

# Cytogenetics with special reference to domestic animals

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Essential Genetics:  
a genomic perspective.

Seminar 4

## Overview

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- Studying chromosomes*
  - The normal karyotypes of domestic animals
  - Chromosome abnormalities
  - Chromosome abnormalities of domestic animals
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## Studying chromosomes

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- Classical karyotyping*
  - Fluorescent in Situ Hybridization (FISH)
  - Chromosome painting
  - Molecular karyotyping
  - Chromosome sorting
- 

## Classical karyotyping

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- Obtaining and preparing cells for chromosome analysis
  - Karyotyping and chromosome banding
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## Obtaining and preparing cells for chromosome analysis

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- Cell source:
    - Blood cells
    - Skin fibroblasts
    - Amniotic cells / chorionic villi
  - Increasing the mitotic index (proportion of cells in mitosis) using colcemid
  - Synchronizing cells to analyze prometaphase chromosomes
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## Karyotyping and chromosome banding

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- Chromosome banding
  - Molecular interpretation of chromosome bands
  - Chromosome nomenclature
-

# Chromosome banding

## Box 2.2: Chromosome banding.

**G-banding** – the chromosomes are subjected to controlled digestion with trypsin before staining with Giemsa, a DNA-binding chemical dye. Positively staining dark bands are known as G bands. Pale bands are G negative.

**Q-banding** – the chromosomes are stained with a fluorescent dye which binds preferentially to AT-rich DNA, such as Quinacrine, DAPI (4',6-diamidino-2-phenylindole) or Hoechst 33258, and viewed by UV fluorescence. Fluorescing bands are called Q bands and mark the same chromosomal segments as G bands.

**R-banding** – is essentially the reverse of the G-banding pattern. The chromosomes are heat-denatured in saline before being stained with Giemsa. The heat treatment denatures AT-rich DNA,

and R bands are Q negative. The same pattern can be produced by binding GC-specific dyes such as chromomycin A<sub>3</sub>, olivomycin or mithramycin.

**T-banding** – identifies a subset of the R bands which are especially concentrated at the telomeres. The T bands are the most intensely staining of the R bands and are visualized by using either a particularly severe heat treatment of the chromosomes prior to staining with Giemsa, or a combination of dyes and fluorochromes.

**C-banding** – is thought to demonstrate constitutive heterochromatin, mainly at the centromeres. The chromosomes are typically exposed to denaturation with a saturated solution of barium hydroxide, prior to Giemsa staining.

# Chromosome banding

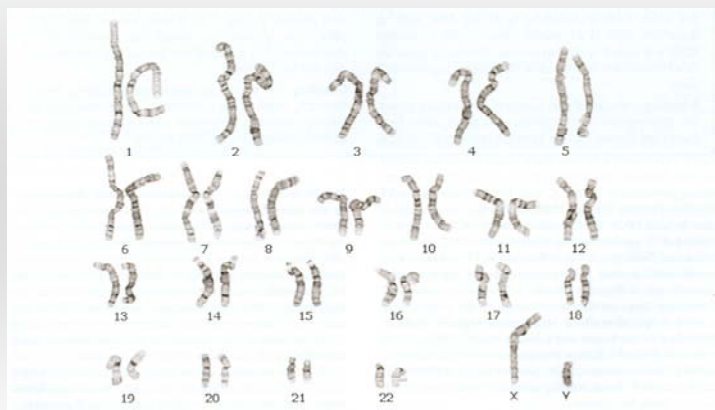


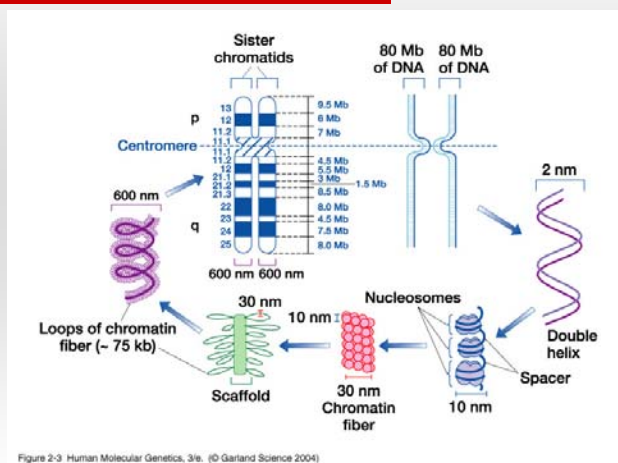
Figure 2.14. G-banded prometaphase karyogram of mitotic chromosomes from lymphocytes of a normal male at between 550 and 850 bands per haploid set.

Compare with the idealized ideograms in Figure 2.15. Overall lengths of metaphase chromosomes range between 2 and 10  $\mu\text{m}$ ; the DNA of the cell, if stretched out, would be about 2 m long. Reproduced from Cross and Wolstenholme (2001). In *Human Cytogenetics: Constitutional Analysis*, 3rd Edn (ed. D. E. Rooney). Reproduced by permission of Oxford University Press.

## Molecular interpretation

- G+ bands (= R- bands)
  - AT rich
  - Gene poor
  - LINE rich
  - Late replicating
  - SAR (Scaffold Attachment Regions) rich

## From DNA duplex to metaphase chromosome



## Molecular interpretation

- G- bands (= R+ bands)
  - GC rich
  - Gene rich
  - SINE rich
  - Early replicating
  - SAR (Scaffold Attachment Regions) poor

## Chromosome nomenclature

Table 2.3: Human chromosome groups

Group	Chromosomes	Description
A	1-3	Largest; 1 and 3 are metacentric but 2 is submetacentric
B	4,5	Large; submetacentric with two arms very different in size
C	6-12, X	Medium size; submetacentric
D	13-15	Medium size; acrocentric with satellites
E	16-18	Small; 16 is metacentric but 17 and 18 are submetacentric
F	19,20	Small; metacentric
G	21,22, Y	Small; acrocentric, with satellites on 21 and 22 but not on the Y

Autosomes are numbered from largest to smallest, except that chromosome 21 is smaller than chromosome 22.

# Chromosome nomenclature

## Box 2.3: Human chromosome nomenclature.

The **International System for Human Cytogenetic Nomenclature (ISCN)** is fixed by the Standing Committee on Human Cytogenetic Nomenclature (see Further reading). The basic terminology for banded chromosomes was decided at a meeting in Paris in 1971, and is often referred to as the Paris nomenclature.

Short arm locations are labeled **p** (*petit*) and long arms **q** (*queue*). Each chromosome arm is divided into regions labeled p1, p2, p3 etc., and q1, q2, q3, etc., counting outwards from the centromere. Regions are delimited by specific *landmarks*, which are consistent and distinct morphological features, such as the ends of the chromosome arms, the centromere and certain bands. Regions are divided into bands labeled p11 (one-one, not

eleven!), p12, p13 etc., sub-bands labeled p11.1, p11.2 etc. and sub-sub-bands for example p11.21, p11.22, in each case counting outwards from the centromere (Figures 2.13, 2.15).

Relative distance from the centromere is described by the words **proximal** and **distal**. Thus, proximal Xq means the segment of the long arm of the X that is *closest to the centromere*, while distal 2p means the portion of the short arm of chromosome 2 that is most *distant from the centromere*, and therefore closest to the telomere. Other common terms are as below.

When comparing human chromosomes with that of another species, the convention is to use the first letter of the genus name and the first two letters of the species name (e.g. HSA18 means human – *Homo sapiens*) chromosome 18.

