fluorescence microscopy and image capturing using SmartCapture 3 software (Digital Scientific UK).

Clear signals were obtained from each subtelomeric probe. These were tested on normal animals and animals that exhibit translocations, providing preliminary evidence that this technique is a valid tool for the identification of translocations that affect fertility in pigs.

When combined with a tool originally developed for humans to enable the simultaneous detection of all porcine chromosomes on one slide (Multiprobe™ Device), the speed and cost of chromosomal analysis for translocations that affect fertility will be greatly improved, therefore offering significant benefits to animal genetic research and the animal breeding industry.

O3

The incidence of translocations in young breeding boars in Canada

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The objective of the project was to carry out the first systematic screening program for chromosomal abnormalities in young breeding boars in Canada. To date, a total 300 young boars from 4 different breeds (Duroc, Landrace, Pietrain and Yorkshire) were karyotyped by G-banding. Four previously unreported reciprocal translocation including rcp(1;5), rcp(3;4), rcp(8;13) and rcp(7;15) and one previously reported Robertsonian translocation rob(13;17) were found. Consequently, the frequency of chromosome abnormalities in this study was 1.67 %. By extending the sampling to other members of the pedigree, it was determined that rcp(3;4) and rob(13;17) were inherited from their dams and rcp(8;13) was a "de novo" event. Comparing with the herd average, average litter size of rcp(3;4), rcp(8;13) and rcp(7;15) translocation carrier boars was noted to be reduced (24 %, 24 % and 38 %, respectively) while for carriers of rob(13;17), it was only slightly reduced (9 %). Interestingly, for rcp(3;4), the overall reduction in litter sizes for female carriers was substantially lower (only 4 %) compared to male carriers (24 %). Chromosome analysis of live offspring from 2 full litters of carrier boars showed a 20 and 40 % transmission rate to progeny for rcp(7;15) and rob(13;17), respectively. More studies need to be carried out to further investigate the effects of these translocations. (Research support was obtained from NSERC, Agriculture and Agri-Food Canada, and the Canada Research Chairs program).

O4

Mix of two chromosomal aberrations in a newborn calf 2n=60,XX, t(11;25)(q11;q14-21)

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A newborn calf of the Agerolese breed underwent cytogenetic investigation because presented hyperflexion forelimbs, red eyes and inability to stand up. Anamnesis revealed the mother, phenotypically normal, was carrier of a t(11;25)(q11,q14-21). The newborn died after a few weeks and no internal alterations were found by veterinarian after the post mortem examination. The mother presented, after a cytogenetic investigation, a reciprocal translocation between chromosome 11 and 25 and the presence of two ders: der11 and der25, for the position of corresponding centromere. On the other hand, the veal revealed a different chromosomal aberration in comparison to her mother. In fact, after R-banded karyotype, the calf showed both chromosomes 25, one chromosome 11 and one der (der25). FISH analysis was performed with the same BAC clones used to detect the translocation in the mother: BAC142G06 mapped on the proximal region of both BTA25 and der25; BAC513H08 mapped to BTA 25q22dist; BAC533C11 mapped to the proximal region of BTA11 and der25. Finally, we confirmed both the localization of the breakpoints on band q11 (centromere) of chromosome 11 and q14-21 of chromosome 25, and the loss of the der11. In this way, it is showed a different cytogenetic aberration in the veal: a partial trisomy of chromosome 25 and a partial monosomy of chromosome 11. We have

been studying a correlation between this aberration and some gene involved comparing it with corresponding human clinical cases.

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O5

A new case of 64,XX sex reversal syndrome in a Spanish purebred horse

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Abnormal morphologies in horses are related in most of the cases to atypical chromosomal complements. An higher percentage of these horses are characterized as animals with unspecified sex or with disorders in the sexual development (DSD). We studied the case of a 3 years old Spanish Purebred horse submitted to karyotiping due to its abnormal sexual morphology. On physical examination, the animal showed an abnormal foreskin, two prominent nipples, a tight scrotum and retained testicles. His sexual behaviour and libido were normal for a male horse. The animal was castrated at 2 years of age and the pathological study showed no abnormal tissue. The animal was karyotyped using conventional citogenetic techniques (chromosome counting and C-banding) and in situ fluorescent hybridization with two specific WCPP for ECAX and ECAY chromosomes showing a female 64,XX chromosomal complement. The presence of three different genes linked to sex chromosomal in the blood DNA of the horse was studied using PCR. Resulting in a positive amplification for ZFX and AMX and no amplification of the SRY, ZFY and AMY genes. Four different microsatellite makers linked to the ECAX chromosome were analysed to determine the number of chromosomal copies. The genotyping of LEX 026, TKY38, TKY270 and LEX003 showed two different alleles in each marker, denoting the presence of at least two different ECAX chromosomes. All the molecular studies were repeated using DNA obtained from hair follicles to discard the presence of a blood chimerism case. The results were identical than those obtained previously. Based on our results we diagnosed the horse as a 64,XX SRY negative DSD carrying a male-like genitalia. To our knowledge, this is the second time that this kind of abnormality was reported in the Andalusian horses and the first time showing this kind of morphological abnormalities. At the present time, the cause of this abnormal sexual development remains certainly unknown. However, the most accepted theory is the occurrence of an androgen exposure during sexual development of the embryo leading to the masculinization of the female foetus. Finally, we suggest the use of genetic and cytogenetic diagnostic tools in the veterinary practice as a valuable tool to determine the origin of reproductive failures among horses.

O6

Disorders of Sexual Development (DSD) in dogs with ambiguous external genitalia—survey of 30 cases

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The genetic background of disorders of sexual development (DSD) may be related with chromosomal aberrations, gene mutations or may have a multifactorial etiology. During the last 10 years we have analyzed 30 dogs subjected to cytogenetic (Giemsa staining and whole sex chromosome painting) and molecular analyses (detection of the *SRY* and *ZFY* genes) due to the abnormalities of their external genitalia. In most cases (21) the enlarged clitoris with a bone was observed in phenotypic females with 78,XX chromosome set and lack of the Y-derived genes (*SRY* and *ZFY*). The consequent characteristics were observed in the following breeds: Bernese Mountain Dog (1), Cocker Spaniel (4), German Shepherd (2), Miniature Pincher (1),