WHAT IS THE OVERALL ORGANIZATION OF THE HUMAN GENOME?

TABLE 9-1 Classification of Eukaryotic DNA

UNIQUE SEQUENCES (65%)

- Protein-coding genes
  - Solitary genes
  - Duplicated and diverged genes (functional gene families and nonfunctional pseudogenes)
- Tandemly repeated genes encoding rRNA, 5S rRNA, tRNA, and histones

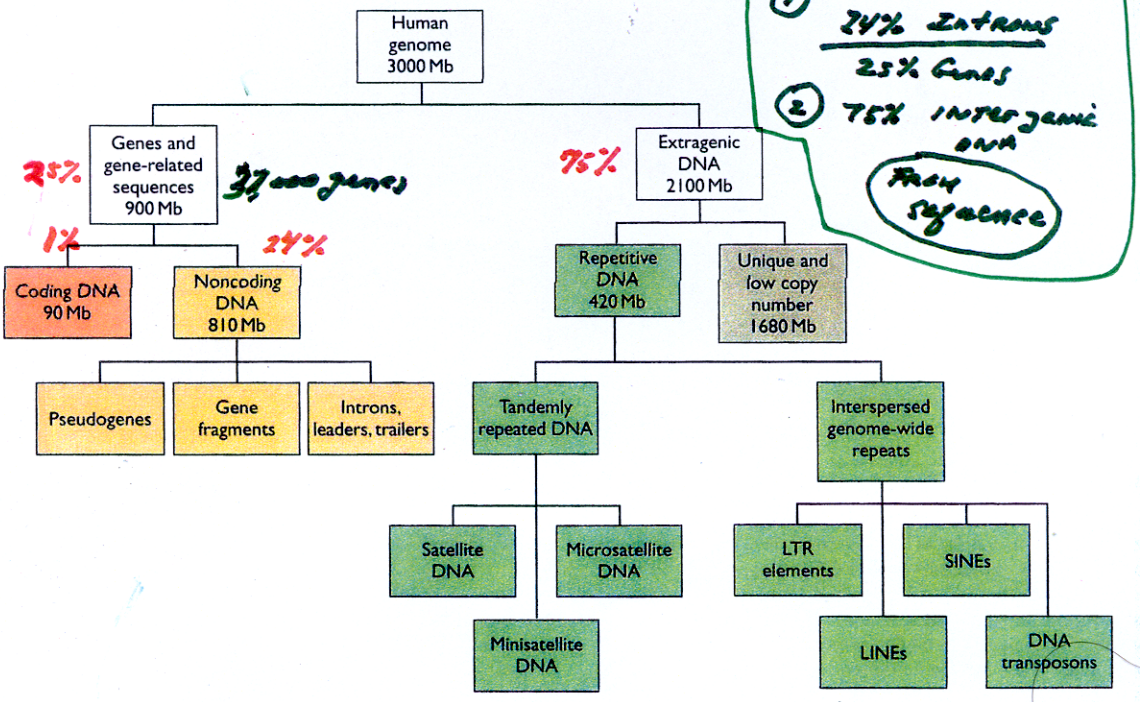
REPEATED SEQUENCES (35%)

- Repetitious DNA
  - Simple-sequence DNA (VNTR's & STR's)
  - Moderately repeated DNA (mobile DNA elements)
    - Transposons
    - Viral retrotransposons
    - Long interspersed elements (LINES; nonviral retrotransposons)
    - Short interspersed elements (SINES; nonviral retrotransposons)
- Unclassified spacer DNA

→ Genes!
ONLY 1% Exons
24% Introns
Remaining intergenic

→ NO KNOWN FUNCTION!
Includes VNTRs!

Box 6.4: The organization of the human genome



Based on Strachan and Read (1996).

THE HUMAN GENOME CONTAINS DIFFERENT CLASSES OF REPEATED SEQUENCES

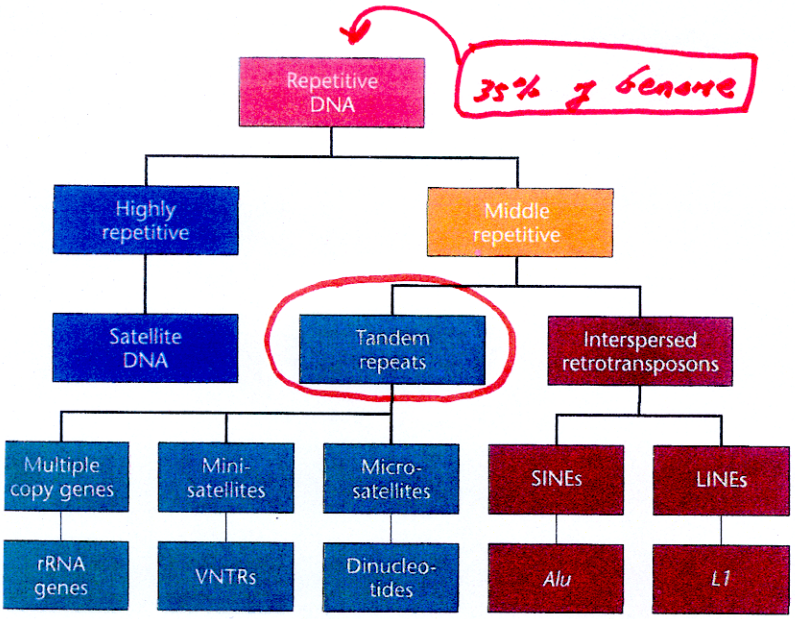


FIGURE 19.16 An overview of the various categories of repetitive DNA.

VNTRs useful for individual identity

STRs as well

Table 7.11: Major classes of tandemly repeated human DNA

USEFUL AS VNTRS

Class	Size of repeat	Major chromosomal location(s)
'Megasatellite' DNA (blocks of hundreds of kb in some cases)	several kb	Various locations on selected chromosomes
RS447	4.7 kb	~50–70 copies on 4p15 plus several copies on distal 8p
untitled	2.5 kb	~400 copies on 4q31 and 19q13
untitled	3.0 kb	~50 copies on the X chromosome
Satellite DNA (blocks often from 100 kb to several Mb in length)	5–171 bp	Especially at centromeres
α (alphoid DNA)	171 bp	Centromeric heterochromatin of all chromosomes
β (*Sau3* A family)	68 bp	Centromeric heterochromatin of 1, 9, 13, 14, 15, 21, 22 and Y
Satellite 1 (AT-rich)	25–48 bp	Centromeric heterochromatin of most chromosomes and other heterochromatic regions
Satellites 2 and 3	5 bp	Most, possibly all, chromosomes
Minisatellite DNA (blocks often within the 0.1–20 kb range)	6–64 bp	At or close to telomeres of all chromosomes
telomeric family	6 bp	All telomeres
hypervariable family	9–64 bp	All chromosomes, often near telomeres
Microsatellite DNA (blocks often less than 150 bp)	1–4 bp	Dispersed throughout all chromosomes