# Chromosome nomenclature

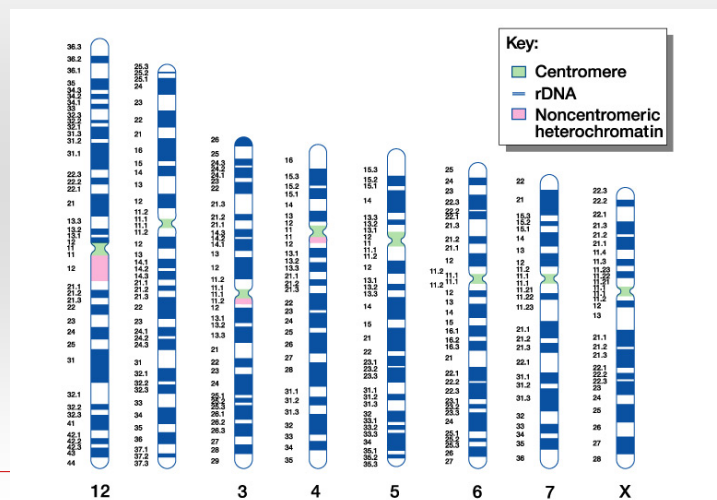


Figure 2-15 part 1 of 2 Human Molecular Genetics, 3/e. (© Garland Science 2004)

# Chromosome nomenclature

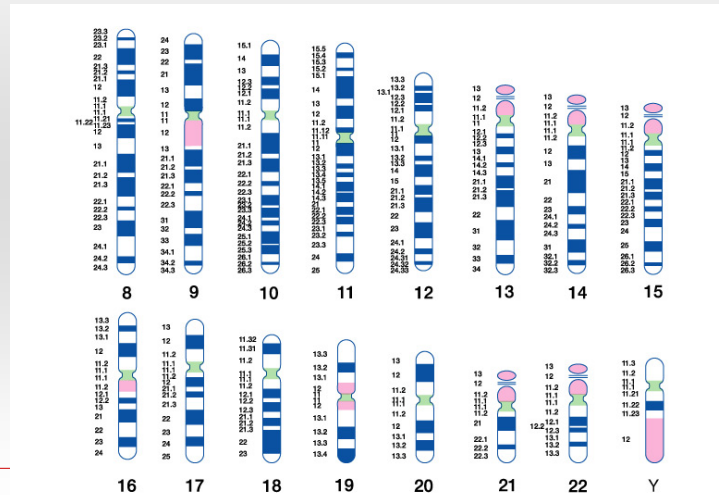


Figure 2-15 part 2 of 2 Human Molecular Genetics, 3/e. (© Garland Science 2004)

# Studying chromosomes

- Classical karyotyping
- Fluorescent in Situ Hybridization (FISH)*
- Chromosome painting
- Molecular karyotyping
- Chromosome sorting

# Fluorescence in situ hybridization (FISH)

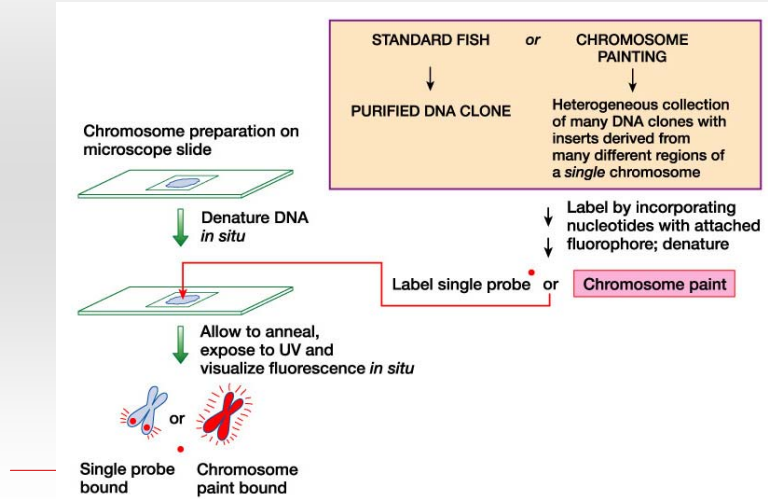


Figure 2\_16 Human Molecular Genetics, 3/e. (© Garland Science 2004)

# Fluorescence in situ hybridization (FISH)

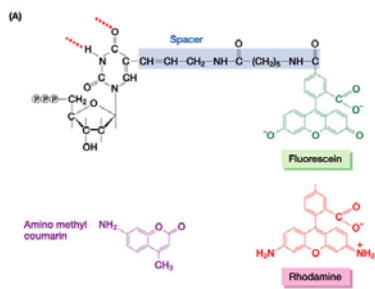


Figure 6-5 part 1 of 2 Human Molecular Genetics, 3/e. (© Garland Science 2004)

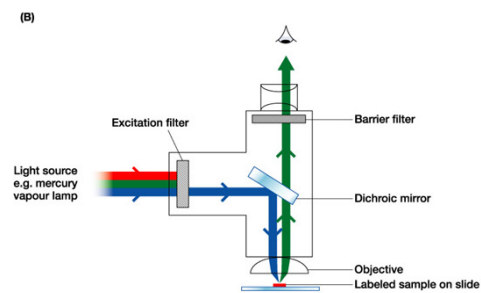
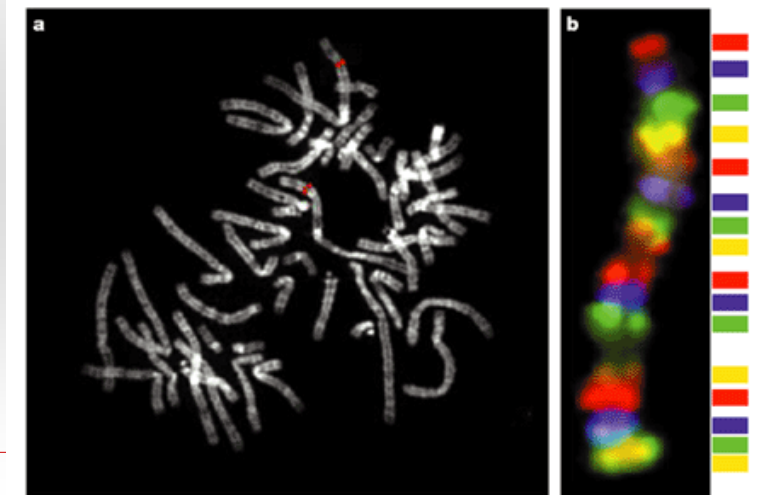


Figure 6-5 part 2 of 2 Human Molecular Genetics, 3/e. (© Garland Science 2004)

## Fluorescence in situ hybridization (FISH)

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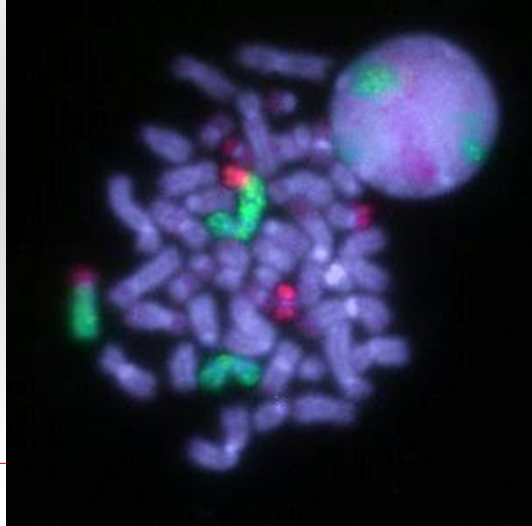
## Studying chromosomes

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  - Chromosome painting*
  - Molecular karyotyping
  - Chromosome sorting
-

