility*

From studies of man it has been learned that numerical aberration of the sex chromosomes are reasonably well tolerated in that they are compatible with survival although, with few exceptions, the bearers are sterile. Those causing complete sterility, e.g. XXY and its variants, are unlikely to be sources of serious loss of fitness in animal populations. However, carriers of structural aberrations are expected to have reduced fertility, so if such mistakes happen at all requently, they can be an important source of loss of reproductive fitness in the population. In mice, semi-sterility of males is a regular concomitant of translocation hetero-zygosis and most other chromosome anomalies (LYON and MEREDITH, 1966). In man, about 10 per cent of individuals presenting themselves to infertility clinics where karyotyping has been done have been found to carry a chromosome aberration (KJESSLER, 1965; McILTREE et al., 1966). It is not known, of course, what proportion of the total population this represents. In the data presented earlier, it was seen that about 0.14 per cent of neonatal babies are carriers of structural aberrations. A large proportion of these will have a reduced fertility or sterility.

The picture is even less clear in farm animals. It has been shown by KNUDSEN (1958, 1961a, 1961b) and other Swedish workers (HENRICSON and BÄCKSTRÖM, 1964) that some of the infertility of cattle and pigs is attributable to structural aberrations possessed by otherwise normal animals. Recent observations on infertile rams have disclosed translocations (BRUÈRE, 1969) as well as the sex chromosome aneuploidy referred to earlier (BRUÈRE et al., 1969). It is too early to attempt an assessment of the importance of chromosome aberrations in causing reduced fertility of farm animals. It is clear however that they are a factor to be taken into account.

VII. — POLYMORPHISMS

The number of chromosome polymorphisms that have been found in farm animals is very surprising in view of the relatively small numbers of animals that have been studied. Centric fusions, i.e. translocation and loss of a centro-mere, have been reported in cattle (GUSTAVSSON, 1969; RIECK et al., 1968), pigs (McFEE et al., 1966; RARY et al., 1968; McFEE and BANNER, 1969) and goats (SOLLER et al., 1966). Each of these variants is thought to be present as a stable polymorphism in the population where it was found. It is probable, but not at all certain, that they became established through genetic drift but to be maintained they must not be severely detrimental. In any case, from this plus other evidence, it does appear that the ungulate chromosomes are particularly prone to centric fusion and it is to be anticipated that more will be found. They should be thoroughly studied not only because they are of interest in their own right but because important insights applicable to similar conditions in man can be gained. An important source of duplication in man is unbalanced gametes

produced by translocation carriers.   Much can be learned in our animals regarding segregation mechanisms in translocation heterozygotes that would be extremely useful to human geneticists.

Need for more work

It is clear from this brief review that cytogenetic research in farm animals has only just begun but promises to contribute answers to some important problems of livestock breeding and genetics.   There are a number of things needing to be done immediately.

Surveys should be conducted in a number of places to determine the frequencies and types of chromosome aberrations in domestic animals.

Attempts should be made to ascertain the expected effects of given aberrations on their carriers and on populations as a whole.

The importance of the various possible causes of aberrations should be carefully determined for each of the groups of farm animals.   When this is done, it is probable that their frequencies can be materially reduced by adopting appropriate husbandry practices.   In addition, when the causes are known, various aberrations can be experimentally produced and their possible beneficial aspects viewed by geneticists.

Chromosome abnormalities in domestic animals will be very useful as models of their counterparts in man.   The mouse, for technical reasons, has a limited usefulness in this respect.

Once this background of information is established, it no doubt will be found that cytogenetic investigations of farm animals have much to contribute to enhancing efficiency of animal production.

RÉSUMÉ

CONSIDÉRATIONS CYTOGÉNÉTIQUES EN SÉLECTION ANIMALE

Cet article résume quelques aspects fondamentaux de la cytogénétique, les travaux faits sur les animaux domestiques et une évaluation des effets connus des aberrations chromosomiques sur les populations animales.

Ces dix dernières années le nombre chromosomique de base ainsi que des caractéristiques précises du caryotype ont été établis avec sûreté pour tous les animaux domestiques.

Des variants intra-spécifiques, c'est-à-dire des polymorphismes chromosomiques ont été mis en évidence chez le Bœuf, le Porc et la Chèvre.

Il existe deux types primaires d'aberrations. Le premier type est une altération du nombre des chromosomes, on l'appelle l'hétéroploïde. Le second type, qui consiste dans le réarrangement de segments chromosomiques, intervient après le bris des chromosomes.

Les facteurs responsables de la fréquence d'apparition des aberrations sont à la fois génétiques et non génétiques. Dans la seconde catégorie, les plus importants sont certains groupes de virus, de produits pharmaceutiques ou chimiques, les radiations ionisantes, l'âge des gamètes, principalement l'ovule et, chez certains animaux, l'âge de la mère.

La présence d'aberrations chromosomiques rend apparemment compte, pour une large part, de la mortalité embryonnaire des mammifères et des oiseaux. Chez l'homme, la proportion des avortons expulsés spontanément et porteurs d'anomalies chromosomiques serait peut-être d'un tiers, ce qui est énorme. Chez le porc, des études préliminaires semblent indiquer que la situation est sans doute fort semblable.

Très peu de travaux de cytogénétique ont été faits sur le bétail présentant des défauts congénitaux, bien qu'un certain nombre d'aberrations aient été signalées. Chez l'homme, un peu moins de 1 p. 100 des nouveaux-nés portent des anomalies chromosomiques.

Quelques aberrations à effets somatiques légers, voire nuls, peuvent être cause de fertilité réduite ou de stérilité des porteurs. Des cas isolés ont été décrits sans qu'une évaluation statistique de leur incidence ait été faite.

Chez le Bœuf, le Porc et la Chèvre on a trouvé des fusions centriques et des travaux sont en cours pour préciser leur effet sur les valeurs adaptatives des populations où elles sont apparues.

Pour conclure, on peut dire que les aberrations chromosomiques réduisent sérieusement la productivité des animaux qui les portent. Il faudra encore préciser l'ampleur de ces pertes et trouver le moyen de les réduire.

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