VNTRs STRs

HUMAN DNA SEQUENCE ORGANIZATION

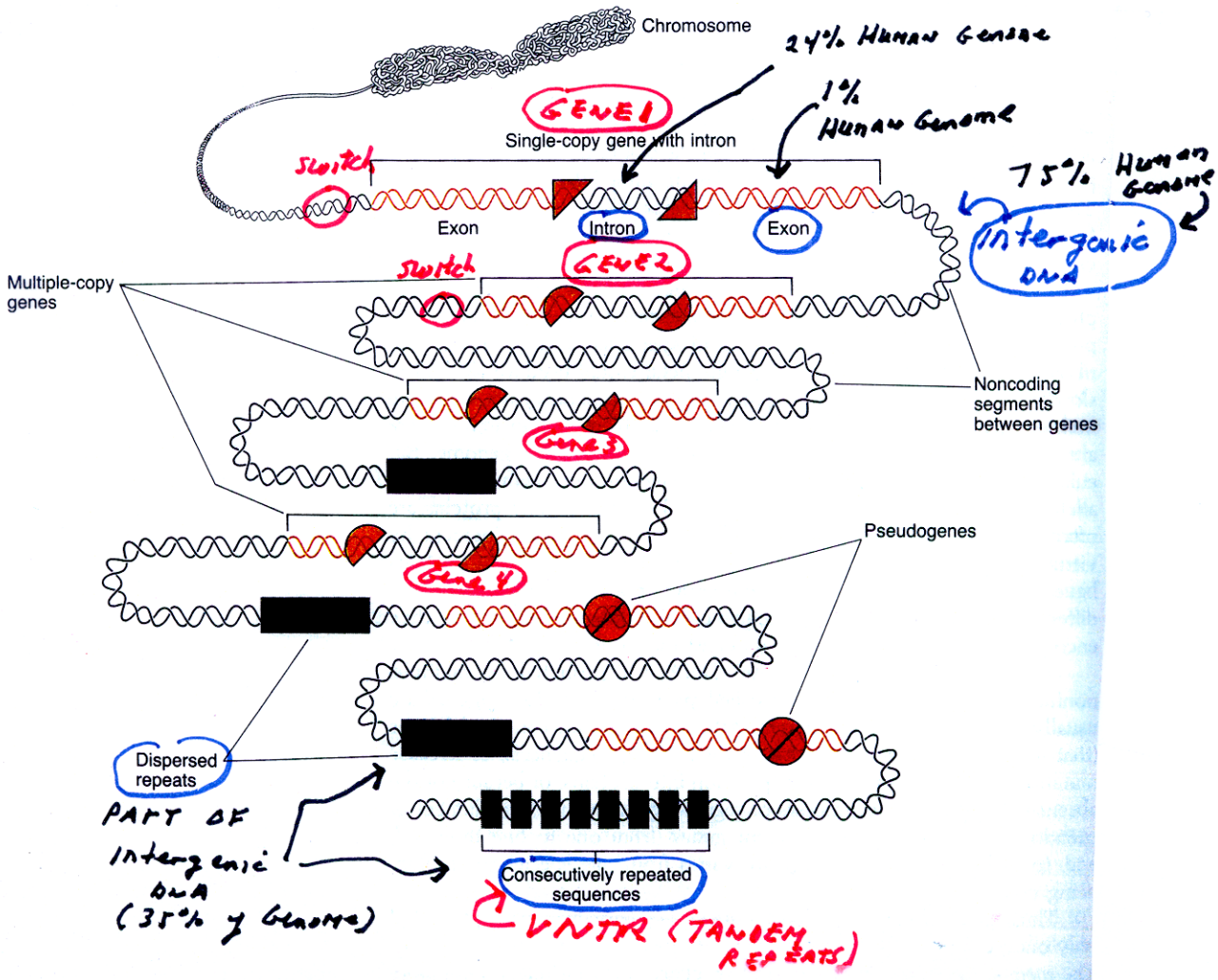


Figure 7.1 Occurrence of different kinds of unique and repeated DNA segments on chromosomal DNA.

CONTINUOUS STRETCH OF GENES + INTERGENIC REGIONS!

VNTRs Are Tandem Repeats & Give Rise to Allelic Variability

Cleavage sites Conserved

Variable # Tandem Repeats

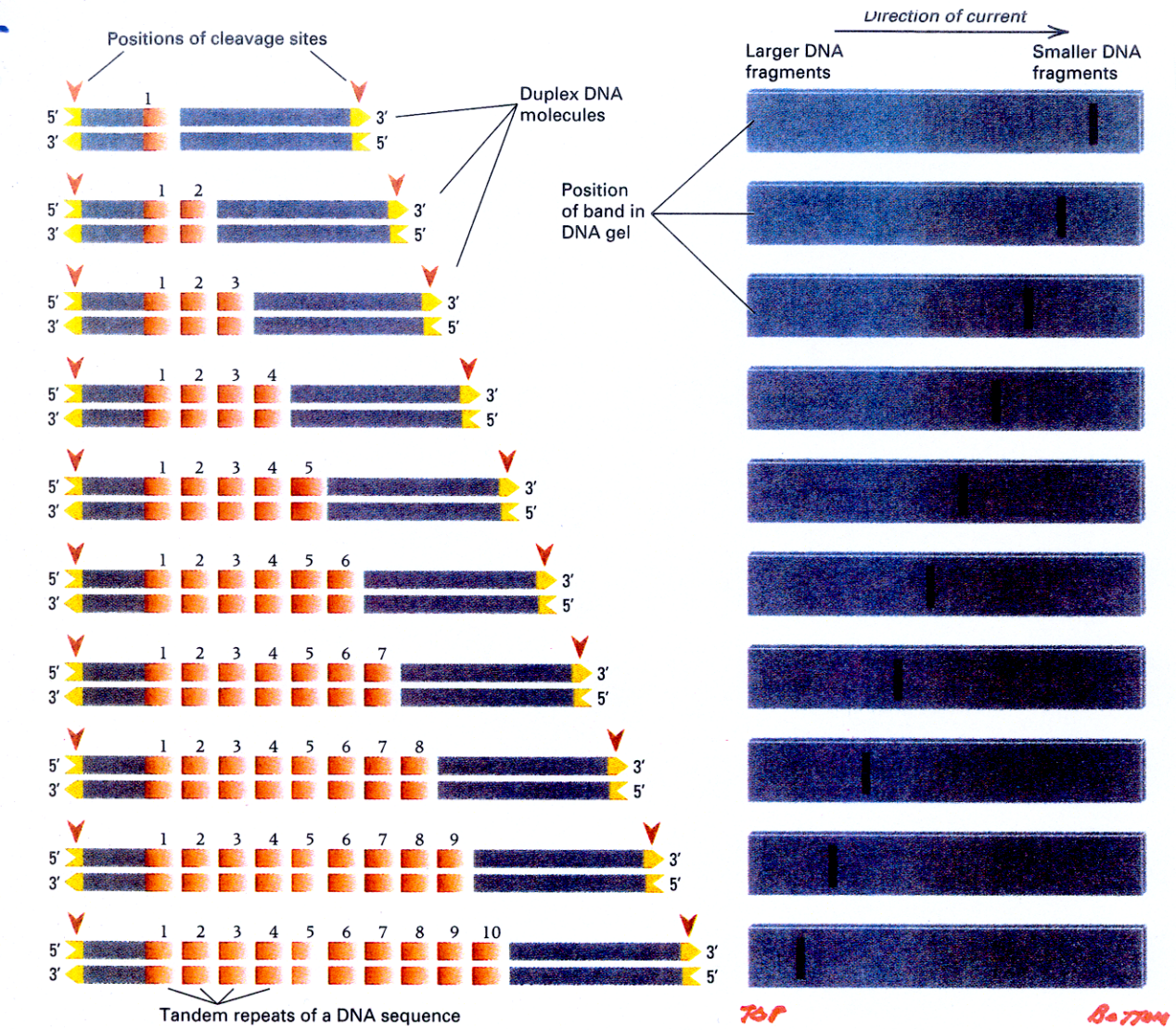


Figure 2.28 In a simple tandem repeat polymorphism (STRP), the alleles in a population differ in the number of copies of a short sequence (typically 2–60 bp) that is repeated in tandem along the DNA molecule. This example shows alleles in which the repeat number varies from 1 to 10. Cleavage at restriction sites flanking the STRP yields a unique fragment length for each allele. The alleles can also be distinguished by the size of the fragment amplified by PCR using primers that flank the STRP.

