Cytogenetic screening of livestock populations in Europe: an overview

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Abstract

Clinical animal cytogenetics development began in the 1960's, almost at the same time as human cytogenetics. However, the development of the two disciplines has been very different during the last four decades. Clinical animal cytogenetics reached its 'Golden Age' at the end of the 1980's. The majority of the laboratories, as well as the main screening programs in farm animal species, presented in this review, were implemented during that period, under the guidance of some historical leaders, the first of whom was Ingemar Gustavsson. Over the past 40 years, hundreds of scientific publications reporting original chromosomal abnormalities generally associated with clinical disorders (mainly fertility impairment) have been published. Since the 1980's, the number of scientists involved in clinical animal cytogenetics has drastically decreased for different reasons and the activities in that field are now concentrated in only a few laboratories (10 to 15, mainly in

Europe), some of which have become highly specialized. Currently between 8,000 and 10,000 chromosomal analyses are carried out each year worldwide, mainly in cattle, pigs, and horses. About half of these analyses are performed in one French laboratory. Ac- curate estimates of the prevalence of chromosomal abnormalities in some populations are now available. For instance, one phenotypically normal pig in 200 controlled in France carries a structural chromosomal rearrangement. The frequency of the widespread 1;29 Robertsonian translocation in cattle has greatly decreased in most countries, but re- mains rather high in certain breeds (up to 20–25% in large beef cattle populations, even higher in some local breeds). The continuation, and in some instances the development of the chromosomal screening programs in farm animal populations allowed the implementation of new and original scientific projects, aimed at exploring some basic questions in the fields of chromosome and/or cell biology, thanks to easier access to interesting biological materials (germ cells, gametes, embryos …).

The identification of various chromosomal rearrangements in livestock species in the 1960's and 1970's (e.g. Robertsonian translocations in cattle – Gustavsson and Rock- born, 1964; Popescu, 1971; Stranzinger and Forster, 1976; reciprocal translocations in pigs – Henricson and Bäck-ström, 1964; Popescu and Legault, 1979) clearly associated with several clinical conditions such as intersexuality and congenital malformations as well as reproductive dysfunction (reduction of the fertility/prolificacy of the carrier animals and/or of their mates – Gustavsson, 1969, 1971; Refs- dal, 1976; Popescu et al., 1984) led to the establishment of many animal cytogenetics laboratories particularly concentrated in Europe. These laboratories were created almost exclusively within academic research institutions with a focus on basic research. Under the leadership of several pioneers (e.g. Ingemar Gustavsson in Sweden, Paul Popescu in France, Gerald Stranzinger in Switzerland, Parvathi Basrur in Canada, and many other prominent researchers world- wide), the field of domestic animal cytogenetics grew rap- idly during this period. The adaptation of some specialized chromosome staining techniques developed in human cytogenetics laboratories (e.g. banding techniques – Seabright, 1971; Dutrillaux et al., 1973) allowed rapid progresses in the acquisition of knowledge of the chromosomes of several animal species. An international study group with the man- date of standardizing the karyotypes of most farm animal species (including cattle, sheep, goats, pigs, horses, rabbits, swine and cats) was created in 1976 during the Reading

Conference (Ford et al., 1980). The Reading standard formed the basis for all subsequent nomenclature reports (e.g. Gustavsson, 1988; ISCNDA1989, 1990; Iannuzzi, 1996; Popescu et al., 1996; Bowling et al., 1997; Ansari et al., 1999), al- though some discrepancies in bovid nomenclatures were identified and, for the most part, solved when both Q/G and R-banding techniques were combined with molecular markers (FISH) (Hayes et al., 2000). These preliminary karyotypes served as the basis for the construction of the most recent nomenclature of bovids (ISCNDB 2000, 2001) where cattle, sheep and goat autosomes were reported using one common chromosome nomenclature.

The research activity of the laboratories involved in animal cytogenetics reached a high level throughout the 1980's and several systematic chromosomal screening programs were initiated, mainly in continental European countries. As a result, a large number of chromosomal rearrangements were identified and reported in many scientific publications (see the reviews of Chowdhary, 1998 and Fries and Popescu, 1999, for pig and cattle, respectively). Several comprehensive review papers and textbooks were also published during this period (e.g. Gustavsson, 1980; Popescu et al., 1984; King, 1990; Long, 1991) which formed the primary reference sources for clinicians and researchers alike. In addition, the characterization of some original and rare chromosomal

rearrangements led to particularly interesting scientific developments (e.g. the X;autosome reciprocal translocation identified in cattle by our Canadian colleagues – Basrur et al., 1992, 2001; Rho et al., 2007) related to the establishment of physical gene maps and understanding of basic developmental phenomena such as X-chromo- some inactivation.

However, since the beginning of the nineties, a clear de- cline of these 'clinical' animal cytogenetics activities (identification of original chromosomal rearrangements and study of their clinical consequences in farm animals) has been noticed. The reduction in the number of scientific publications and doctoral theses in this field is one objective indicator of this evolution. Several explanations can be pro- posed. First, some groups initially involved in clinical animal cytogenetics were reorientated towards new scientific objectives (e.g. towards genome mapping projects). On the other hand, the eradication of particular chromosomal re- arrangements in some populations made the continuation of the corresponding animal screening programs no longer justified. Finally, the retirement of some 'historical leaders' in our field and the dissolution of their laboratory groups also contributed to the decline. Currently, the number of countries in which significant clinical animal cytogenetics activities are carried out

is very limited (less than ten). Most are located in Europe. Nonetheless, new initiatives adopted by several breeding and artificial insemination companies (e.g. in pigs, some companies are now interested in systematically analyzing all purebred boars at the selection level, instead of only hypoprolific boars at the production level) as well as the improvement of the techniques used in the laboratories (use of new software allowing semi-automatic karyotyping, and therefore a dramatic augmentation in the productivity of the labs) has generated a very significant in- crease in the number of analyses carried out in some laboratories. This was clearly the case in France. In pigs for in- stance, as illustrated below, the annual number of analyses carried out increased 20-fold in only 15 years, and the number of original chromosomal rearrangements identified in this species during the 1996–2007 period in only one laboratory alone is larger than the total number of rearrangements published worldwide during the previous 30 years. The development of the few remaining laboratories allowed us to reaffirm the interest of 'clinical cytogenetics' in farm animal species, and opens new scientific opportunities in that field.

In the current paper, we present an overview of the main cytogenetic screening programs carried out in farm animal species in some currently active European cytogenetics laboratories, and summarize the main results and scientific outcomes obtained within these programs.

Description of the main European screening programs

The French programs
Only one laboratory is currently involved in large scale animal cytogenetics screening programs in France. This laboratory, located at the National Veterinary School of Toulouse, was created in 1968 by Prof. Roland Darré, assisted by Mrs Hélène Berland. It is in this laboratory that the most widespread chromosomal rearrangement in cattle (the 1;29 Robertsonian translocation) was identified for the first time in France, both in the Blonde d'Aquitaine and Limousine beef cattle breeds (Darré et al., 1972a). Screening for this particular rearrangement and diagnosis of the bovine freemartin syndrome (Darré et al., 1972b) were the main activities of the lab for many years. At the beginning of the 1980's, these programs were extended to include the chromosomal screening of wild pig populations (wild and breeding populations). Being located in a veterinary faculty, the laboratory also provided chromosomal analyses for hospital patients (mainly horses and dogs). However, the most important and recent evolution corresponded to the implementation of a

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systematic chromosomal screening program in swine. The pig clinical cytogenetics activity started in Toulouse at the beginning of the 1990's. At that time, only hypoprolific boars were karyotyped (less than 50 analyses per year). From the middle of the nineties, the majority of the French pig breeding companies (and more recently other European breeding and artificial insemination companies) started to ask for a systematic screening of all their purebred boars before using them in artificial insemination (AI) centers. With more than 2,000 pigs karyotyped annually, pig cytogenetics is currently the main component of the activity of the French laboratory (Ducos et al., 2007). The results obtained during the last five years in France are sum- marized below.