## Chromosome painting

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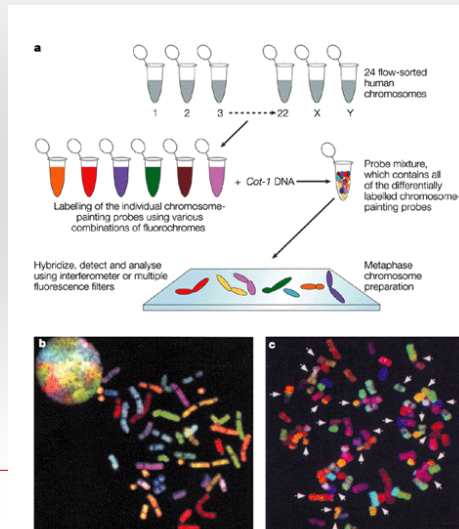


## Studying chromosomes

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- Classical karyotyping
  - Fluorescent in Situ Hybridization (FISH)
  - Chromosome painting
  - Molecular karyotyping*
  - Chromosome sorting
-

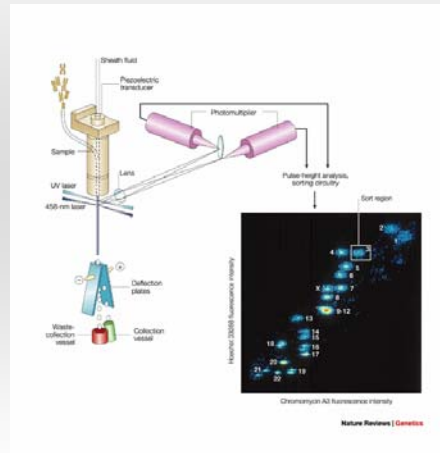
## Molecular karyotyping



## Studying chromosomes

- Classical karyotyping
- Fluorescent in Situ Hybridization (FISH)
- Chromosome painting
- Molecular karyotyping
- Chromosome sorting*

## Flow-sorting chromosome



## Overview

- Studying chromosomes
- *The normal karyotypes of domestic animals*
- Chromosome abnormalities
- Chromosome abnormalities of domestic animals

# The normal karyotype of domestic animals

Human, <i>Homo sapiens</i>	23	Cat, <i>Felis catus</i>	19
Horse, <i>Equus caballus</i>	32	Dog, <i>Canis familiaris</i>	39
Pig, <i>Sus scrofa</i>	19	Mouse, <i>Mus musculus</i>	20
Cattle, <i>Bos taurus</i>	30	Rat, <i>Rattus norvegicus</i>	21
Sheep, <i>Ovis aries</i>	27	Chicken, <i>Gallus domesticus</i>	Ca. 39

(haploid chromosome number)

# Pig

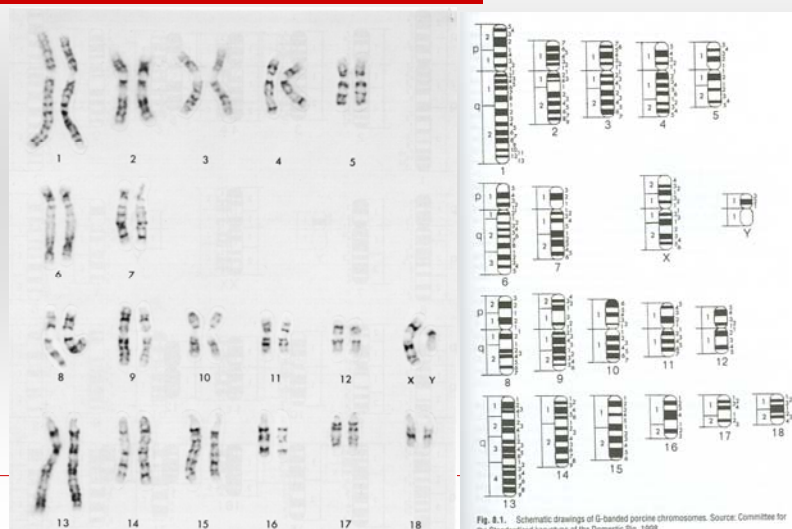


Fig. 8.1. Schematic drawings of G-banded porcine chromosomes. Source: Committee for the Standardized karyotype of the Domestic Pig, 1998.

# Pig

## SUIDAE

	2n	Comments	References
<b>Babirusa</b> <i>Babirusa babirusa</i>	38	11 autosomes and X identical to dom. pig. 5 pairs with no direct equivalents. Y isacro, with distinct p arm	Bosma (1980) Bosma and de Hann (1981)
<b>Phacochoerus</b> <i>Phacochoerus aethiopus</i> (Wart hog)	34	14 autosomal pairs and sex chromosomes similar to dom. pig. Others are rob. transl. of chr. no. 13-16, 15-17	Bosma (1978); Melander and Hansen-Melander (1980)
<b>Potamochoerus</b> <i>Potamochoerus porcus</i> (Bush pig)	34	12 pairs of meta/submeta. autosomes; 4 pairs of acro. X-largest meta/submeta. Y-smallest submeta.	Melander and Hansen-Melander (1980); Bosma et al. (1991b)
<b>Hylchoerus</b> <i>Hylchoerus meinertzhageni</i> (Giant forest hog)	32	all autosomes and X are meta/submeta. Y still not analysed	Melander and Hansen-Melander (1980)
<b>Sus</b>			
<i>Sus scrofa</i>			
<i>Sus scrofa domestica</i> (Domestic pig)	38	Chromosome number same in all breeds of domestic pig hitherto studied	see Bosma et al. (1991b)
Wild pig	36 or 38	Polymorphic system present	
Asian wild pig	36	16/17 centric fusion translocation <sup>1</sup>	Tikhonov and Troshina (1975)
West (former USSR, Europe, USA, etc.)	36	15/17 centric fusion translocation <sup>1</sup>	McFee et al. (1966); Gropp et al. (1969); Rittmannsperger (1971); Gustavsson et al. (1973); Tikhonov and Troshina (1975); Bosma (1976); Popescu et al. (1980)
<i>Sus vittatus leucomystr</i> (Japanese wild pig)	38	Karyotype identical to domestic pig	Muramoto et al. (191965); Okamoto et al. (1982); Bosma et al. (1991a)
<i>Sus porcula salivatus</i> (Pigmy hog)	38	Karyotype similar to dom. pig centromeric region of acrocent. chromosomes have extra band	Bosma (1983)
<i>Sus verrucosus</i> Javan warty pig	38	Karyotype similar to dom. pig except chromosome 10 and Y	Bosma et al. (1991a)
<i>Sus barbatus</i> The bearded pig	38	Karyotype shows MANY similarities to dom. pig	Bosma et al. (1991b)
<i>Sus celebensis</i> Sulawesi warty pig	38	Karyotype similar to dom. pig except the Y chromosome	Bosma et al. (1991a)

<sup>1</sup>Involvement of chromosome 17 in both polymorphic systems of wild pigs indicates that centric fusion translocation has been involved in the evolution of the domestic pig (Gustavsson, 1990) and that such translocations have significantly influenced karyotype evolution within the family Suidae.