Size Varies Between Conserved Regions Like an Accordion — AT SAME LOCUS or CHROMSOME Location

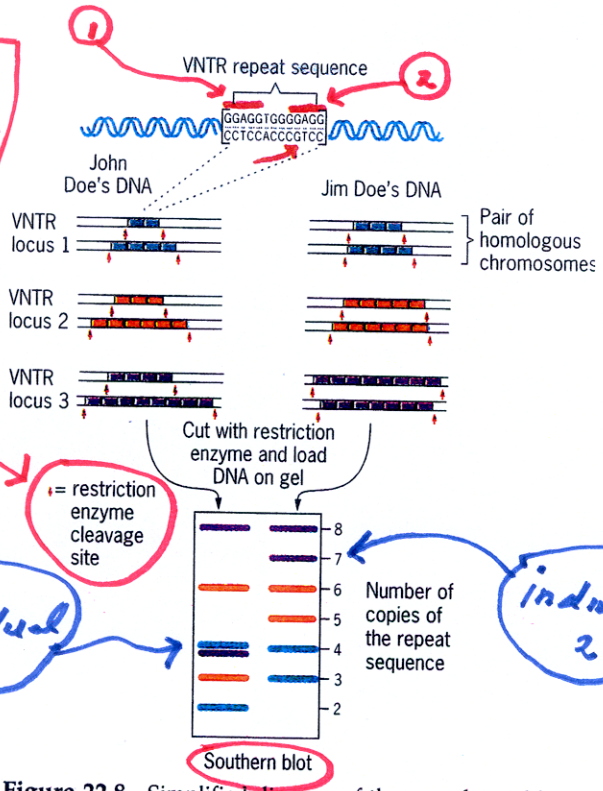
ANALOGOUS TO DISC FINGERPRINTS

VNTRs Are Sequence-Specific Tandem Repeats Present Throughout the Genome

BLOT METHOD

conserved flanking Restriction Sites

Repeat = (GGAGG) (GGAGG)$_n$



How was this experiment done?

individual 1

individual 2

MANY different types! Differ in Sequence & location!

Figure 22.8 Simplified diagram of the use of variable number tandem repeats in preparing DNA fingerprints.

VARY in Repeat Length (2bp & 4bp!)

2bp + 4bp!

VNTRs Generally Have Many Different Alleles at a Given Locus

Repeat = $(CA)_n$

PCR METHOD

Population of Alleles!

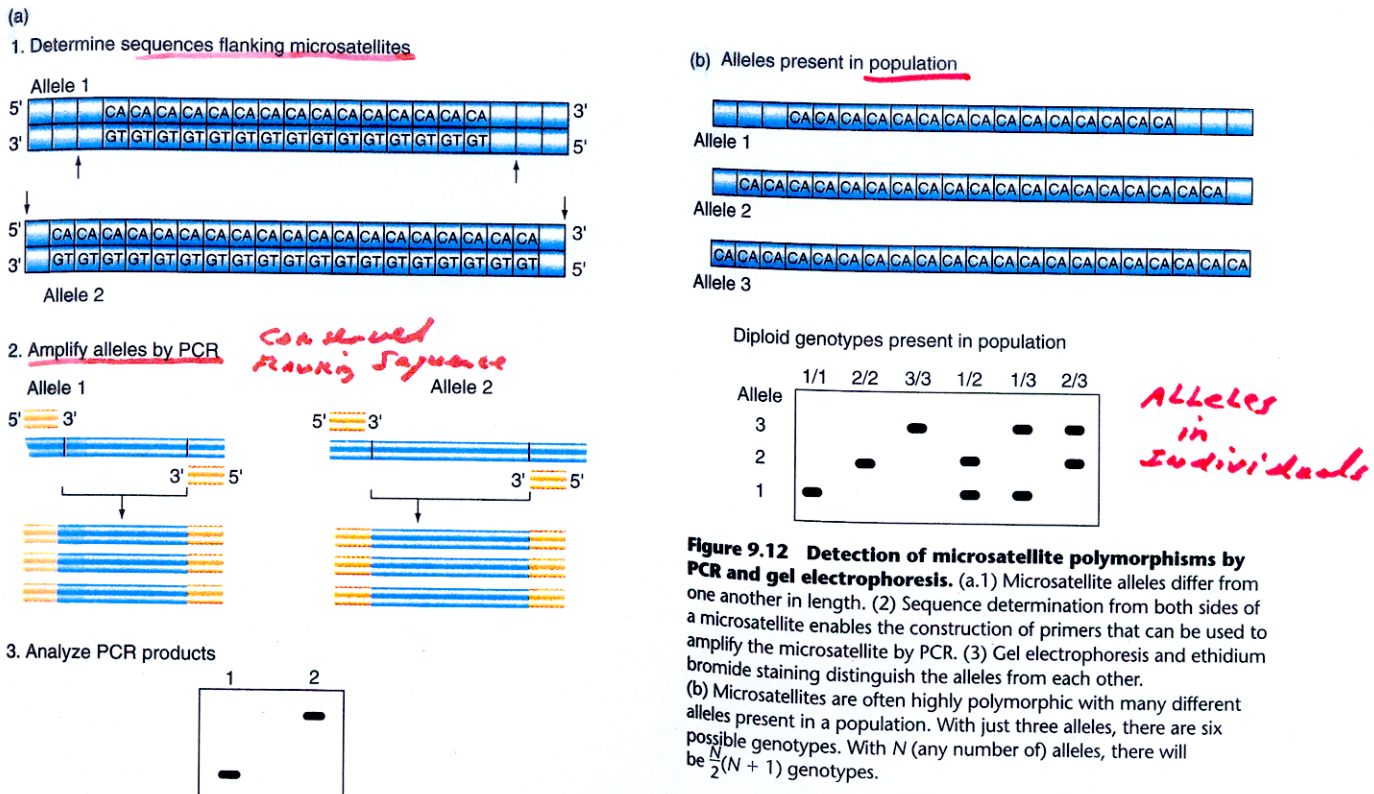


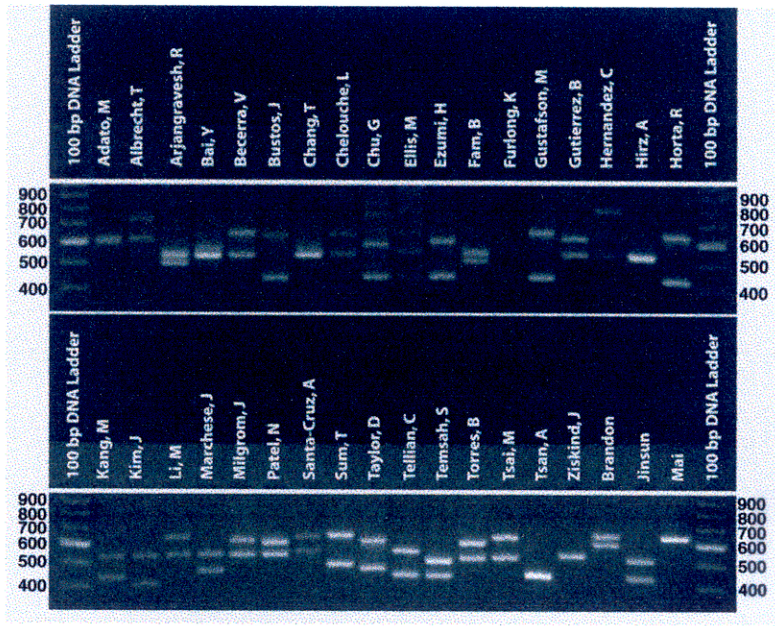
Figure 9.12 Detection of microsatellite polymorphisms by PCR and gel electrophoresis. (a.1) Microsatellite alleles differ from one another in length. (2) Sequence determination from both sides of a microsatellite enables the construction of primers that can be used to amplify the microsatellite by PCR. (3) Gel electrophoresis and ethidium bromide staining distinguish the alleles from each other. (b) Microsatellites are often highly polymorphic with many different alleles present in a population. With just three alleles, there are six possible genotypes. With N (any number of) alleles, there will be $\frac{N}{2}(N+1)$ genotypes.

Useful for comparing Individuals & Populations (e.g., HCTOA) are there races?

Method Used in HCTOA Class!