Screening programs in cattle.

The historical 1;29 Robertsonian translocation screening program in cattle has continued without interruption until now. This program is based on a statutory obligation for the breeders to control all the young bulls before being used in AI centers. This obligation concerns all the breeds considered as 'non-free' of the translocation at the beginning of the 1980's, i.e. mainly the beef cattle breeds (Charolaise, Limousine, Blonde d'Aquitaine) and some dairy cattle breeds (e.g. Montb liarde). The analyses carried out only aim at detecting the 1;29 Robertsonian translocation. Therefore, simple conventional chromosome staining techniques are routinely used (GTG-banding techniques are not used systematically). About 1,300 individuals are examined annually. More than 50% of the analyses concerned animals from the Blonde d'Aquitaine breed. General statistics concerning this particular screening program for the last five years are presented in Table 1

Table 1. Overview of the French 1;29 Robertsonian translocation screening program in cattle

Breed[a]	2002		2003		2004		2005		2006		Total per breed (% 1;29)
	N[b]	1;29 (%)[b]	N	1;29(%)	N	1;29 (%)	N	1;29 (%)	N	1;29 (%)	
BA	524	50 (9.5)	624	53 (8.5)	834	56 (6.7)	703	73 (10.4)	762	40 (5.2)	3,447 (7.9)
LI[c]	402	100 (24.9)	209	15 (7.2)	158	7 (4.4)	143	2 (1.4)	146	6 (4.1)	1,058 (12.3)
CH	125	0	127	1 (0.8)	327	3 (0.9)	287	3 (1.0)	224	6 (2.7)	1,090 (1.2)
MB	208	0	173	0	202	0	136	0	180	0	899 (0)
INRA95	71	1 (1.4)	81	10 (12.3)	86	1 (1.2)	79	0	43	0	360 (3.3)
Other breeds	14	0	6	0	14	0	9	0	14	0	57 (0)
Total	1,344		1,220		1,621		1,357		1,369		6,911

[a] BA = Blonde d'Aquitaine; LI = Limousine; CH = Charolaise; MB = Montbéliarde; INRA95 = synthetic line; other breeds: Aubrac, Gasconne, Bazadaise.
[b] N: total number of animals controlled; 1;29: animals carrying the 1;29 Robertsonian translocation (both heterozygotes and homozygotes).
[c] The values reported in the table for the Limousine breed don't correspond to the real prevalence of the 1;29 Robertsonian translocation in this population (see the text).

Only the analyses carried out for males (about 90% of the total number of analyses) are

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considered in Table 1. Indeed, the females examined are generally daughters (or sisters or dams) of carrier bulls. The frequency of female carriers is therefore higher than the one estimated for males. For in- stance, in 2006, 21.5% of the females analyzed carried the translocation in the Blonde d'Aquitaine breed (121 females), 46.8% in the Limousine breed (32 females) and 25% in the Charolaise breed (24 females).

On the other hand, the values indicated in Table 1 for the Limousine breed for years 2002–2003 do not correspond to the real prevalence of the translocation in this population. Indeed, a carrier bull was accidentally used at the beginning of year 2000. A large scale eradication program was later carried out at the request of the breeding association concerned. During years 2001, 2002 and 2003, many offspring of this bull were checked, which explains the relatively high frequencies of carrier animals during this period.

The higher prevalence of the 1;29 translocation is ob- served in the Blonde d'Aquitaine breed (about 8% for the 2002–2006 period). The estimated frequency sharply de- creased from 1990 to 1997, then became almost stable after that date (Fig. 1). This can be explained by the fact that only future AI bulls are systematically analyzed. Natural mating bulls generally escape the screening program, whereas this reproduction mode still represents more than 50% of the calves born in this breed. An effort should be made in this direction to eradicate the rearrangement in the Blonde d'Aquitaine population.

Complementary GTG-banding analyses are carried out when apparently abnormal chromosomes are identified using conventional staining techniques. This led to the detection of six original chromosomal rearrangements during the last five years: one 1;7 Robertsonian translocation, two mosaics for Robertsonian translocations (21;29 and 3;12), one pericentric inversion of chromosome 29 and one reciprocal translocation (7p+;7q–) in the Blonde d'Aquitaine breed; one reciprocal translocation (1q–;15q+) in the Charolaise breed. Moreover, one 61,XXY karyotype and one 60,XX/90,XXY chimeric karyotype were identified in the Blonde d'Aquitaine breed, as well as one 60,XY/61,XYY chimeric karyotype in the Montbéliarde breed (all found in phenotypically normal young bulls). Finally, the analyses carried out in hypofertile bulls (Ducos et al., 2000) allowed the detection of two original reciprocal translocations, one involving chromosomes 9 and 12, and the other one chromosomes 8 and 21.

About 250 analyses are carried out each year in young females born co-twin to males (diagnosis of the freemartin syndrome; 1,253 analyses since year 2002). Globally, 86.4% of these females presented an XX/XY blood chimerism (13.6% had normal XX

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karyotypes).

Screening program in pigs.

As mentioned above, the great majority (about 90%) of the pigs currently screened in the Toulouse laboratory are young purebred boars waiting for an approval for use in artificial insemination (AI) centers. At the same time (2002–2007 period), 20–70 hypoprolific boars were screened annually. This represented only 3% of the total number of analyses carried out in the laboratory during this period. Finally, 7% of the analyses carried out in Toulouse during the 2002–2007 period concerned animals belonging to the families of carrier individuals: parents, (half) sibs, offspring.

The analyses carried out aim at detecting all types of chromosomal rearrangements. Therefore, GTG banding is systematically used. For some particular rearrangements the presumptive chromosomes involved and/or the location of break- points on the chromosomes were verified using molecular cytogenetic techniques (see for instance Ducos et al., 2002).

As of July 1st, 2007, 15,114 pigs have been karyotyped in the Toulouse laboratory with the great majority of the animals belonging to French breeding companies. As shown in Fig. 2, the number of pigs controlled has increased regularly for more than 15 years. None of these analyses were mandatory.

In total 115 original structural chromosomal rearrangements have been identified in the laboratory, including 78 since 2002 (Table 2).

Table 2. Distribution of the constitutional structural chromosomal rearrangements identified in France during the 2002–2007[a] period in pigs

	Reciprocal translocations			Inversions		Robertsonian translocations	Total
	Hypoprolific boars	Routine boars[b]	Females	Routine boars	Females	Routine boars	
2002	4	8	1	3			16
2003	4	5					9
2004	4	4	3	1	1		13
2005	4	6	1	2		1	14
2006	6	6	2	1		2	17
2007[a]		7	1	1			9
Total	22	36	8	8	1	3	78

[a] End of the period: July 1st 2007.
[b] Young purebred boars waiting for an approval for use in artificial insemination (AI) centres.

Sixty-six (out of 78) were reciprocal translocations and nine were peri- or paracentric inversions. For the first time since the beginning of the screening program, after

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more than 11,000 pigs were karyotyped, one Robertsonian translocation was identified in 2005 and two others in 2006. Also in 2006 for the first time one reciprocal translocation involving a sex chromosome was identified in an azoospermic boar: t(Y;14)(q10;q11) (Pinton et al., 2007).

The estimated prevalence of balanced structural chromosomal rearrangements in a sample of more than 7,700 young boars karyotyped before service was 0.47% (Ducos et al., 2007). To the best of our knowledge, the pig (*Sus scrofa domestica L.*) is the only mammalian species other than humans and laboratory mice for which an accurate estimate of the prevalence of structural chromosomal rearrangements is available.