# Cattle

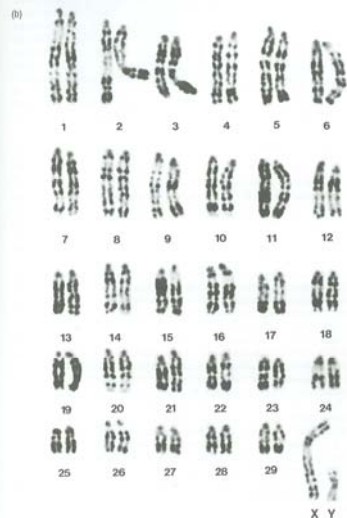
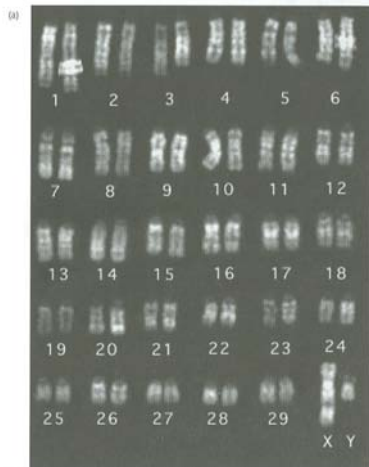


Fig. 10.2 (and opposite). (a) QFQ- and (b) RBG-banded cattle chromosomes arranged according to the revised ISCNDA ideogram.

# Cattle

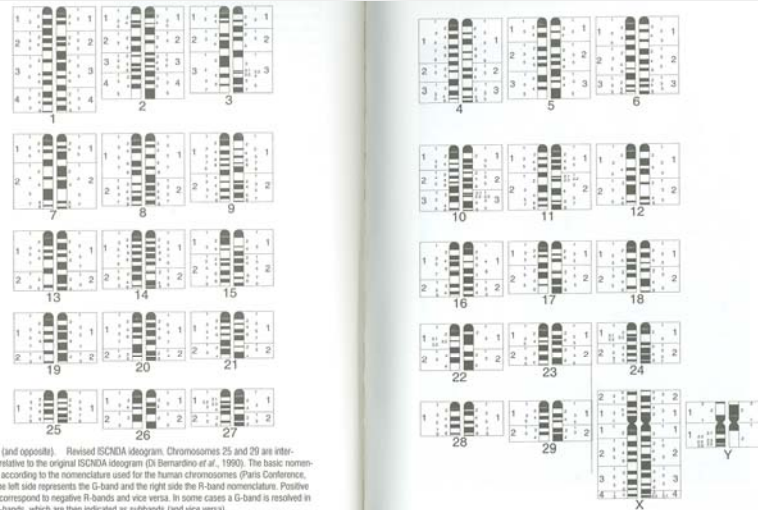


Fig. 18.1 (and opposite). Revised ISCNDA ideogram. Chromosomes 25 and 29 are interchanged relative to the original ISCNDA ideogram (Barnardino *et al.*, 1990). The basic nomenclature is according to the nomenclature used for the human chromosomes (Paris Conference, 1975). The left side represents the G-band and the right side the R-band nomenclature. Positive G-bands correspond to negative R-bands and vice versa. In some cases a G-band is resolved in several R-bands, which are then indicated as subbands (and vice versa).

Y of *Bos indicus* underwent inversion => acrocentric

# Sheep: (Differences between species often involve Robertsonian fusions)

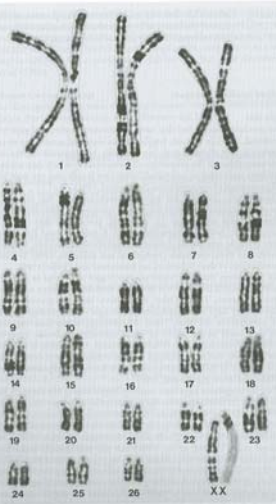


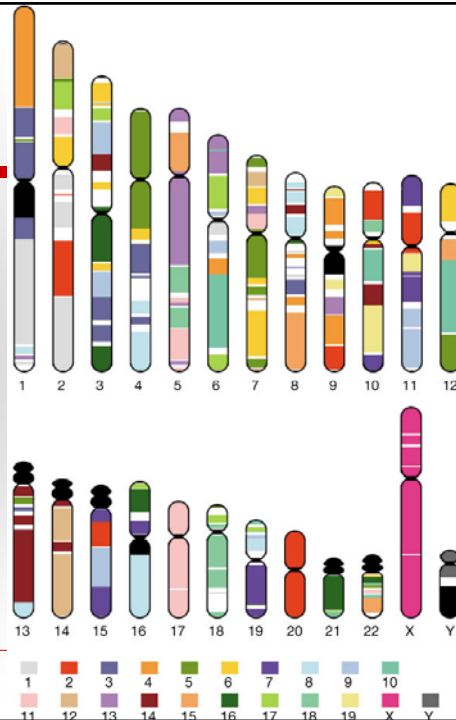
Fig. 10.1. An RBG-banded sheep karyotype (prepared by Dr Helene Hayes).

Texas standard group	U	Marker gene	Human chromosome	Length % *	Reading (GTG)	ISCNDA (GTG)	ISCNDA (DFO)	ISCNDA (RBA/RBG)	Sheep chromosome
1	U 10	SOD1	3, 21	5.87	1	1	1	1	1q
2	U 17	VIL	1p, 2q	5.12	2	2	2	2	2q
3	U 6	HSD3B	1p	4.71	3	3	3	3	1p
4	U 13	INHBA	7p	4.67	4	4	4	4	4
5	U 3	IFNG	1q, 12, 22	4.48	5	5	5	5	3q
6	U 15	CSN@	4	4.33	6	6	6	6	6
7	U 22	RASA	5q, 19p	4.18	7	7	7	7	5
8	U 18	IFNA	8p, 9q	4.13	8	8	8	8	2p
9	U 2	IGF2R	6q	3.86	9	9	9	9	8
10	U 5	CYP19	5q, 14, 15	3.67	10	10	10	10	7
11	U 16	LGB	2, 9q	3.94	11	11	11	11	3p
12	U 27	RB1	13	3.29	12	12	12	12	10
13	U 11	IL2RA	10p, 20	3.09	13	13	13	13	13
14	U 24	TG	8q	3.15	14	14	14	14	9
15	U 19	FSHB	5, 11p	3.11	15	15	15	15	15
16	U 1	PIGR	1q	3.07	16	16	16	16	12
17	U 23	FGG	4q, 12q, 22	2.83	17	17	17	17	17
18	U 9	GPI	16q, 19q	2.60	18	18	18	18	14
19	U 21	GH	17	2.54	19	19	19	19	11
20	U 14	MAP18	5	2.75	20	20	20	20	16
21	U 4	IGH@	14, 15	2.72	21	21	21	21	18
22	U 12	LTF	3	2.51	22	22	22	22	19
23	U 20	BOLA	6p	2.09	23	23	23	23	20
24	U 28	DSCI	18	2.37	24	24	24	24	23
25	U 8	ELN	7q, 16p	1.97					24
26	U 26	APT1	10q	1.96	26	26	26	26	22
27	U 25	DEFB@	8	1.83					26
28	U 29	CGN1	10q	1.73	28	27	27	28	25
29	U 7	LDHA	11	1.99					21
X		PGK1	X	5.45	X	X	X	X	X
Y		ZFY	Y	2.13	Y	Y	Y	Y	Y

\*Chromosome measurement expressed as relative length of the haploid genome.  
@ indicates a cluster of loci.

# Zoo-FISH

Conserved synteny between the human and mouse genomes. Regions from different mouse chromosomes (indicated by the colors of each mouse in B) show conserved synteny (gene order) with the indicated regions of the human genome (A). For example the genes present in the upper portion of human chromosome 1 (*orange*) are present in the same order in a portion of mouse chromosome 4. Regions of human chromosomes that are composed primarily of short, repeated sequences are shown in *black*. Mouse centromeres (indicated in *black* in B) are located at the ends of chromosomes; no known genes lie beyond the centromere on any mouse chromosome. For the most part, human centromeres, indicated by constrictions, occupy more internal positions on chromosomes (see Figure 4–11). (Adapted from International Human Genome Sequencing Consortium, *Nature* 409:860–921, 2001.)