D1S80 VNTR ALLELES IN HC70AL CLASS

Figure 1. ***D1S80*** **Alleles in the Winter, 2004 HC70A UCLA Class Population.**

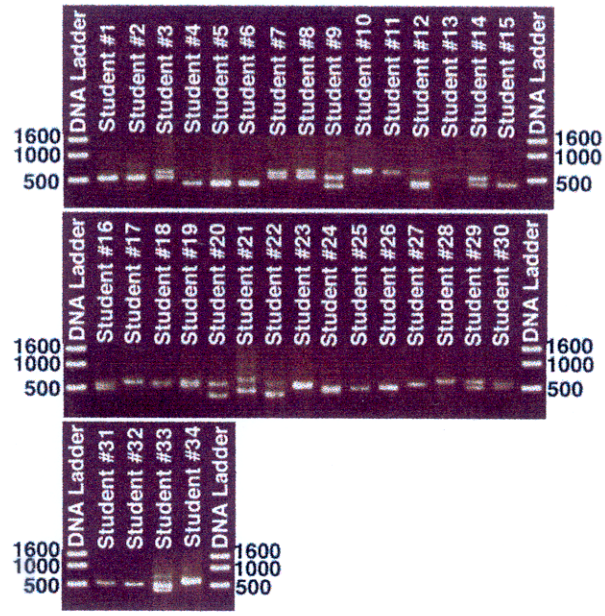


PCR Approach as Shown on Previous Page

8-15 ALLELES

CORE REPEAT SEQUENCE = 16bp!

Figure 2. ***D1S80*** **Alleles in the Winter, 2004 HC70A Kyoto Class Population.**



VNTR D1S80 ALLELES VARY in Different HUMAN Populations

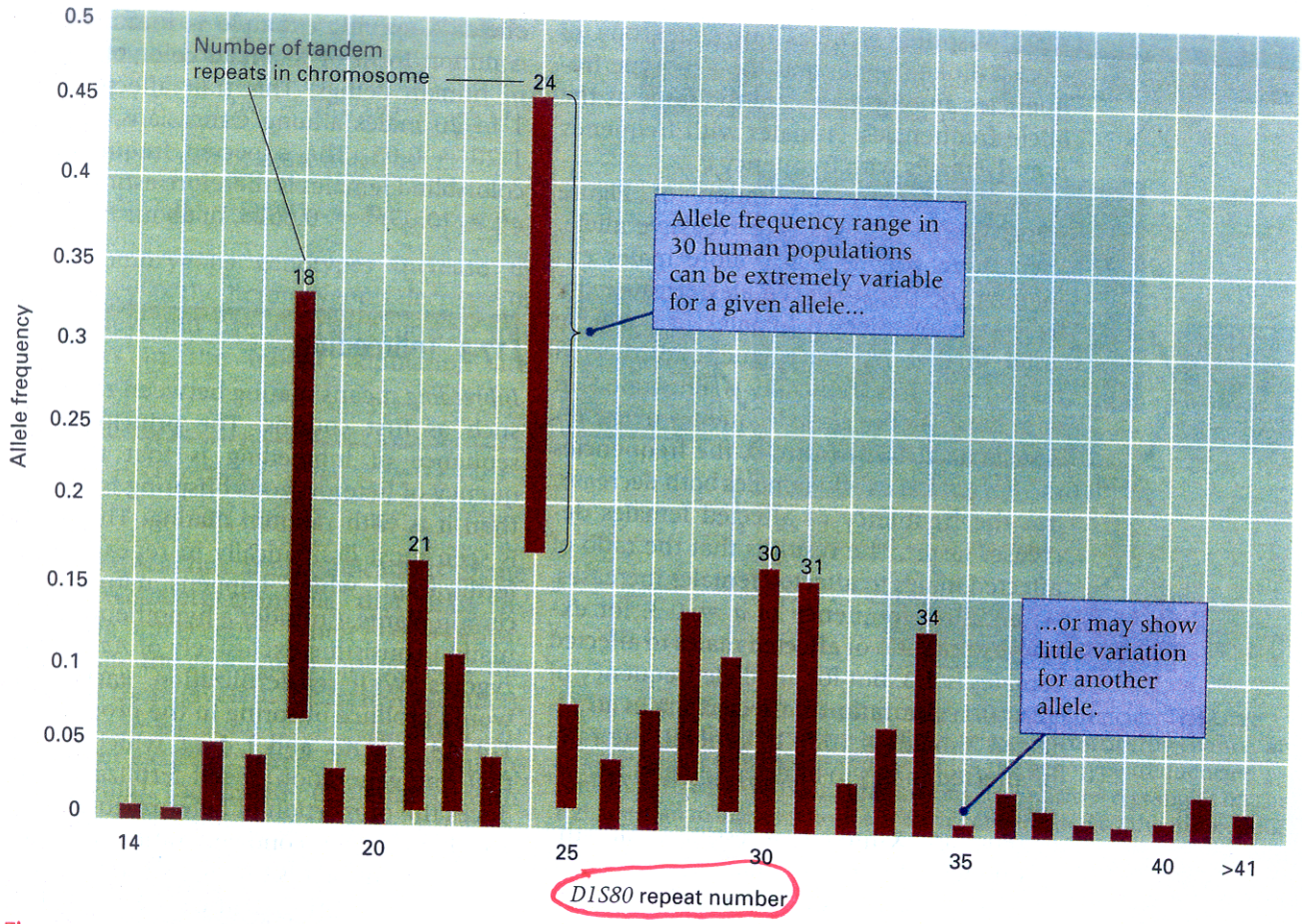
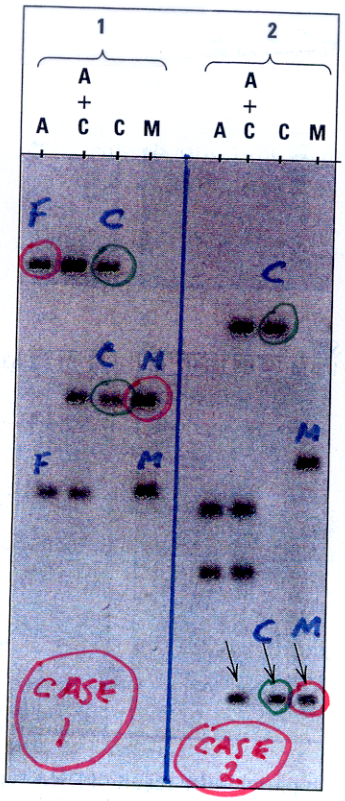


Figure 17.15 Range of allele frequencies found among human subpopulations for the VNTR *D1S80*. [Data from B. Budowle et al. *J. Forensic Science* 1995. 40:38.]

USING VNTR LOCI IN PATERNITY CASES

one allele shared with child

yes A is OK



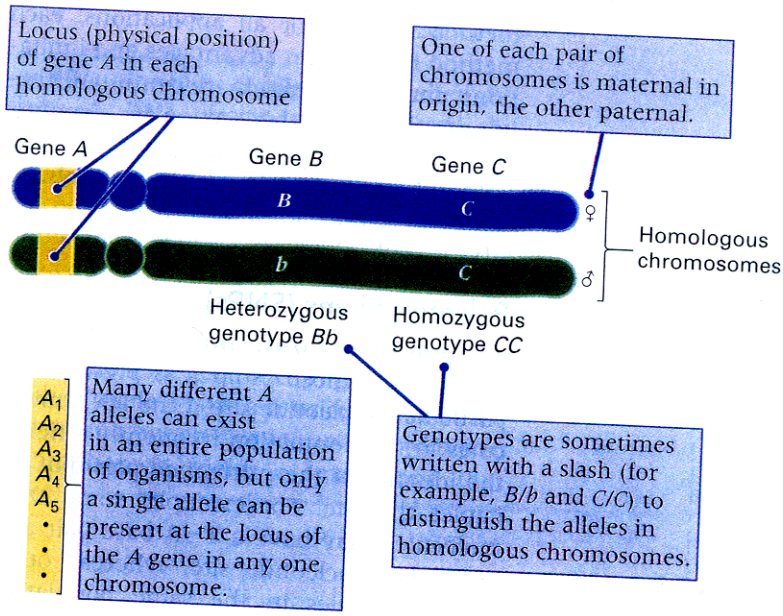
NO A is not OK

no allele shared with child

Figure 17.14 Use of DNA typing in paternity testing. The sets of lanes numbered 1 and 2 contain DNA samples from two different paternity cases. In each case, the lanes contain DNA fragments from the following sources: M, the mother; C, the child; A, the accused father. The lanes labeled A + C contain a mixture of DNA fragments from the accused father and the child. The arrows in case 2 point to bands of the same size that are present in lanes M, C, and A + C. Note that the male accused in case 2 could not be the father because neither of his bands is shared with the child. [Courtesy of R. W. Allen.]