Twenty-two of the 78 rearrangements described since 2002 were identified in hypoprolific boars. All were reciprocal translocations. The estimated effect of the chromosomal rearrangements identified in hypoprolific boars since the beginning of the program (decrease of the average litter size of the mates) varied between 10 and 70% (40% on average). One translocation, the t(3;16)(q23;q22), was responsible for malformations in some of the offspring (cleft palate – Ducos et al., 2004).

Twelve cases of chimerism (XX/XY in 11 individuals, XY/XXY in one individual) were also diagnosed. Two of these were hypoprolific boars, and three were intersex animals.

Screening program in other species.
Besides cattle and swine, the Toulouse laboratory is involved in another large scale control program aimed at detecting 'domestic pig wild pig hybrids' (having 37 chromosomes – Darré et al., 1992). Since 2002, the total number of analyses carried out in that field is 2,257. 427 animals (i.e. 18.9%) were hybrids (37 chromosomes). This frequency was halved in 15 years.

The number of analyses carried out in horses and dogs since 2002 is very low. Only 31 horses were karyotyped, including 29 sterile and two intersex mares. Six 63,X cases and seven 64,XY cases were diagnosed among the 29 sterile mares. All the other animals had normal karyotypes. One intersex individual presented a 64,XX karyotype, whereas the other one had a 64,XY karyotype. Only seven dogs were karyotyped. All were intersex individuals with three having a 78,XX karyotype, and four a 78,XY karyotype.

The Italian program
The Italian cytogenetic screening program concerns only bovine populations. It mainly focuses on meat breeds (mostly Chianina, Marchigiana, Romagnola and Maremmana breeds) investigated at the cytogenetics laboratory of the Animal Production Institute, University of Milan. Cytogenetic investigations have been performed in other breeds by the cytogenetic laboratories

of both Milan and Naples (CNR-ISPAAM). Most of the animals investigated (92.6%) have been males which underwent cytogenetic analysis at about four months of age, before breed performance testing. Almost all animals were studied by using conventional Giemsa staining to detect numerical and structural (Robertsonian – rob-, and evident reciprocal – rcp-translocations) chromosome abnormalities, as well as XX/XY chimerism. Additional studies have been performed in animals carrying or suspected of carrying chromosome abnormalities. For these animals, both C- and R-banding techniques, as well as FISH-map- ping studies were performed for precise identification of the chromosomes and chromosome regions involved in the abnormalities (Iannuzzi et al., 2001a, b, c; Molteni et al., 2007). Table 3 reports all investigated breeds (and animals) and all types of chromosome abnormalities identified during the last 15 years.

Some animals were found to carry both rob(1;29) and XX/XY chimerism. For these animals, we prefer to list them in XX/XY chimerism because this syndrome is responsible for more deleterious effects on fertility than rob(1;29), especially in females, also because few animals were found to be carriers of both types of chromosome anomalies.

20,030 cattle were investigated during the last 15 years (Table 3).

Table 3. Results of the chromosomal screening program carried out in Italy for ten cattle breeds during the last 15 years

Breed	Animals N (F)[a]	rob(1;29)[b] HT	HM	HT+HM (%)	Rob N/ chroms[c]	Rcp N/ chroms[d]	Inv N/ chroms[e]	XX/XY[f]	XXX/XY[f]	XXY	frag[g]	Carriers N (%)
Bruna	57 (18)	2		2 (3.5)	3/1;8;9							5 (8.8)
Chianina	8,303 (223)	119		119 (1.4)	1/Y;9 1/11;21			66 (M)	1 (M)	6	3	197 (2.4)
Friesian	136 (72)	1		1 (0.7)				6 (M), 15 (F)				22 (16.2)
Grey Alpine	580 (76)	5		5 (0.9)	45/26;29	4/1;5		4 (M)				58 (10.0)
Marchigiana	5,522 (538)	618	26	644 (11.7)	6/14;17 1/13;19			45 (M), 1 (F)	1 (M)	1		699 (12.7)
Mucca Pisana	122 (109)	3		3 (2.5)								3 (2.5)
Podolian	256 (75)	29	1	30 (11.7)			12/Y					42 (16.4)
Romagnola	3,876 (256)	477	26	503 (13.0)				18 (M)				521 (13.4)
Maremmana	1,057 (70)	183	16	199 (18.8)				14 (M), 1 (F)				214 (20.2)
Ottonese	121 (55)	23		3 (2.5)				1 (F)				4 (3.3)
Total	20,030	1,440	69	1,509 (7.5)	52	9	12	71	2	7	3	1,765 (8.8)

[a] N = total number of animals controlled (F = females).
[b] Carriers of rob(1;29); HT = heterozygotes; HM = homozygotes.
[c] Rob = Robertsonian translocations; N = number of carriers; chroms = chromosomes involved.
[d] Rcp = reciprocal translocations; N = number of carriers; chroms = chromosomes involved.
[e] Inv = inversions; N = number of carriers; chroms = chromosomes involved.
[f] Chimeric karyotypes; in brackets: M = males, F = females.
[g] Frag = fragile X.

About 90% of these analyses concerned only three meat breeds (Chianina, Marchigiana and Romagnola). In the most investigated breed (Chianina), the frequency of rob(1;29) carriers was very low (1.4%) although this breed is closely related to others where the translocation was found in appreciable frequencies (Marchigiana and Podolian). On the other hand, two new reciprocal translocations, involving chromosomes Y and 9, and 11 and 21, respectively, were identified in this breed, as well as a substantial number of XX/XY males. In the two other highly investigated breeds (Marchigiana and Romagnola), the frequency of rob(1;29) carriers is much higher (11.7 and 13.0%, respectively). Twenty-six homozygotes were even found in these two breeds. Two original Robertsonian translocations, involving chromosomes 14 and 17, and 13 and 19, respectively, were also identified in the Marchigiana breed. Among all investigated Italian breeds, the one with the highest percentage of carriers of rob(1;29) is Maremmana (18.8%). This breed is the closest relative of the ancient Podolian cattle living in central Europe. In this latter breed, raised in southern Italy, the frequency of rob(1;29) carriers is thus logically rather high (11.7%). The number of investigated animals in the other breeds (Bruna, Grey Alpine, Mucca Pisana and Ottonese) is relatively limited. The frequencies of rob(1;29) carriers are low (0.9–3.5%). Yet, a new Robertsonian

translocation involving chromosomes 26 and 29 was found in 45 animals (7.8%) of the Grey Alpine breed. Among the various Robertsonian translocations found so far in just a few animals, the rob(26;29) (De Giovanni et al., 1979; Di Meo et al., 2000) has the highest frequency after the rob(1;29). Moreover, BTA29 has been involved in both translocations, although these two rearrangements had different evolutionary origins (re- viewed in Di Meo et al., 2006).

The frequency of rob(1;29) has decreased during the last 15 years due to systematic elimination of male carriers, al- though this chromosomal rearrangement is still present with appreciable frequencies in some breeds (Table 3), due to maternal transmission. The high frequency of carriers of chromosome abnormalities also observed in the Friesian breed (16.1%, almost all XX/XY chimeric bulls), suggests that systematic cytogenetic screening would also be relevant in this breed, which is the most common in Italy among the milk breeds. However, the Italian Breeder's Association still does not require cytogenetic investigations for milk breeds.