## Overview

- Studying chromosomes
- The normal karyotypes of domestic animals
- Chromosome abnormalities*
- Chromosome abnormalities of domestic animals

## Chromosome abnormalities

- Types according to chromosome change:
  - *Aberrant chromosome number*
    - Aberrant euploidy (polyploidy)
    - Aneuploidy (trisomy, monosomy)
    - Mixoploidy
  - Aberrant chromosome structure (del, inv, dup, ins, R, mar, t, der)
  - Aberrant chromosome parental origin
- Types according to body extent:
  - Constitutional
  - Somatic (= > mosaic)

## Aberrant euploidy (polyploidy)

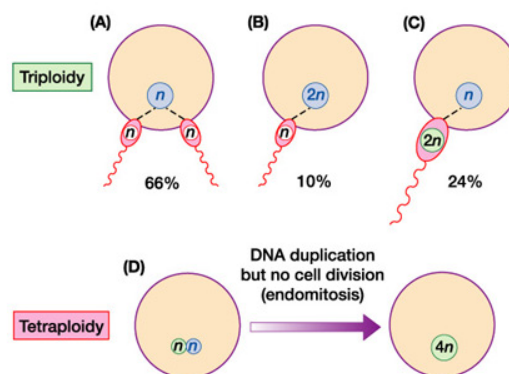


Figure 2-19 Human Molecular Genetics, 3/e. (© Garland Science 2004)

## Aneuploidy

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- ❑ Trisomy and monosomy
  - ❑ Chromosomal non-disjunction or anaphase lag
- 

## Consequences of numerical chromosomal abnormalities

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Table 2.4: Consequences of numerical chromosomal abnormalities.

<b>Polyploidy</b>	
Triploidy (69,XXX, XXY or XYY)	1–3% of all conceptions; almost never live born; do not survive
<b>Aneuploidy (autosomes)</b>	
Nullisomy (missing a pair of homologs)	Pre-implantation lethal
Monosomy (one chromosome missing)	Embryonic lethal
Trisomy (one extra chromosome)	Usually lethal at embryonic or fetal stages, but trisomy 13 (Patau syndrome) and trisomy 18 (Edwards syndrome) may survive to term and trisomy 21 (Down syndrome) may survive to age 40 or longer
<b>Aneuploidy (sex chromosomes)</b>	
Additional sex chromosomes	(47, XXX; 47, XXY; 47, XYY) present relatively minor problems, with normal lifespan
Lacking a sex chromosome	45,X = Turner syndrome. About 99% of cases abort spontaneously; survivors are of normal intelligence but infertile and show minor physical signs. 45,Y = not viable

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## Mixoploidy: Mosaicism versus chimerism

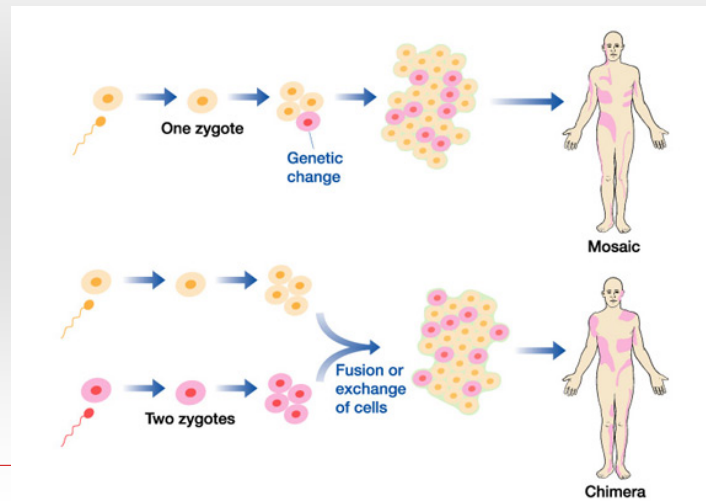


Figure 4-10 Human Molecular Genetics, 3/e. (© Garland Science 2004)

## Chromosome abnormalities

- Types according to chromosome change:
  - Aberrant chromosome number
    - Aberrant euploidy (polyploidy)
    - Aneuploidy (trisomy, monosomy)
    - Mixoploidy
  - *Aberrant chromosome structure* (del, inv, dup, ins, R, mar, t, der)
  - Aberrant chromosome parental origin
- Types according to body extent:
  - Constitutional
  - Somatic (= > mosaic)

# Nomenclature of chromosome abnormalities

## Box 2.4: Nomenclature of chromosome abnormalities.

### Numerical abnormalities:

Triploidy	69,XXX, 69,XXY, 69,XYY
Trisomy	e.g. 47,XX,+21 <sup>a</sup>
Monosomy	e.g. 45,X
Mosaicism	e.g. 47,XXX/ 46,XX

### Structural abnormalities:

<b>Deletion</b>	e.g. 46,XY,del(4)(p16.3) <sup>b</sup> ; 46,XX,del(5)(q13q33) <sup>b</sup>
<b>Inversion</b>	e.g. 46,XY,inv(11)(p11p15)
<b>Duplication</b>	e.g. 46,XX,dup(1)(q22q25)
<b>Insertion</b>	e.g. 46,XX,ins(2)(p13q21q31) <sup>c</sup>
<b>Ring</b>	e.g. 46,XY,r(7)(p22q36)
<b>Marker</b>	e.g. 47,XX,+mar <sup>d</sup>
<b>Translocation, reciprocal</b>	e.g. 46,XX,t(2;6)(q35;p21.3) <sup>e</sup>
<b>Translocation, Robertsonian</b>	e.g. 45,XY,der(14,21)(q10;q10) <sup>f</sup>
<b>(gives rise to one derivative chromosome)</b>	46,XX,der(14;21)(q10;q10),+21 <sup>g</sup>

### Notes:

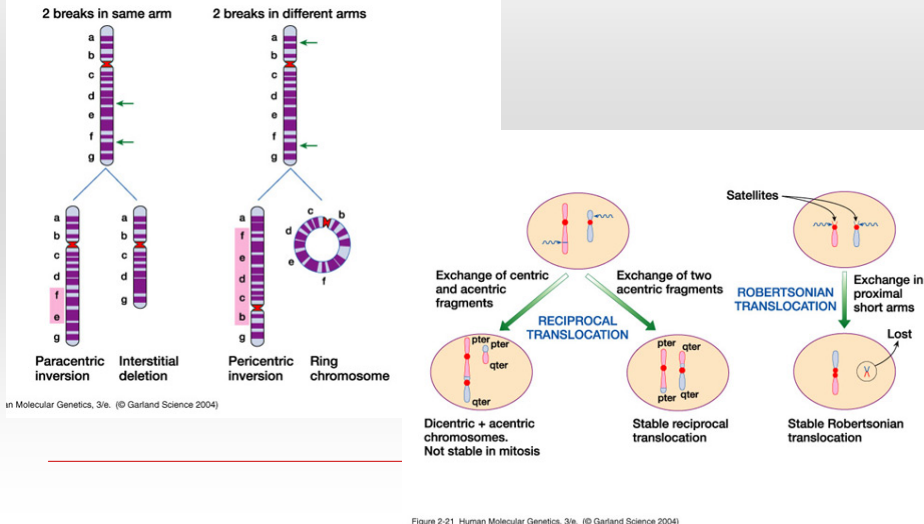
- <sup>a</sup>Gain of a chromosome is indicated by +; loss of a chromosome by -.
- <sup>b</sup>Terminal deletion (breakpoint at 4p16.3) and interstitial deletion (5q13-q33).
- <sup>c</sup>A rearrangement of one copy of chromosome 2 by insertion of segment 2q21-q31 into a breakpoint at 2p13.
- <sup>d</sup>Karyotype of a cell that contains a **marker chromosome** (an extra unidentified chromosome).
- <sup>e</sup>A balanced reciprocal translocation with breakpoints in 2q35 and 6p21.3.
- <sup>f</sup>A balanced carrier of a 14;21 Robertsonian translocation. q10 is not really a chromosome band, but indicates the centromere; **der** means **derivative chromosome** (used when one chromosome from a translocation is present).
- <sup>g</sup>Translocation Down syndrome; a patient with one normal chromosome 14, a Robertsonian translocation 14;21 chromosome and two normal copies of chromosome 21.
- This is a short nomenclature; a more complicated nomenclature is defined by the ISCN that allows complete description of any chromosome abnormality – see Further reading.