RECALL --- RELATIONSHIP BETWEEN CHROMOSOMES, ALLELES, Individuals, & Populations

VNTR Locus

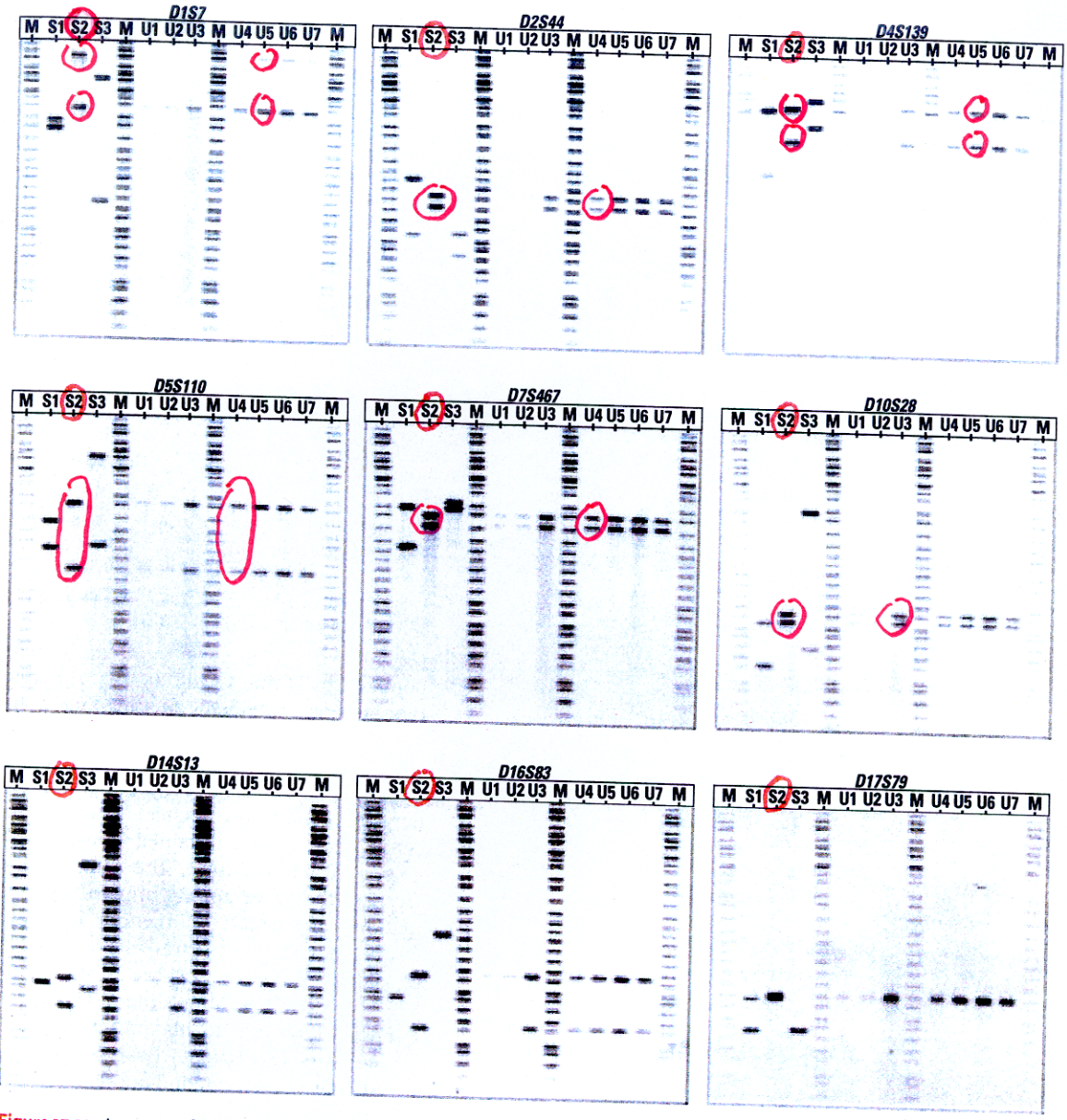


MANY VNTR alleles

Figure 2.22 Key concepts and terms used in modern genetics. Note that a single gene can have any number of alleles in the population as a whole, but no more than two alleles can be present in any one individual.

Multiple Single-Locus VNTRs used in a Criminal Case

U = Semen from Rape Victim 7 victims



NOTE! Power of using many different VNTR markers!

Figure 17.13 An example of DNA typing in a criminal case. Each panel is the result of DNA typing for a different VNTR. The lanes marked S1, S2, and S3 contain DNA from blood samples of three male suspects; those in columns U1 through U7 contain DNA from semen samples collected from seven female victims of rape. The lanes marked M contain molecular-weight markers. In each case, the DNA from suspect S2 matches the samples obtained from the victims. [Courtesy of Steven J. Redding, Office of the Hennepin County District Attorney, Minneapolis, and Lowell C. Van Berkom and Carla J. Finis, Minnesota Bureau of Criminal Apprehension.]

Who done it!!

But also who is innocent?

ORIGINS OF VNTR VARIABILITY

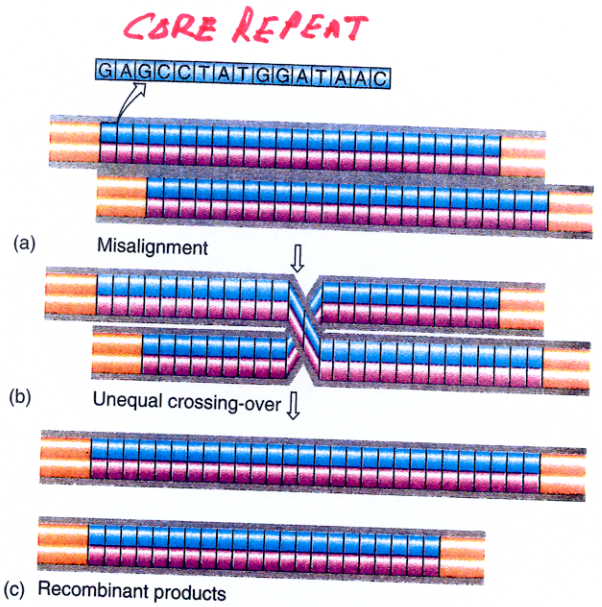
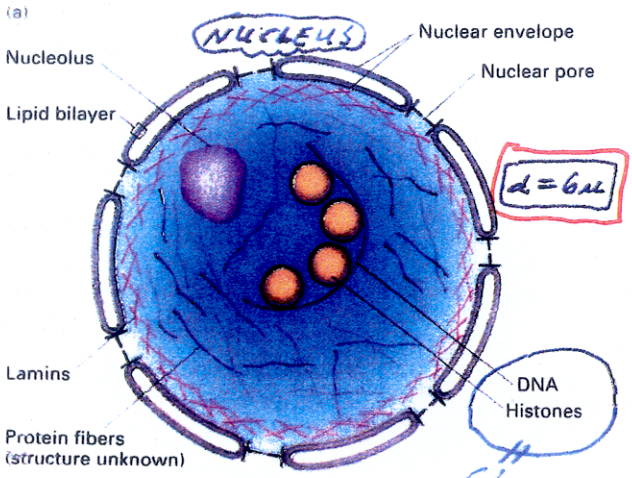


Figure 9.4 Minisatellites are highly polymorphic because of their potential for misalignment and unequal crossing-over. Minisatellites are composed of relatively long tandem repeating units of identical sequence. (a) Misalignment and (b) unequal crossing-over produce (c) recombinant products that contain different numbers of repeating units than either parental locus; each new recombinant product is a new allele.