The effects of Robertsonian translocations on the reproductive performance of female carriers have been investigated in the Grey Alpine and Marchigiana breeds. In the Grey Alpine breed, reproductive parameters of cows heterozygous for the rob(26;19) (sired by a carrier bull) were com- pared to the performance of cows sired by the same bull but

having normal 2n = 60 karyotypes. The same experiment was carried out in the Marchigiana breed, but for the classical rob(1;29). The results showed a strong decrease of all reproductive parameters in the heterozygotes. For instance, the percentage of negative services was significantly higher in the carrier cows than in the ones having normal karyotypes (30.2 vs. 22.2% in the Grey Alpine breed, 39.9 vs. 29.6% in the Marchigiana breed). This was also the case for the percentage of irregular returns to heat and the average (inter)calving interval (414 days vs. 381 in the Grey Alpine breed, 434 days vs. 412 days in the Marchigiana breed). These results are in good agreement with results obtained in Hungary (see section 'The Hungarian programs'), and justify the continuation of the screening activities.

The Romanian program

Considering the great interest in the artificial insemination of cattle in Romania, chromosomal analysis mainly concerns sires. The investigated bulls mostly belonged to the Romanian Spotted, Romanian Black Spotted and Brown breeds which are the most common in the country. Overall 2,576 bulls of Romanian cattle breeds have been investigated during the last 20 years. Different types of abnormalities were identified (Table 4).

Table 4. Results of the chromosomal screening program carried out in Romania for three cattle breeds during the last 20 years

Breed	Animals N[a]	rob(1;29) N (%)[b]	Rob N/chroms[c]	TF N/Chrom[d]	XX/XY N[e]	Sex reversal N	Carriers N (%)[f]
Romanian Spotted	1,357	11 (0.8)	1/14;20		13	1 (XX male)	26 (1.9)
Romanian Black Spotted	932		1/3;27	2/?	12		15 (1.6)
Brown	287	2 (0.7)	1/5;23				4 (1.4)
			1/11;21				
Total	2,576	13 (0.4)	4	2	25	1	45 (1.7)

[a] N = total number of animals controlled (all males).
[b] Carriers of rob(1;29); N = number of carriers.
[c] Rob = Robertsonian translocations; N = number of carriers; chroms = chromosomes involved.
[d] TF = tandem fusion; N = number of carriers; chroms = chromosomes involved.
[e] XX/XY chimerism.
[f] Total number of carriers.

The most frequent abnormality in the screened population was XX/XY chimerism (25 cases). Because the opinions concerning the reproductive performance of the male carriers are relatively contradictory (Padula, 2005), their elimination is recommended.

Five original centric fusions involving chromosomes of different pairs (1;29, 3;27, 5;23, 11;21 14;20) have also been described (Nicolae and Popescu, 2001), but the most com- mon was rob(1;29): 13 cases were identified, including 11 in the Romanian

Spotted breed alone. This situation might be explained, on the one hand, by the high number of animals studied, or, on the other hand, by the massive import of animals and frozen semen from the Simmental breed. The carriers of Robertsonian translocations, for which negative effect on reproduction could be demonstrated, were eliminated from the herds.

One tandem fusion was identified in the Romanian Black Spotted breed (Nicolae and Livescu, 1995). The consequences regarding the reproductive performance were similar to that of the 1;29 translocation and the male carrier was there- fore eliminated from the AI center.

A sex reversal constitution was identified in a bull be- longing to the Romanian Spotted breed. Even if the repro- ductive behavior was seemingly normal, the presence of a female karyotype in this bull (XX male) justified its elimination.

A dicentric chromosome was observed in the Romanian Black Spotted breed (Nicolae, 2003). This abnormality is very rare and previously had only been identified in humans, with a very low frequency (0.082% in the general population; Lloyd et al., 1992). It is particularly interesting to mention that the carrier of this abnormality was born seven months after the Chernobyl nuclear accident which affected Romania. This seemed to be the only explanation for this particular chromosomal rearrangement,

even if a mutation during the pregnancy should have resulted in a mosaic embryo.

The Polish programs

Screening programs in cattle. In 1989, the Ministry of Agriculture issued a directive to cytogenetically evaluate all young bulls undergoing animal breeding evaluation. Following this directive, five new local cytogenetic laboratories were established. However, the leading laboratory was the one chaired by Prof. Ewa Slota at the National Institute of Animal Production. The latest summary of this program was presented by Sysa et al. (2002) at the 15th European Cytogenetic Colloquium on Cytogenetics and Gene Mapping in Sorrento, Italy. Altogether, over 7,500 young bulls were evaluated and among them 89 (1.2%) were carriers of the XX/XY chimerism and 35 (0.47%) carriers of a centric fusion, mainly 1;29 in the Charolais breed (Rejduch et al., 1994) and one case of 5;22 fusion (Słota and Switonski, 1992). Also one case of the 61,XYY trisomy was diagnosed. Recently, new sex chromosome aneuploidies in young bulls were also described in the Holstein-Friesian breed: 61,XYY (Krumrych et al., 2002) and 61,XXY (Krumrych, 2003).

Since cytogenetic evaluation has also been conducted outside the national program, more cases of abnormal karyotypes have been detected (Table 5).

In addition, an indigenous cattle breed (Polish Red) was analyzed to estimate the incidence of abnormal karyotypes in this population (Slota et al., 2004). Among 451 animals investigated, three appeared to be carriers of the 1;29 Robertsonian translocation and four were carriers of the 60,XX/60,XY lymphocyte chimerism. Cytogenetic analysis was also applied to determine the etiology of congenitally malformed calves (polymelia and amelia). In both cases, frequent chromatid and chromosome breaks were observed (Szczerbal et al., 2006; Nowacka et al., 2007).

Screening programs in sheep and goats. These species are not systematically screened in

Poland, however, a large number of animals have been analyzed (Table 6). Leukocyte chimerism (XX/XY) appeared to be the predominant chromosome abnormality.

Screening programs in pigs. Extensive cytogenetic evaluation of pigs has been performed at the National Institute of Animal Production in Balice. Two groups of animals were considered: (a) random group of 1,600 animals and (b) 258 boars from AI stations. Altogether six cases of reciprocal translocations and one case of a pericentric inversion were identified (Table 7). In earlier studies, a case of paracentric inversion of chromosome 8 was also found (Switonski, 1991).

Table 5. Abnormal karyotypes detected in Polish crossbred cattle (Polish Black and White or Polish Red and White Holstein Friesian) subjected to cytogenetic evaluation due to fertility problems

Sex	Animals with abnormal karyotype	Karyotype	Reference
Female	1	60,XX, inv(X)(p12;q24)	Switonski, 1987
Male	6	61,XXY or 60,XY/61,XXY	Slota et al., 1982, 2003; Sysa and Slota, 1984; Danielak et al., 1988; Krumrych, 2003
Male	1	60,XY/61,XYY	Jaszczak et al., 2003
Male	2	60,XX/59,XY, rob(13;24)	Slota et al., 1988
Female (quintuplets)	3		
Female	87	60,XX/60,XY	Reiduch et al., 1999, 2000
(freemartins)	19		Nowacka et al., 2004

Table 6. Cytogenetic surveys of sheep and goats bred in Poland

Species	Total number of animals	Chromosome abnormalities[a]	Number of cases	Reference
Sheep	970 (random group of 50% females and 50% males)	rcp(2p+;3p–)	1	Slota et al., 1986
		54,XX/54,XY	33 (3.4%)	Sysa et al., 1996; Rejduch et al., 2004
	454 (females of Leine breed, originating from heterosexual twins)	54,XX/54,XY	23 (5.1%)	Szatkowska and Switonski, 1996
	104 (Booroola females and males originating from 48 heterosexual litters)	54,XX/54,XY	32 (30.8%)	Keszka and Jaszczak, 1996
Goat	130 (random group of 70% females and 30% males)	60,XX/60,XY	4 (3.1%)	Rychlik et al., 2005

[a] rcp = Reciprocal translocation.