# Origin of structural abnormalities

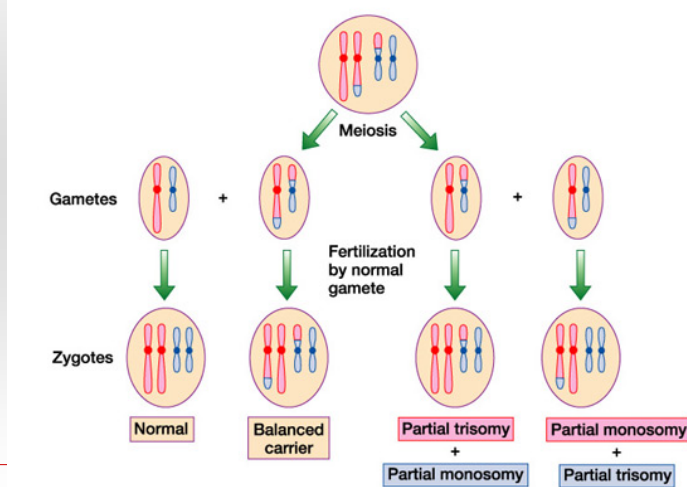
Table 2.5: Structural abnormalities resulting from misrepair of chromosome breaks or recombination between non-homologous chromosomes.

	One chromosome involved	Two chromosomes involved
One break	Terminal deletion (healed by adding telomere)	-
Two breaks	Interstitial deletion; Inversion;	Reciprocal translocation (Figure 2.21)
	Ring chromosome (Figure 2.20)	Robertsonian translocation (Figure 2.21)
	Duplication or deletion by unequal sister-chromatid exchange (Figure 11.7)	Duplication or deletion by unequal recombination (Figure 11.7)
Three breaks	Various rearrangements, e.g. inversion with deletion, intrachromosomal insertion	Interchromosomal insertion (direct or inverted)

# Origin of structural abnormalities



# Consequences of reciprocal translocations



## Consequences of Robertsonian fusions

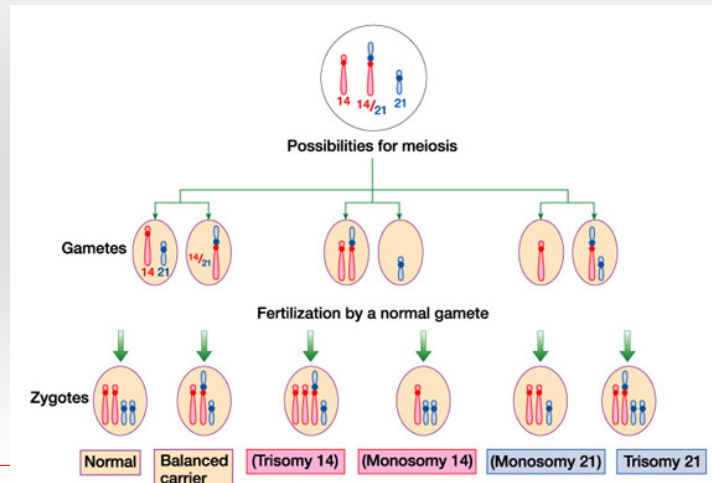


Figure 2-23 Human Molecular Genetics, 3/e. (© Garland Science 2004)

## Chromosome abnormalities

- Types according to chromosome change:
  - Aberrant chromosome number
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    - Aneuploidy (trisomy, monosomy)
    - Mixoploidy
  - Aberrant chromosome structure (del, inv, dup, ins, R, mar, t, der)
  - *Aberrant chromosome parental origin*
- Types according to body extent:
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  - Somatic (= > mosaic)

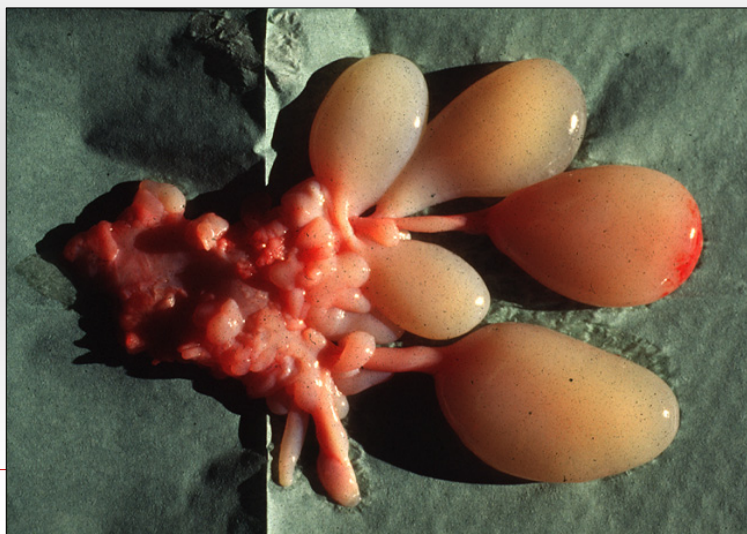
## Aberrant parental origin

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- Uniparental diploidy
    - Hydatiform moles
      - Paternal uniparental diploidy
      - Trophoblast hyperplasia
      - Risk of transformation in choriocarcinoma
      - Most moles are homozygous at all loci (⇔chromosome doubling from single sperm)
    - Ovarian teratoma
      - Maternal uniparental diploidy
      - Disorganized embryonic tissue
      - ⇔Activation of unovulated oocyte
- 

## Hydatiform mole

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## Aberrant parental origin

- Uniparental disomy:
  - Paternal or maternal
  - Isodisomy or heterodisomy
  - Matings between heterozygotes for reciprocal translocations
  - Trisomy or monosomy rescue
  - Anomalies if involved region contains imprinted genes.

## Parental imprinting

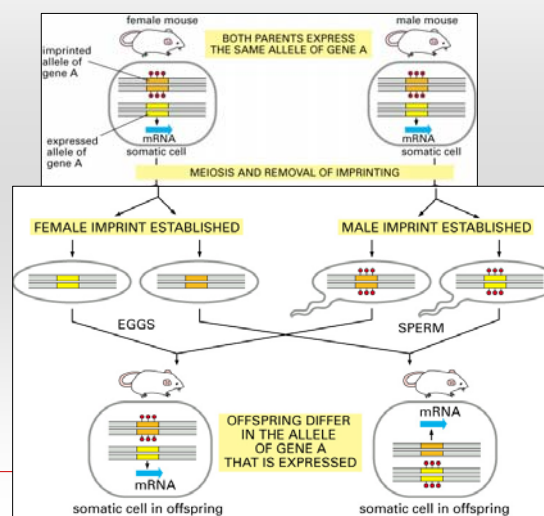


Figure 7-83 part 2 of 2, Molecular Biology of the Cell, 4th Edition

## Chromosome abnormalities

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- Types according to chromosome change:
    - Aberrant chromosome number
      - Aberrant euploidy (polyploidy)
      - Aneuploidy (trisomy, monosomy)
      - Mixoploidy
    - Aberrant chromosome structure (del, inv, dup, ins, R, mar, t, der)
    - Aberrant chromosome parental origin
  - Types according to body extent:
    - Constitutional
    - Somatic (= >mosaic)
- 