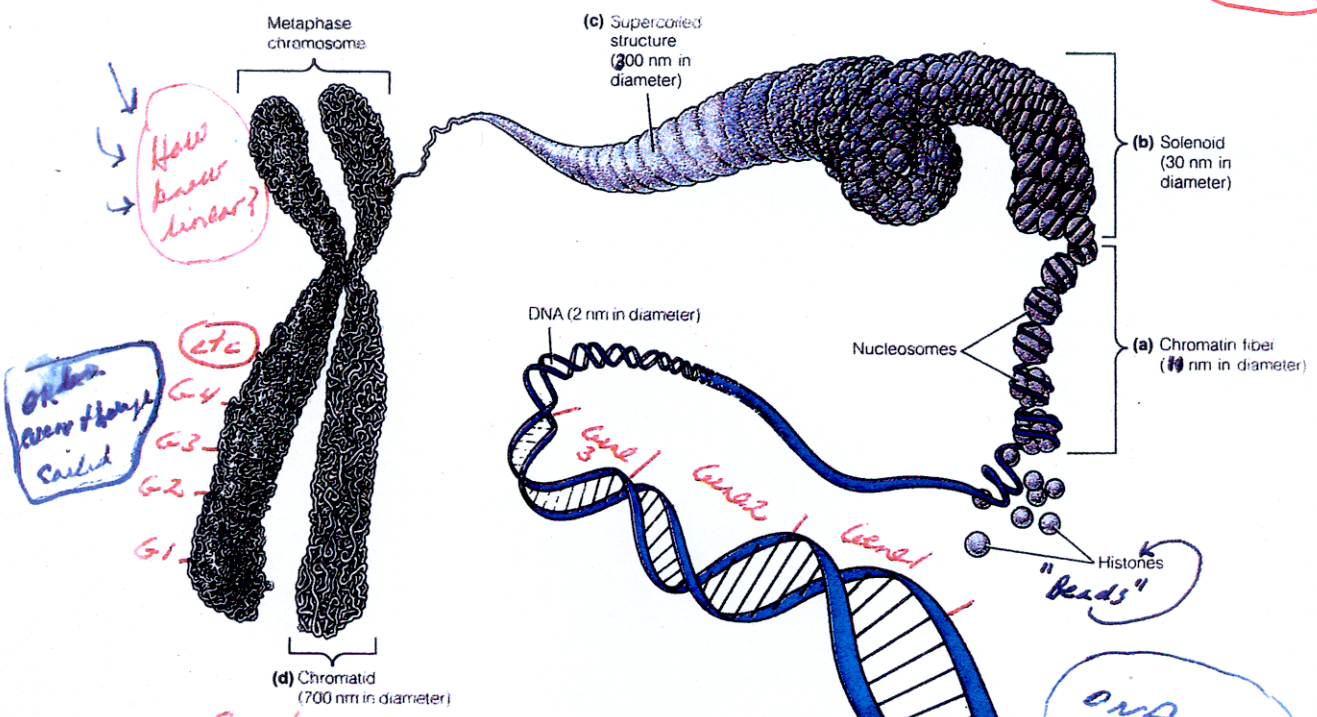
During Crossing over in Meiosis

The HUMAN GENOME IS PACKAGED INTO CHROMOSOMES



◀ **Figure 8-60** (a) Generalized diagram of eukaryotic nucleus showing identifiable structural elements. (b) Electron micrograph of nuclear pore–lamin complexes isolated from rat nuclei. Nuclear pores *(arrows)* are embedded in fibrous lamin proteins (la). (c) A transmission electron micrograph of a whole mount of a HeLa cell, showing a skeletal network within the nucleus. The cell was prepared by removing lipids and soluble factors with a mild detergent. The remaining skeletal struture was then treated to remove most of the DNA. The sample was fixed with glutaraldehyde, but no heavy-metal shadowing was done. [See S. Penman et al., 1982, *Cold Spring Harbor Symp. Quant. Biol.* 46:1013.] *Photograph (b) courtesy of N. Dwyer. Reproduced from the* Journal of Cell Biology, *1976, by copyright permission of Rockefeller University Press. Photograph (c) courtesy of S. Penman.*

Nucleus = 6μ diameter
DNA = 6×10^6 m in length!



each chromatid is a duplicate of the other

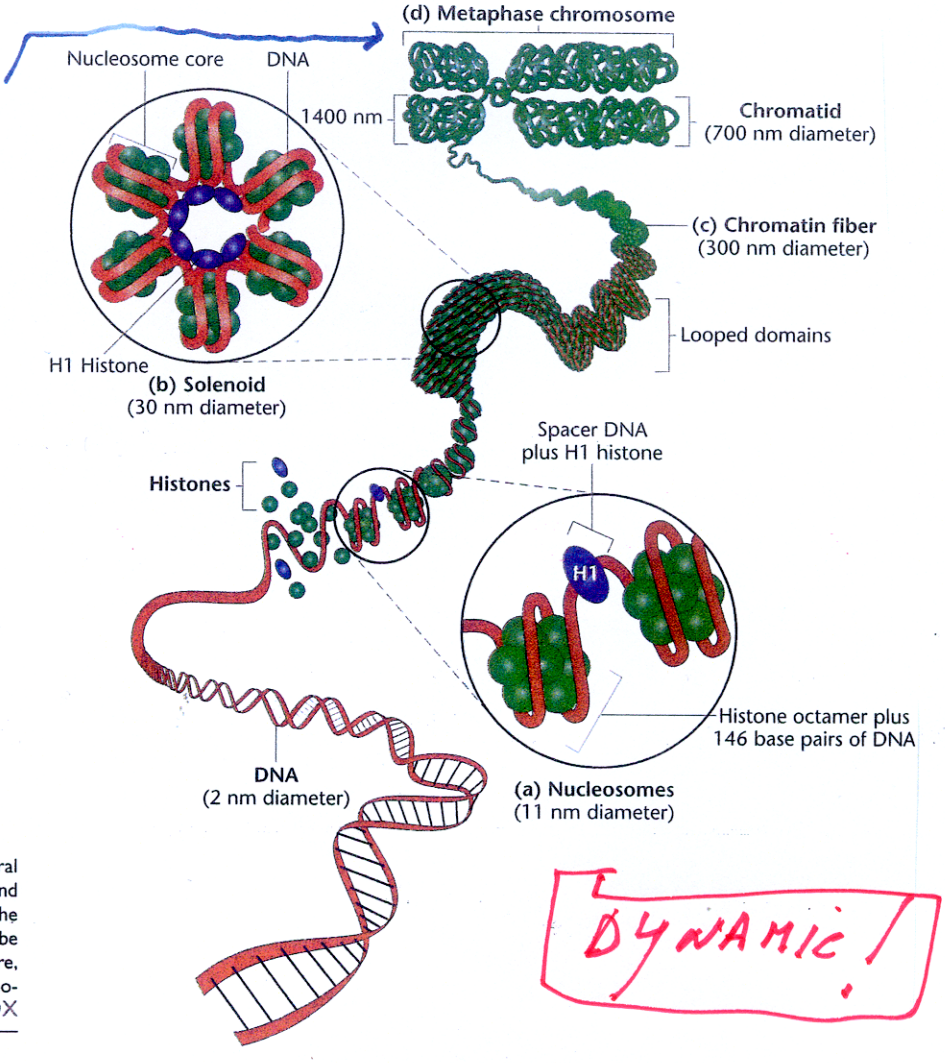
Hundreds of Genes per chromosome

DNA tightly coiled why?

DYNAMIC Coiling Chromosomes

Figure 28.10
Levels of chromatin structure. The beaded string structure is a 11-nm fiber, which folds into a "solenoidal" 30-nm fiber with about six nucleosomes per turn. This can further fold to form thick 200-nm fibers that can be observed in electron micrographs of chromosomes or nuclei.