Table 7. Large scale cytogenetic survey of pigs bred in Poland

Population	Total number of analysed animals	Chromosome rearrangements[a]	References
Random	1,600 (approx. 50% males and 50% females)	rcp(8;14)(p21;q25) rcp(7;13)(p13;q46) rcp(1;5)(p21;q21) rcp(9;14)(p14;q23) inv(1)(p22;q11)	Danielak-Czech et al., 1997 Rejduch et al., 2003 Danielak-Czech et al., 1996
AI boars	258	rcp(10;13)(q11;q11)	

[a] rcp = Reciprocal translocation; inv = inversion.

Screening programs in horses. A cytogenetic survey of 500 young horses was performed recently by Bugno et al. (2007a). This analysis revealed that the incidence of X mono- somy in mares reached 3%, but no abnormalities were found in males (Table 8). Another group of mares were subjected to cytogenetic investigations due to fertility problems. Survey of such mares was performed by two groups (Table 8).

Table 8. Cytogenetic surveys of horses bred in Poland

Population	Total number of animals	Chromosome abnormalities	Number of cases	References
Young horses (random group)	272 (females)	63,X	1 (0.4%)	Bugno et al., 2007a
		63,X/64,XX	7 (2.6%)	
		64,XX/65,XX,+31	1 (0.4%)	
		64,XX/64,XY	1 (0.4%)	
	228 (males)	none	0 (0%)	Bugno et al., 2007a
Females with fertility problems	215	65,XXX	1 (0.5%)	Bugno et al., 2003c
		63,X	1 (0.5%)	Bugno et al., 2003b
		63,X/64,XX	14 (6.5%)	Bugno et al., 2003b; Bugno et al., 2005; Bugno et al., 2006
		63,X/64,XX/65,XXX	3 (1.4%)	Bugno et al., 2006; Bugno et al., 2007b
		64,XX/65,XXX	1 (0.5%)	Bugno et al., 2006
		64,XX/65,XX,+Xp	1 (0.5%)	Bugno and Słota, 2007
		64,XY – sex reversal	1 (0.5%)	Bugno et al., 2003a
		64,XX/64,XY	2 (0.9%)	Bugno et al., 1999
		63,X/64,XX/65,XX,+Xq	1 (0.5%)	
		63,X/64,XX/65,XX,+delY	1 (0.5%)	
Females with fertility problems	244	63,X	3 (1.2%)	Parada et al., 1999
		64,XX/63,X	4 (1.6%)	Bugno et al., 2001
		64,XX/64,XY	2 (0.8%)	

Moreover, other cases of sex chromosome aneuploidy were also reported: X monosomy (Pawlak et al., 2000), XXY trisomy (Kubien et al., 1991), X/XX/XXX mosaicism (Wieczorek et al., 2001) and XY-male pseudohermaphroditism (Switonski et al.,

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2005).

The Dutch programs
Screening programs in cattle. The scale of the historical 1;29 Robertsonian translocation screening program in cattle is currently relatively limited in The Netherlands. Indeed, such analyses are not mandatory for the breeders, and only limited pressure is applied by the breeding organizations to encourage cytogenetic analysis.

Screening programs in pigs. Routine karyotyping of AI boars is performed on a large scale by the Cooperative Pig Centers for Artificial Insemination in Pigs. G-banding karyotypes are carried out systematically. Up to now, more than 4,000 pigs have been karyotyped (about 1,000 per year in the recent years). At the beginning of the program, the estimated frequency of chromosomal rearrangements was higher (1.5%) than expected from the literature. This may be due to the fact that some chromosomal aberrations were present in the populations without a specific effect on fertility, and therefore remained undetected. In recent years, the percentage of chromosomal translocations has dropped (e.g. only six confirmed translocations since 2006).

The Spanish programs
After the important observations concerning the 1;29 translocation in Sweden by I. Gustavsson and in France by P. Popescu, a systematic chromosomal control program was initiated in Spain (Zaragoza) for cattle and sheep. Table 9 summarizes the results obtained pertaining to the 1;29 Robertsonian translocation screening program in different cattle breeds (Arruga and Zarazaga, 1984, Arruga et al., 1984; Arruga, 1987).

More than 30% of the analyses have been carried out in the Holstein Friesian dairy cattle breed. No carrier individual was detected in this breed. On the other hand, a rather high frequency of carriers was observed in other breeds, as for instance the Retinta and Rubia Gallega (16.1 and 21.9%, respectively), which are major beef cattle breeds in Spain. The frequency of carriers appears even higher in other breeds (up to 57.1% in the Cachena breed), but the estimated values should be considered with caution due to the very low number of animals screened.

At the same time, other cytogenetic abnormalities were detected, such as the identification of freemartinism in cattle and sheep, or deletions of chromosome 3 in sheep (e.g. Pascual and Arruga, 1996; Arruga and Pascual, 1997).

Thousands of animals were studied during the 1970's,
1980's and 1990's. However, a large decrease of these screen- ing activities has occurred in Spain since that period. The main reasons

for this decline are, on the one hand, the loss of interest in the official and private sectors, and, on the other hand, the lack of financial support for the Spanish Laboratory.

The Hungarian programs
Screening programs in cattle.
Cattle chromosome investigations in Hungary were started in 1972 and since then more than 9,000 animals, mainly AI and other breeding bulls including Hungarian Grey herds (Kovacs, 1978) and relatives of carriers of different chromosome abnormalities have been evaluated (Table 10).

Table 9. Results of the 1;29 Robertsonian translocation screening program carried out in Spain

Breed	Animals controlled	HT (%)[a]	HM (%)[a]
Holstein Friesian	717		
Retinta	254	41 (16.1)	3 (1.2)
Rubia Gallega	146	32 (21.9)	
Parda Alpina	89		
Asturiana de los Valles	126	4 (3.2)	
De lidia	49	4 (8.2)	
Pirenaica	262	7 (2.7)	
Maine-Anjou	69		
Charolaise	72	1 (1.4)	
Alistana	41	2 (4.9)	
Morucha	35	5 (14.3)	
Sayaguesa	25	6 (24.0)	
Avileña	22		
Limousine	62	1 (1.6)	
Fleckvieh	29		
Cachena	7	4 (57.1)	
Normande	10		
Simmental	8		
Holstein-Friesian in Morocco	32		
Rubia d'Ulmes in Morocco	32	1 (3.1)	
Morena del Atlas in Morocco	62	4 (6.5)	
Creole of Argentine	25	1 (4)	
Other breeds[b]	77		
Total	2,251	113 (5.0)	3 (0.1)

[a] HT = heterozygotes; HM = homozygotes.
[b] Including (number of animals): Caldelana (13), Mirandesa (5), Vianesa (6), Limiana (3), Asturiana de los Montes (2), Tudanca (3), Palmeña (2), Canaria (2), Berrenda en Negro (1), Cárdena Andaluza (1), Blonde d'Aquitaine (7), Wild cattle (Doñana) (1), Blanca Cacereña (3), Blanco Azul Belga (12), Mertolenga (1), Piamontes (3) and Jersey (12).

Table 10. Results of the chromosomal screening of breeding bulls carried out in Hungary

Breed	Animals	rob(1;29)(%)	XX/XY (%)	Others[a]
Holstein-Fresian	2,885		81 (2.8)	2 (0.07)
Simmental	858	26 (3.0)	10 (1.2)	2 (0.2)
Hungarian Grey	208	10 (4.8)		
Belgian White-Blue	140			
Limousin	84		1 (1.2)	
Charolais	74	3 (4.1)		
Hereford	62			
Europ. Red and White	16			
Blonde d'Aquitaine	15	2 (13.3)		
Jersey	10			
Lincoln Red	6			
Kostroma	2			
Crossbred	209	1 (0.5)	4 (1.9)	
Total	4,569	42 (0.9)	96 (2.1)	4 (0.09)

[a] In Holstein-Friesian: one rob(13;21) in mosaic form, not found among the offspring, and one XY/XXY chimeric karyotype; in Simmental: one rob(14;21), hereditary, and one rob(5;18) in mosaic form, not found among the offspring.