## Overview

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- Studying chromosomes
  - The normal karyotypes of domestic animals
  - Chromosome abnormalities
  - *Chromosome abnormalities of domestic animals*
-

# Pig

- Numerical aberrations
  - Cfr. Above
  - Chimerism
    - XX/XY chimerism
    - Does not necessarily results in intersexuality (vs bovine- Free-martinism)
- Structural aberrations
  - Reciprocal translocations
    - « ... at least 50% of breeding boars removed due to lower than average litter size carried reciprocal translocation. »
    - Other: cfr. Above
- Sex reversal / (pseudo-)hermaphroditism

# Reciprocal translocations of the pig

Translocation	Breakpoints	Breed	References*	Translocation	Breakpoints	Breed	References*
1q-11q+	1p11.5q25	LR Szw	Hansen-Melander and Melander (1970)	7q-9q	7q24.15q12		Ravasarmanana et al. (1992)
1p-9q-	1q11.16q11	LW	Lockstein (1974)	7q-15q-	8q23.14q27	LW	Konfortova et al. (1995)
1q-18q+	1p13.8q27	LR	Förster et al. (1981)	8p-14q	8q27.13q36		Ravasarmanana et al. (1992)
1p-30q+	1q23.14q21	Yo Szw	Gustavsson et al. (1982)	8q-13q	9q24.11q11		Ravasarmanana et al. (1992)
1q-14q-	1q25.14q15	SML	Golish et al. (1982)	9p-11q-	9q24.15q13	Yo Szw	Gustavsson et al. (1982)
1p-17q+	1q21.17q11	Yo Szw	Gustavsson (1984)	9p-15q-	11p1.5.15q13	LW x LF	Ducos et al. (1997)
1p-30q+	1q17.14q21	LW Ru	Konovalev et al. (1987)	11p-15q-	11p14.16q14	LR Fin	Henricsson and Blåsdén (1964)
1q-14q+	1q23.11q15	LW	Tarocco et al. (1987)				Gustavsson, quoted by Kuusikainen and Mäkinen (1988)
1p-11q+	1p25.15q13	LR Fin	Kuusikainen and Mäkinen (1988)	11p-16q-		LW x PI	Ducos et al. (1997)
1p-15q+	1q27.15q26	LR Fin	Kuusikainen and Mäkinen (1988)	11q-13q-		LW	Ducos et al. (1997)
1q-15q+	1q2.13.7q24	LW	Blaise et al. (1988)	12q-15q-	12q13.13q11	LW Ru	Konovalev et al. (1987)
1q-7q-	1p25.11p15	LR Szw	Gustavsson et al. (1988)	12q-13q-	13q21.14q27	Mimub	Astachova et al. (1990)
1p-11p+	BL	Tzschew (1900)		13q-14q+		Yo Szw	Aggelinos et al. (1976)
1q-18q+	1p11.5q11	Ha	Villagomez et al. (1990)		14q29.15q24		Golish and Riller (1990)
1p-5q-	1q2.11.10p15	LW Szw	Yang et al. (1992)	14q-15q-	15q26.16q21	Ha	Gustavsson and Jonsson (1992)
1q-15q	1q2.12.14q22		Ravasarmanana et al. (1992)	15q-16q-	15q13.17q21	Yo	Gustavsson et al. (1988)
1q-14q+			Zhang et al. (1992)	15q-17q+	16q23.17q21	LW	Ducos et al. (1997)
1p-30q+	1q12.6q22	LW	Ducos et al. (1997)	16q-17q-	Xq24.13q21	LR x Du	Popescu and Boscher (1986)
1q-9q+	2p13.15q24	Ga	Ducos et al. (1997)			LR x Viet	Astachova et al. (1991)
2p-15q	2p17.4q11	LR Fin	Mäkinen et al. (1987)	Xq-13q-		Ha	Gustavsson et al. (1989)
2p-4q-	2p14.14q23		Gustavsson, quoted by Kuusikainen and Mäkinen (1988)	Xp-14q-			Villagomez et al. (1990)
			Villagomez et al. (1993)	rec(1-14)(p-1-1)			Singh et al. (1994)
2p-14q+	3p13.7q21	LW	Popescu et al. (1983)				
3p-7q-	3p15.13q21	Indian	Sharma et al. (1991)				
3p-13q-	3p14.5q23	LW	Ducos et al. (1997)				
3p-5q+	4p11.14q11	LF	Ducos et al. (1997)				
4p-14q-	4q25.13q41	LW x LF	Popescu and Legault (1979)				
4q-13q-		LR Fin	Mäkinen and Rimes (1986)				
4q-15q+		PI	Blaise et al. (1988)				
4q-14q+	4q16.6q28	Indian	Sharma et al. (1991)				
4q-8p-	5q12.8q27	LW	Ducos et al. (1997)				
5q-8q+	5q11.14q-	Yo Szw	Gustavsson (1984)				
5q-14q-	5q25.15q25	HA x PI	Popescu et al. (1984)				
5q-14q+		LR	Parkanyi et al. (1992)				
6p-15q-	6p11.14q11	LR Fin	Bouters et al. (1974)				
6p-14q+	6p15.15q13	LW x Essex	Madan et al. (1978)				
6q-18q+	6q33.8q26	PI x LW	Bonneau et al. (1991)				
6q-18q-	6q11.16q11	Ga x Ma	Bonneau et al. (1991)				
6q-16q+	6q27.14q21	SML	Ducos et al. (1997)				
6q-14q-	6p15.13q41	LW x PI	Ducos et al. (1997)				
6p-13q-	7q21.11q11	SML	Ducos et al. (1997)				
7q-11q+	7q24.12q15	Yo Szw	Gustavsson et al. (1982)				
7q-12q+	7p13.13q21	Yo Fin	Kuusikainen and Mäkinen (1987)				
7p-13q-	7q35.17q11	Ha	Gustavsson et al. (1988)				
7q-17q-	7q13.8q27	Ha	Villagomez et al. (1995)				

Abbreviations: LR, Landrace; LF, French Landrace; LW, Large White; Yo, Yorkshire; PI, Pietrain; Ha, Hampshire; Ga, Gascon; Du, Duroc; MS, Meishan; BL, Belgian Landrace; SLM, Synthetic Male Line; Szw, Swedish; Fin, Finnish; Ru, Russian; Viet, local breed from Vietnam. It is expected that approximately another 10-15 new translocations will be added to this list in the very near future. (Courtesy, Dr Alain Ducos.)

\*Individual references cited in this table are not listed in the reference list but may be found in Ducos et al., 1997.

extra chromosomes and chromosome fragments (37,XY,-18/38,XY/39,XY+18 and 37,XY,-18/38,XY) have been described in liveborn individuals (Voigt et al., 1974). Furthermore, Bösch et al. (1985) incidentally discovered a pregnant true hermaphrodite, 38,XY/39,XX,+14 individual, which was mosaic in 29% of the cells examined.