HISTONE PROTEINS INTERACT WITH DNA TO MAKE A CHROMOSOME



DNA division occured?

DYNAMIC!

FIGURE 19.12 General model of the association of histones and DNA in the nucleosome, illustrating the way in which the chromatin fiber may be coiled into a more condensed structure, ultimately producing a mitotic chromosome. GenCDX

Significance of coiling?

CHROMOSOMES CAN BE CHARACTERIZED USING A MICROSCOPE AND CONSTRUCTING A Karyotype

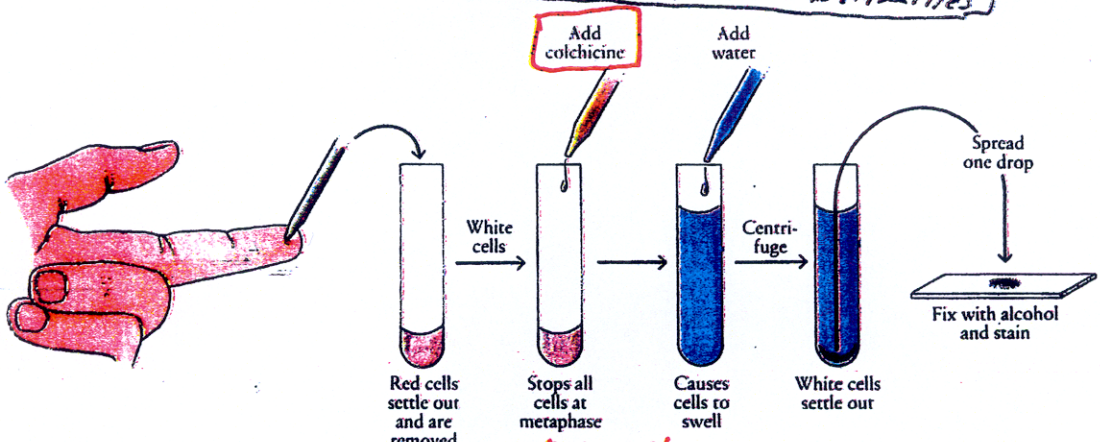
Preparation of a Karyotype

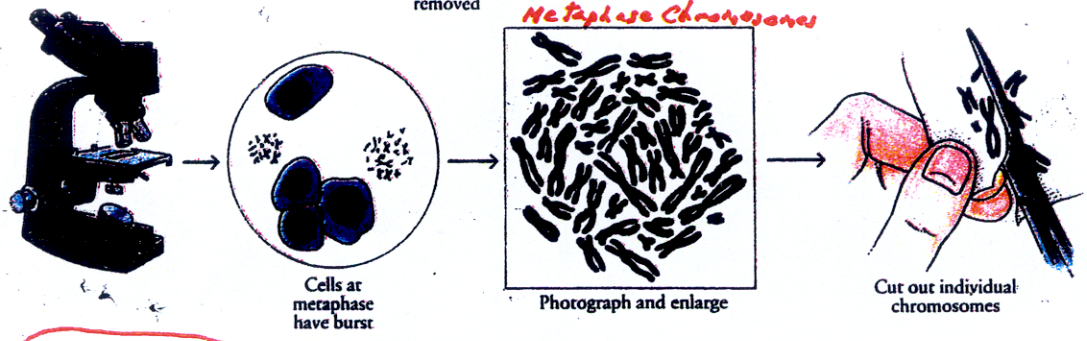
AT METAPHASE

why "short" then?

Chromosome typing for the identification of gross chromosomal abnormalities is being carried out at an increasing number of genetic counseling centers throughout the United States. The result of the procedure is a graphic display of the chromosome complement, known as a karyotype. The chromosomes shown in a karyotype are mitotic metaphase chromosomes, each consisting of two sister chromatids held together at their centromeres. To prepare a karyotype, cells in the process of dividing are interrupted at metaphase by the addition of colchicine, a drug that prevents the subsequent steps of mitosis from taking place by interfering with the spindle microtubules. After treating and staining, the chromosomes are photographed, enlarged, cut out, and arranged according to size. Chromosomes of the same size are paired according to centromere position, which results in different "arm" lengths. From the karyotype, certain abnormalities, such as an extra chromosome or piece of a chromosome, can be detected.

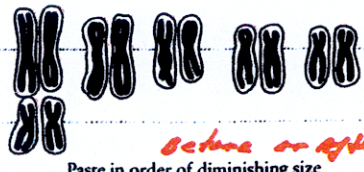
Karyotypes are needed to detect Chromosomal abnormalities





C- Metaphase Chromosomes

colchicine Metaphase!

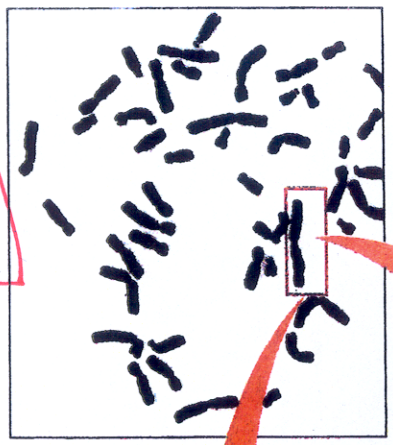


Paste in order of diminishing size with centromere on pencil line

before or after DNA replication?

2 Chromatids after DNA replication = 2 DNA molecules

CHROMOSOMES HAVE STRUCTURES That ARE VISIBLE in Light and Electron Microscopes

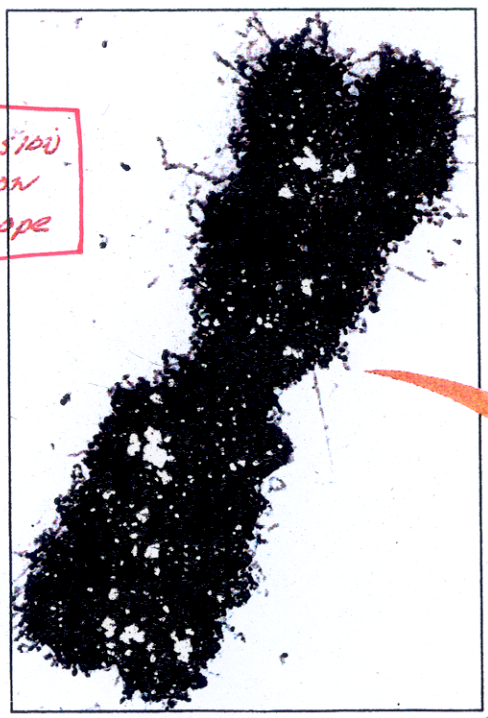


Light MICROSCOPE

ight micrograph of
uman chromosomes
enlarged 600 times)

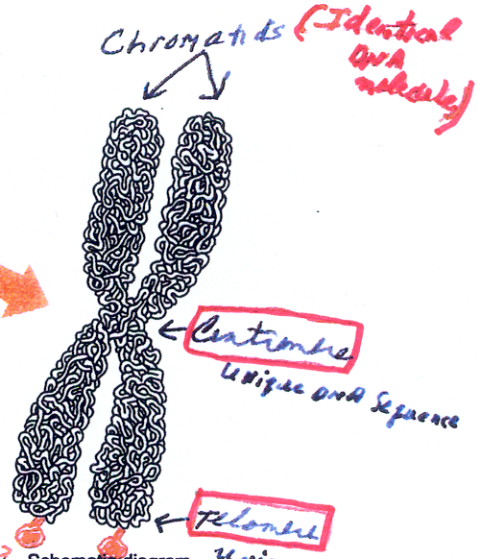
Electron micrographic 3-D image
(enlarged 30,000 times)

SCANNING Electron MICROSCOPE



TRANSMISSION Electron MICROSCOPE

Electron micrograph
of fixed chromosome
(enlarged 30,000 times)



repetitive DNA → "satellite"

one chromatid / one DNA molecule

Figure 1.3 Human chromosomes.