Two hereditary abnormalities have been identified: the 1;29 centric fusion in the Blonde d'Aquitaine (Kovacs and Szepeshelyi, 1987), Charolais (Kovacs and Szepeshelyi, 1987; Tozser et al., 1995), Maremmana—Hungarian Grey (Kovacs, 1989), Simmental (Kovacs and Szepeshelyi, 1987), Swedish Red and White (Gustavsson and Kovacs, 1977), and the 14;21 centric fusion in the Simmental breed (only in one AI bull and its relatives, Kovacs, 1989). In the seventies, the frequency of the 1;29 translocation carriers was 3.6% among Simmental bulls. Since 1975, all AI bulls have been karyotyped. Carriers of structural chromosome abnormalities as well as their stored semen were culled. As the calves could inherit chromosome abnormalities only from their mothers, the frequencies of those were halved in each generation. Today, the Simmental population of Hungary may be considered to be free of the 1;29 translocation. However, the 1;29 translocation was introduced into some herds of the Hungarian Grey cattle by a single Maremmana bull imported from Italy in 1971 (Kovacs, 1989). Some 720 Hungarian Grey cattle have been in- vestigated and carriers of both sexes were culled. Currently, this ancient breed is practically free of the 1;29 translocation again. Among the 140 Belgian White-Blue bulls investigated, none was found to be a carrier of the 1;29 translocation (Nicolas et al., 1995).

In a joint four year project with the U.S.A., examination of 69 cattle revealed the 1;29 translocation in the Charolais and Brown Swiss breeds. As well, an original 1;8;9 complex translocation in a Brown Swiss bull was identified. This difficult case was

diagnosed using synaptonemal complex analysis and G-banding (Kovacs et al., 1992a). The bull had greatly reduced fertility and there were multiple lethal malformations in some of the offspring. Semen was imported to Europe (Denmark and Italy). In Denmark, insemination of 223 cows resulted in only 11 calves and the abnormality was found among them (Christensen et al., 1992). No information is available on the outcome of the importation to Italy.

The 1;29 translocation was imported from France (Blonde d'Aquitaine and Charolais breeding animals and Montbéliarde semen), Germany (Simmental breeding ani- mals), Italy (Maremmana), Sweden (Swedish Red and White heifers) and The Netherlands (Blonde d'Aquitaine embry- os). However, the last 1;29 carrier AI bull was found in 1999, and all of the 372 bulls (320 Holstein-Friesian, 42 Simmental, six Limousin, two Belgian White-Blue, one Blonde d'Aquitaine and one Polled Charolais) investigated so far in this century had normal karyotypes.

Large-scale testing of bulls allowed the identification of other chromosomal abnormalities. Two cases of Robertsonian translocations in mosaic form: 5;18 in a Simmental bull (Kovacs and Szepeshelyi, 1987) and 13;21 in a Holstein- Friesian AI bull (Kovacs et al., 1973). XX/XY chimaerism was diagnosed in more than 100 individuals of different breeds including one supposedly primary chimera single- born Simmental bull (Kovacs et al., 1977). XXX/XY and XY/XXY chimeric karyotypes were also identified in two bulls each. An exceptional opportunity to survey the losses connected to the 1;29 translocation occurred in Hungary at a large state farm (Kovacs and Csukly, 1980; Kovacs, 1989, 1994; Kovacs et al., 1992b). Almost three hundred half-sib daughters of a Simmental bull heterozygous for the 1;29 translocation were involved in a blind study (the farm had not been informed of the results of individual chromosome investigations) between 1975 and 1992. Most of the follow- ing results were confirmed by numerous studies as reviewed in Kovacs (1989). Heterozygous carrier cows had fewer but longer lactations than their half-sisters bearing the normal karyotype, with the result that the two groups had practically equivalent lifetime milk production values (Kovacs et al., 1992b; Kovacs, 1994). In the whole half-sister group, the number of t+ individuals was lower than that of the t– ones. The difference was most apparent among cows. Among calves, the expected Mendelian distribution of 50:50 was actually almost fully observed, while there were 3.19 times more t+ heifers among the culled ones. The insemination index (number of inseminations/pregnancy) of the t+ group was 28.43% higher and its fertility (pregnancies % after the first insemination)

was 32.41% lower as compared to the t–control group. Disadvantageous differences were found in the service period (+20.64 days) and in the ages at first breeding (+11.86 days), in age at the first (+30.51 days), second (+53.03 days), third (+109.48 days) and fourth (+123.63 days) calvings, in the calving interval (+21.62 days), in the days open (+17.16 days) as well as in the calving rate (–4.57%). The gestation length was the same, and the involution period was shorter by 7.79 days. This single advantage recognized was not statistically significant and was probably the effect of the higher culling rate. There were no significant differences in the occurrence of abortions and dead calves between the t+ and t– control group (Kovacs et al., 1992b; Kovacs, 1994). The interval between two inseminations did not differ from the control, thus indicating a normal cycle of 20–21 days and an early preimplantation loss within the first half of the cycle. The total zygotic loss for the heterozygous carrier group was calculated to be 22.7% higher. The estimated yearly loss in Hungary connected to the 1;29 translocation was the culling of 920 heifers, 9,555 surplus inseminations and 451 fewer calves (Kovacs, 1989). This loss was caused by the production of gametes (and therefore embryos) with unbalanced chromosomal constitutions (Bonnet-Garnier et al., 2006, 2007).

Screening programs in other species. More than 500 artificial insemination boars were investigated for the occurrence of translocations at the G-band level. There was no positive diagnosis, possibly due to the very strict selection for litter size. Animals showing reduced prolificacy were culled very quickly, and could therefore not be subjected to chromosomal investigations. C-band polymorphism was also studied revealing sporadic occurrence of large hetero-chromatic blocks on the acrocentrics. This condition is suspected to be related to reduced litter size.

Large scale chromosome investigations were also carried out in pedigree stocks of poultry lines, for use in the selection procedures. Dead embryos at the early stages of incubtion were analysed cytogenetically to determine the potential accumulation of chromosome abnormalities in certain families or individuals. In two layer hybrid lines the proportion of embryos presenting abnormal karyotypes was esti- mated between 20 and 24% (482 and 572 dead embryos analyzed, respectively – Hidas et al., 1996). Similar investigations were carried out in goose breeding stocks (Liptoi et al., 2005).

Besides the programs mentioned above, investigation of a few individuals in other farm animal species has been con- ducted. A 63,X mare (Bozsaky et al., 2003) as well as one case of XX-sex reversal in one polled goat

(pseudomale) were found.

The Portuguese programs

Screening programs in cattle. Cytogenetic screening pro- grams are not mandatory for the Portuguese breeders, and limited pressure is applied by the breeding organizations as already reported for the Netherlands case. This situation is not a ref lection of a low incidence of the 1;29 Robertsonian translocation in Portugal. On the contrary, the translocation in many Portuguese commercial cattle breeds (e.g. Alentejana, Barrosã, Maronesa and Mirandesa) is wide- spread and the heterozygotes are common in these popula-tions (Rangel-Figueiredo and Iannuzzi, 1990, 1993; Chaves et al., 2003a). It is also important to note that the highest frequency for the 1;29 Robertsonian translocation was found in a Portuguese breed (Barrosã, 6,000 animals in this population, 206 karyotyped) with 70% of individuals carry-ing the rob(1;29) (17% of which were homozygous – Rangel- Figueiredo and Iannuzzi, 1993). Furthermore, in the Barrosã breed, two more Robertsonian translocations were de- tected, namely (15;25) (Iannuzzi et al., 1992) and (16;18) (Iannuzzi et al., 1993).