Structural aberrations of the porcine chromosomes may be broadly grouped into translocations and duplications/deletions. During recent years, inversions have also been detected in pigs. Among translocations, reciprocal (rec) translocations are commonly observed in pigs. Of the more than 50 different translocations described in pigs (Table 8.2), approximately 50% have been detected in our laboratory by I. Gustavsson and co-workers during the past 20 years. Comparative analysis of various rec translocations in pigs (Table 8.2) shows that there are certain chromosomes which are preferentially involved in

## Pig

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- Numerical aberrations
    - Cfr. Above
    - Chimerism
      - XX/XY chimerism
      - Does not necessarily results in intersexuality (vs bovine- Free-martinism)
  - Structural aberrations
    - Reciprocal translocations
      - « ... at least 50% of breeding boars removed due to lower than average litter size carried reciprocal translocation. »
    - Other: cfr. Above
  - Sex reversal / (pseudo-)hermaphroditism
- 

## Bovine

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- Numerical aberrations
    - Cfr. Above
    - Chimerism
      - Male-female twins
      - XX/XY chimerisms of blood and haematopoeitic organs
      - Masculinization of females ⇔ AMH hormone of brother
  - Structural aberrations
    - Reciprocal translocations
      - T(1;29)
-

## Bovine recipr. translocations

Robertsonian translocation	Breed	Country	Method of chromosome identification	Reference
1:4		Czechoslovakia	M	Lojda <i>et al.</i> (1976)
1:7				Cited by Frank and Robert (1981)
1:21	Holstein Friesian	Japan	G	Miyake <i>et al.</i> (1991)
1:23		Czechoslovakia	M	Lojda <i>et al.</i> (1976)
1:25	Piebald	Germany	G	Stranzinger and Friester (1976)
1:26	Holstein Friesian	Japan	G	Miyake and Kaneda (1987)
1:28		Czechoslovakia	M	Lojda <i>et al.</i> (1976)
1:29	Different breeds	Different countries		Gustavsson and Rockborn (1964)
2:8	Friesian	England	G	Pollock (1974)
2:27				Yu and Xin (1991)
3:4	Limousine	France	R	Popescu (1977)
3:27	Friesian	Romania	M	Samarineanu <i>et al.</i> (1977)
4:4		Czechoslovakia	M	Lojda <i>et al.</i> (1975)
4:8	Chianina	Italy	R	De Giovanni <i>et al.</i> (1988)
4:10	Blonde d'Aquitaine	France	G, R, C	Balvi-Darwich <i>et al.</i> (1993)
5:18	Simmental	Hungary	G	Papp and Kovacs (1980)
5:21	Japanese Black	Japan	G	Masuda <i>et al.</i> (1978)
5:22	Polish Red	Poland	G	Syta and Slota (1992)
5:23	Bone Roumaine	Romania	M	Samarineanu <i>et al.</i> (1977)
6:16	Dexter	England	G	Logan and Harvey (1978)
6:28		Czechoslovakia	M	Lojda <i>et al.</i> (1976)
7:21	Japanese Black	Japan	G	Hanada <i>et al.</i> (1981)
8:9	Brown Swiss	Switzerland	M	Tschudi <i>et al.</i> (1977)
8:23	Ukrainian Grey	Russia	G, R, C	Bilbaeva <i>et al.</i> (1994)
9:23	Blonde D'Aquitaine	France	G, R	Cribiu <i>et al.</i> (1989)
11:16	Simmental	Hungary	G	Kovacs (1975)
11:22		Czechoslovakia	M	Lojda <i>et al.</i> (1976)
12:12	Simmental	Germany	G	Herzog and Hohn (1984)
12:15	Holstein Friesian	Argentina	G	Roldan <i>et al.</i> (1984)
13:21	Holstein Friesian	Hungary	M	Kovacs <i>et al.</i> (1973)
13:24	Red and White	Poland	G	Slota <i>et al.</i> (1988)
14:19	Braunvieh	Switzerland	G	Stranzinger (1989)
14:20	Simmental	England	G	Logan and Harvey (1978)
14:21	Simmental	Hungary	G	Kovacs and Szepeshegyi (1977)
14:24	Podolian	Italy	R	Di Bernardino <i>et al.</i> (1979)
14:28	Holstein Friesian	USA	G	Elsworth <i>et al.</i> (1979)
15:25	Barrosa	Portugal	G, G + C, G, R	Iannuzzi <i>et al.</i> (1992)
16:18	Barrosa	Portugal	G, R	Iannuzzi <i>et al.</i> (1993a)
16:19	Marchigiana	Italy	R, C	Malerba (1997)
16:20	Ger. Red Pied x Czech. Red Pied	Czechoslovakia	R, C	Rubes <i>et al.</i> (1996)
16:21	Ger. Red Pied x Czech. Red Pied	Czechoslovakia	R, G	Rubes <i>et al.</i> (1992)
19:21	Holstein Friesian	France	G	Pinton <i>et al.</i> (1997)
20:20	Simmental	Germany	G	Herzog and Hohn (1984)
21:27	Blonde d'Aquitaine	France	R, G, NOR	Berland <i>et al.</i> (1988)
24:27	Holstein hybrid	Egypt	G, C	Mahrous <i>et al.</i> (1994)
25:27	Grey Alpine	Italy	R	De Giovanni <i>et al.</i> (1979)

M, chromosome measurement; C, G, NOR, R: banding method.

## Chromosomal abnormalities in domestic sheep



Table 10.3. Chromosomal abnormalities in domestic sheep (*Ovis aries*).

Abnormality	Chromosomes	First report
Centric-fusion translocations	$t_1(6;24)^*$	Bruere (1969)
	$t_2(9;10)^*$	Bruere and Mills (1971)
	$t_3(7;25)^*$	Bruere <i>et al.</i> (1972)
	$t_4(5;8)$	Pearce <i>et al.</i> (1994a)
	$t_5(8;22)$	Pearce <i>et al.</i> (1994a)
Reciprocal translocations	$1p^-;19q^+†$	Glahn-Luft and Wassmuth (1978)
	$13q^-;19q^+‡$	Anamthawat-Jonsson <i>et al.</i> (1992)
	$2q^-;3q$	Slota <i>et al.</i> (1986)
Autosomal aneuploidy	53,XY/54,XY	Dunn and Roberts (1972)
Sex-chromosome aneuploidy	53,XO	Zartman <i>et al.</i> (1981)
	53,XO/54,XX	Baylis <i>et al.</i> (1984)
	55,XXY	Bruere <i>et al.</i> (1969a)
	54,XY/55,YYY	Moraes <i>et al.</i> (1980)
Deletions	Numerous	See Long (1990)
Autosomal duplication	20§	Matejka and Cribiu (1989)
Intersaxes	54,XX	Dain (1972)
Freemartins	54,XX/54,XY	Germeke (1967)
Testicular feminization	54,XY	Bruere <i>et al.</i> (1969b)
True hermaphrodites	54,XX	Fayrer-Hosken <i>et al.</i> (1992)

Chromosomes are numbered according to the 1995 (Texas) standard. Original designation for:  $t_1 = (5;26)$  and  $t_2 = (8;11)$  in Bruere *et al.* (1974). The numbers cited in the table are from Ansari *et al.* (1993a) (which are consistent with those of the 1995 standardization).

†  $1p^-;19q^+ = 1p^-;19q^+$  in Glahn-Luft and Wassmuth (1978).

‡  $13q^-;19q^+ = 13q^-;20q^+$  in Anamthawat-Jonsson *et al.* (1992).

§ 20 = 19 in Matejka and Cribiu (1989).

## Association between Polled and intersexuality in goats

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- In goats, Polled (absence of horns) is inherited as an autosomal dominant trait.
- It is always associated with sex reversal in females which is however transmitted as an autosomal recessive trait.
- The PIS locus has been mapped to 1q43 and the mutation has been identified by positional cloning as an 11.7 Kb deletion which is probably affecting the expression of the nearby FOXL2 gene.

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<http://dga.jouy.inra.fr/lgbc/projets/PIS.html>