In the Portuguese laboratory, cytogenetic screening in cattle has been conducted since 2000, especially regarding the cattle breeds from the North of Portugal. However, these analyses are conducted for research purposes, and not because the breeders or breeding organizations request the services of the laboratory. The number of analyses carried out is therefore rather limited (about 200 animals screened in each breed). The most frequent rearrangement found is certainly the 1;29 translocation (from 9 to 77% of carriers, depending on the breed). Recently, efforts have focused on the study of the fundamental features of this chromosomal rearrangement as it constitutes an excellent chromosome model (Chaves et al., 2003a; Di Meo et al., 2006).

In addition, the involvement of chromosomal abnormalities in some congenital defects has been studied. A mixoploid (data not published) and a complex intersex condition with the existence of Y chromosome material in the two X chromosomes (Payan-Carreira et al., 2008) was found in two Holstein calves, respectively.

Screening programs in other species. Sheeps, goats and pigs are also not systematically screened in Portugal. Nevertheless, in 2003, an 8;11 translocation in a female sheep belonging to 'Churra da Terra Quente' breed was detected (Chaves et al., 2003b).

Discussion

The aim of this paper was not to do an exhaustive survey of all published results concerning the cytogenetic screnning of domestic animal populations worldwide, but rather to illustrate this activity by presenting the data obtained in eight European countries in which animal cytogenetics laboratories are active: France, Italy, Romania, Poland, The Netherlands, Spain, Hungary and Portugal. Even if a large proportion of chromosomal studies in farm animal species have been carried out in these eight countries during the last 15 years, original studies were conducted in other countries too, but usually on a more limited scale. This was for instance the case of Switzerland and Finland. In Switzerland, Tschudi (1984) reported that among the 2,941 bulls investigated between 1973 and 1984, 31 (1%) carried the classical rob(1;29), whereas three carried other centric fusions and 32 presented a blood XX/XY chimerism. Comparable results were obtained later (1994–2001 period): 11 bulls, mainly sons from imported (U.S.A.) semen, carrying the rob(1;29), over 2,315 controlled (0.5%) – Stranzinger (unpublished results). Complementary, molecular studies carried out by Joerg et al. (2001) revealed significant molecular differences in the centromeric region of different centric fusions, and proved the very ancient origin of some of them. No systematic and large scale control program exists in Finland, but animals with reproductive problems have been occasionally studied, allowing the discovery of original chromosomal rearrangements in cattle, pigs and horses (e.g. Villagomez et al., 1993; Mäkinen et al., 1999b, 2000, 2006) (Table 11).

Table 11. Chromosomal rearrangements identified in hypoprolific boars in Finland

(Reciprocal) translocation[a]	Average litter size	Reference
(7q–;12q+)	7.9	Kuokkanen and Mäkinen, 1987
(7;15)(q24;q26)	5.2	Mäkinen et al., 1997
(2;9;14)(q23;q22;q25)	3.3	Mäkinen et al., 1997
(1p+;15q–)	7.1	Kuokkanen and Mäkinen, 1988
(8;10)(p11;q13)	9.0	Mäkinen et al., 1999a
(4q+;13q–)	7.4	Mäkinen and Remes, 1986
(2;4;15)	8.2	
(1p–;11q+)	7.8	Kuokkanen and Mäkinen, 1988

[a] The first four translocations were identified in on-farm breeding boars; the last four translocations were identified in AI boars

Similarly, studies in Canada were conducted on the effects of either the t(1;29) (Schmutz and Moker, 1989; Schmutz et al., 1991) or t(14;20) (Schmutz et al., 1997) on the karyotype of embryos from carrier parents. A small scale screening study of 134 bulls of 11 breeds detected seven t(1;29) carriers (Schmutz et al., 1990). In other small scale screening studies of cattle abortuses, one monosomy, seven trisomies, and one translocation were detected in 73 of the 107 samples success- fully cultured and karyotyped (Coates et al., 1988; Schmutz et al., 1996).

The results presented above and in the other papers of this special issue clearly illustrate that, even if the number of scientists and laboratories involved in clinical animal cytogenetics has substantially decreased over the last 15 years, our discipline is still active, scientifically attractive and important for livestock breeders. Overall, the cytogenetic investigations carried out for a large number of AI bulls have had very positive technical and economic repercussions. On the one hand, removing the carriers from reproduction during the first year of the selection procedure avoided the dissemination of the chromosomal rearrangements in the off- spring. As a consequence, the frequency of the widespread 1;29 Robertsonian translocation, for instance, has been dramatically reduced in most countries during the last 20 years. On the other hand, the costs corresponding to the complete selection procedure of the carrier animals were saved. The development of the activities in the most active laboratories largely compensates for the decrease in the number of laboratories involved in that field. The recent results obtained, showing for instance that the prevalence of chromosomal rearrangements is much higher than that initially considered, at least in some species (e.g. in pigs the prevalence of structural chromosomal rearrangements is 1/200, i.e. com- parable to man, and not 1/1,500 as published earlier by Legault and Popescu, 1993), make the professional organizations as well as the scientific community more attentive to our work. In pigs again, the breeding and AI companies wishing to carry out systematic controls in their populations have never been so numerous as now. The perspectives of development of the different laboratories are therefore still important. In cattle for instance, one can argue that it would be very pertinent to systematically screen all the bulls used in artificial insemination (AI) centers, as already carried out for purebred boars in some countries. Such an objective should concern the beef cattle breeds, as already considered in the past in some countries, as well as the dairy cattle breeds, and especially the Holstein Friesian breed, which is numerically the most important in almost all European countries, and for which artificial insemination is

nearly the only mode of reproduction which results in very high diffusion levels of genetics from selected bulls. Indeed, there is a non-negligible risk that some of the selected bulls will carry particular chromosomal rearrangements with low to moderate effects on the fertility of the mates (e.g. Robertsonian translocations or inversions). As shown in man (Anton et al., 2005; Roux et al., 2005) and verified in some animal studies (Bonnet-Garnier et al., 2006), the individuals carrying such rearrangements may produce a low proportion of unbalanced gametes (leading to early embryonic mortality). The probability of not detecting these rearrangements during the progeny testing phase of the bulls is therefore important (average fertility decrease of the mates is too low to be detected). Without cytogenetic screening, the undetected carrier animals will be used and then sire tens if not hundreds of thousands offspring. With such dif- fusion levels, even with low effects of the rearrangements, the economical consequences could be very substantial, and probably much more important than the global cost of the chromosomal screening program. The full progeny testing cost of one AI bull is approximately 40,000 euros. The cost of carrying out one karyotype is less than 100 euros. Such a disproportion between these two values should incite the breeding organizations to more systematically screen their breeding animals, especially in a context

where fertility has become one of the main limiting factors of the economic efficiency of the herds, and was therefore introduced into the global selection goals of most dairy cattle breeds (Weigel, 2006). In pig production, one can also demonstrate easily that the overall cost of a chromosomal screening program is much lower than the cost of using translocation carrier males in AI stations. Indeed, considering a 1/200 incidence of reciprocal translocations in this species, the cost of detecting one particular rearrangement is about 12,000 euros (200 60, where 60 euros is the cost of carrying out the karyotype of one animal). In contrast, the cost of using one translocated boar in an AI centre is at least 20,000 euros. Indeed, the translocation carrier boar will be used until the hypoprolificacy of his mates is detected, i.e. at least for four months. During that period, it will produce at least 160 litters (40/month). The total number of piglets lacking at the end of the 4-month period will be 640 (1604, where 4 is the average litter size reduction connected to the translocation), which corresponds to a 19,200 euros economic loss for the breeders (640— 30, where 30 euros is the economical value of one piglet). The economic loss is even much higher if we consider that the chromosomal rearrangement is carried by a purebred boar at the selection or multiplication levels of the production pyramid, as 50% of their offspring will in turn carry the rearrangement.

In Western European countries, where

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farmers are in a stable financial situation and the profitability of animal husbandry is better than in countries with reorganizing agricultural structures, the above mentioned facts and arguments are recognized to be easier to implement. A good ex- ample is France where almost all AI bulls (beef cattle breeds) and purebred boars are now under cytogenetic control. In contrast, governmental support of national screening pro- grams is decreasing in a number of countries. Breeding companies and associations could incorporate the chromo- some analysis into their quality control systems, which would have great marketing importance demonstrating their careful business policy.

The importance of cytogenetics in veterinary medicine needs reaffirmation. The involvement of chromosomal abnormalities in many congenital defects and cancers has been documented in man for a long time (Lejeune et al., 1959,1963; Nowell and Hungerford, 1960). Major medical stakes justified the extraordinary development of human cytogenetics during the 20th century (some 900,000 cytogenetic analyses are now performed each year in approximately 500 laboratories worldwide – Gersen, 1999). In humans, cytogenetic investigations are carried out systematically in cases of congenital malformations. This is far from being the case in animal species, mainly for economic reasons (the economic value of a piglet, a lamb or a calf in some breeds is lower than the cost of carrying out one karyotype). Nevertheless, the identification and characterization of particular chromosomal rearrangements paved the way for discovering many deleterious genes in humans. This approach has been almost systematically neglected in animal species. However, the improvement of the molecular cytogenetic techniques, well mastered in our laboratories now, gives us new opportunities in that field. Some recent examples in constitutional (Pinton et al., 2002; Payan-Carreira et al., 2008) and cancer animal cytogenetics (Thomas et al., 2003, 2005; Santos et al., 2006) illustrate these opportunities.

Finally, it can also be argued that farm animal species are very interesting and informative alternative models in bio- medical research (Pliska and Stranzinger, 1990). They can be useful in particular to study some fundamental aspects of the cell and/or chromosome biology, as well as for evolutionary studies (Iannuzzi et al., 2000; Chaves et al., 2005; Di Meo et al., 2005; Iannuzzi, 2007). For instance, the karyotype structure of the domestic pig (*Sus scrofa domestica* L.) is much more similar to human than that of the mouse. The females of this species are relatively prolific (12–14 progeny per litter on average), which means that the number of oocytes or embryos that can be analysed per female is relatively high. In

addition, the generation interval is relatively short (about two years), and the experimental production of individuals with particular karyotypes is possible at reason- able expense. In other animal species, as for instance bovines, the reproductive biotechnologies (e.g. ovum pick-up and in vitro fecundation, or somatic cell nuclear transfer) are well mastered, this facilitates the collection of biological material of interest. In France, these animal model species have been used to study the impact of chromosomal rearrangements on the course and products of meiosis. Some questions, very difficult to investigate in humans for technical and ethical reasons, as for instance the difference of segregation profiles between males and females, or the variability of segregation profiles between individuals having the same karyotype, or between sperm samples for the same individual, could be thoroughly documented (e.g. Pinton et al., 2005; Bonnet-Garnier et al., 2007). In Canada, the limited access to relevant foetal oocytes which

precluded direct study of meiotic events in female carriers was over- come by the use of somatic cell nuclear transfer in cattle to study meiosis in a female carrying a sex-dependent fertility- impairing X-chromosome abnormality (Rho et al., 2007). Other up to date biological questions could be investigated using these animal species, as for instance the impact of chromosomal rearrangements on the spatial organization of chromosome territories and gene expression in somatic cells, or the spatial organization of chromosome territories in the gametes. The large scale chromosomal screening pro- grams carried out in several European countries now makes the raw material necessary for such studies available.

Acknowledgements

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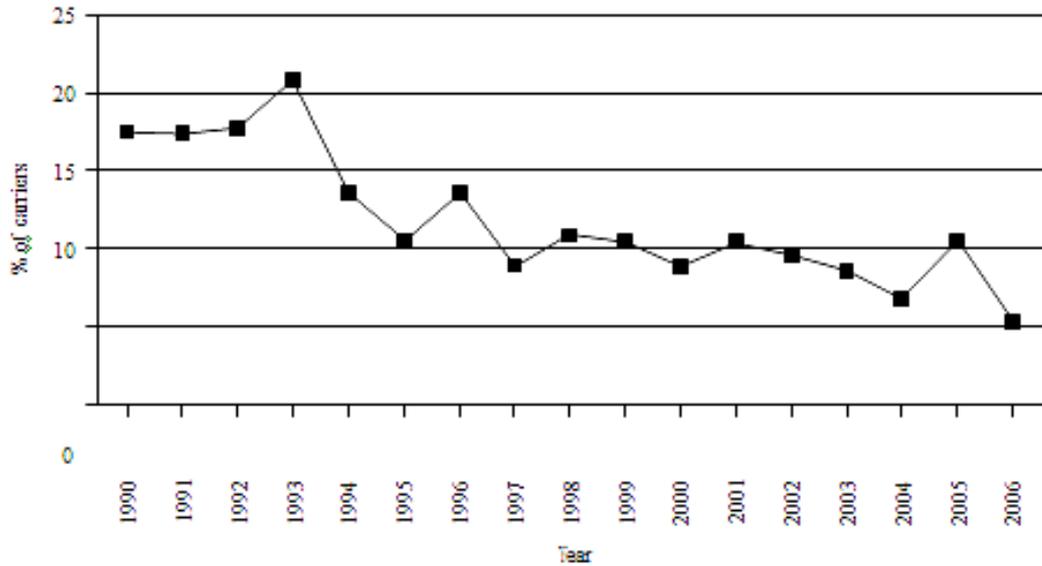527 (2001).

Fig. 1- Evolution of the frequency of bulls carrying the 1;29 Robertsonian translocation in the Blonde d'Aquitaine beef cattle breed in France.
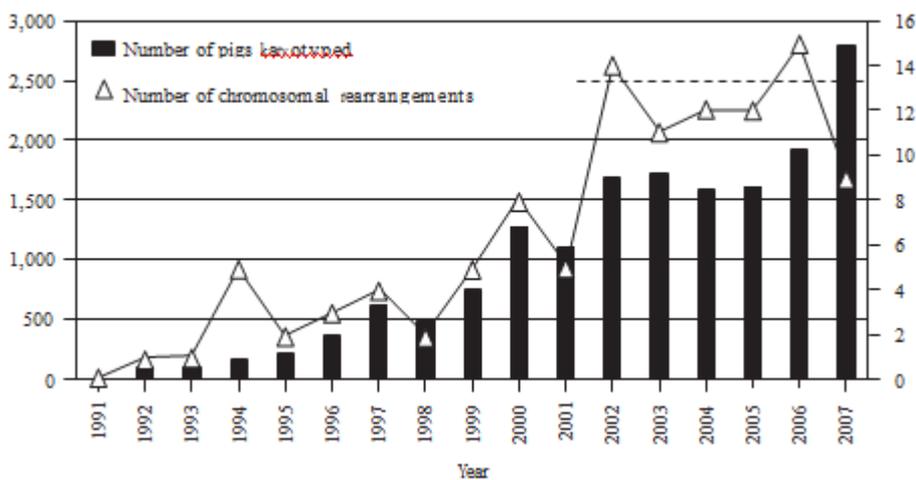

Fig. 2- Evolution of the annual number of pigs karyotyped in France and of the number of structural chromosomal rearrangements identified. For year 2007: the total number of pigs karyotyped was extrapolated from the number of pigs karyotyped from January 1st to July 1st; the number of chromosomal rear rangements indicated in the figure corresponds to the January 1st to July 1st period (only 6 months).