Chromatids
centromere
telomere

A chromosome during division

EACH CHROMOSOME HAS A UNIQUE MORPHOLOGY + BANDING PATTERN

22 PAIRS OF AUTOSOMES ♀ X ♂ Y = 24 Pieces of DNA!

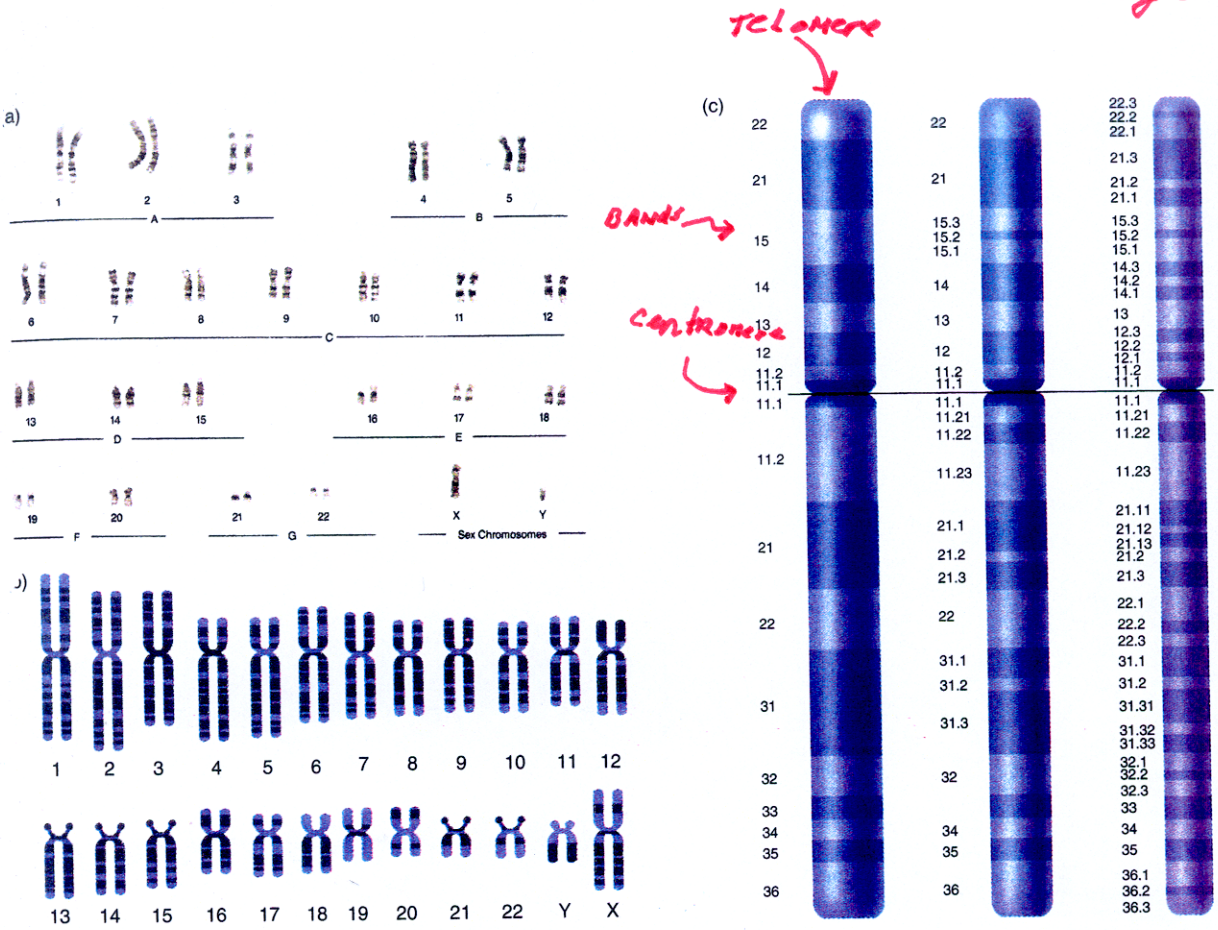


Figure 10.3 The human karyotype: Banding distinguishes the chromosomes. (a) Photograph of a complete set of human chromosomes at metaphase. Staining with Giemsa dye accentuates the bands and interbands. (b) Idiograms for the complete set of human chromosomes. An idiogram is an idealized diagram of the banding pattern associated with a stained chromosome. (c) Chromosome 7 at three different levels of banding resolution. As staining techniques improve, it becomes possible to resolve what previously appeared as a single band into a series of bands and interbands, producing more and more bands along each chromosome. Thus, at one resolution, 7q31 appears as one band. At a slightly higher resolution, 7q31 becomes two bands (7q31.1 and 7q31.3) flanking an interband (7q31.2); and at an even higher resolution, 7q31.3 itself appears as two bands (7q31.31 and 7q31.33) and an interband (7q31.32).

What causes banding patterns of chromosomes to be unique?

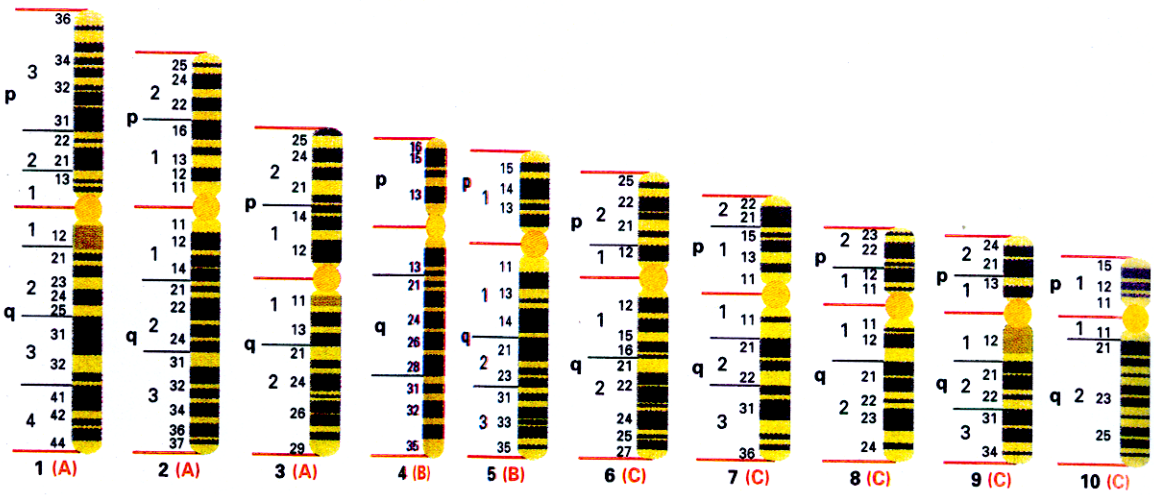
Size of bands?

SP

CHROMOSOME NOMENCLATURE

Table 9.1 Conventional karyotype symbols used in human genetics

A–G	Chromosome groups
1–22	Autosome designations
X, Y	Sex-chromosome designations
p	Short arm of chromosome
q	Long arm of chromosome
ter	Terminal portion: pter refers to terminal portion of short arm, qter to terminal portion of long arm
+	Preceding a chromosome designation, indicates that the chromosome or arm is extra; following a designation, indicates that the chromosome or arm is larger than normal
−	Preceding a chromosome designation, indicates that the chromosome or arm is missing; following a designation, indicates that the chromosome or arm is smaller than normal
mos	Mosaic
/	Separates karyotypes of clones in mosaics—e.g., 47, XXX/45,X
dup	Duplication
dir dup	Direct duplication
inv dup	Inverted duplication
del	Deletion
inv	Inversion
t	translocation
rcp	Reciprocal translocation
rob	Robertsonian translocation
r	Ring chromosome
i	Isochromosome (two identical arms attached to a single centromere, like an attached-X chromosome in *Drosophila*)



BANDING PATTERNS CAN BE USED TO DISTINGUISH CHROMOSOMES & LOCATE GENES

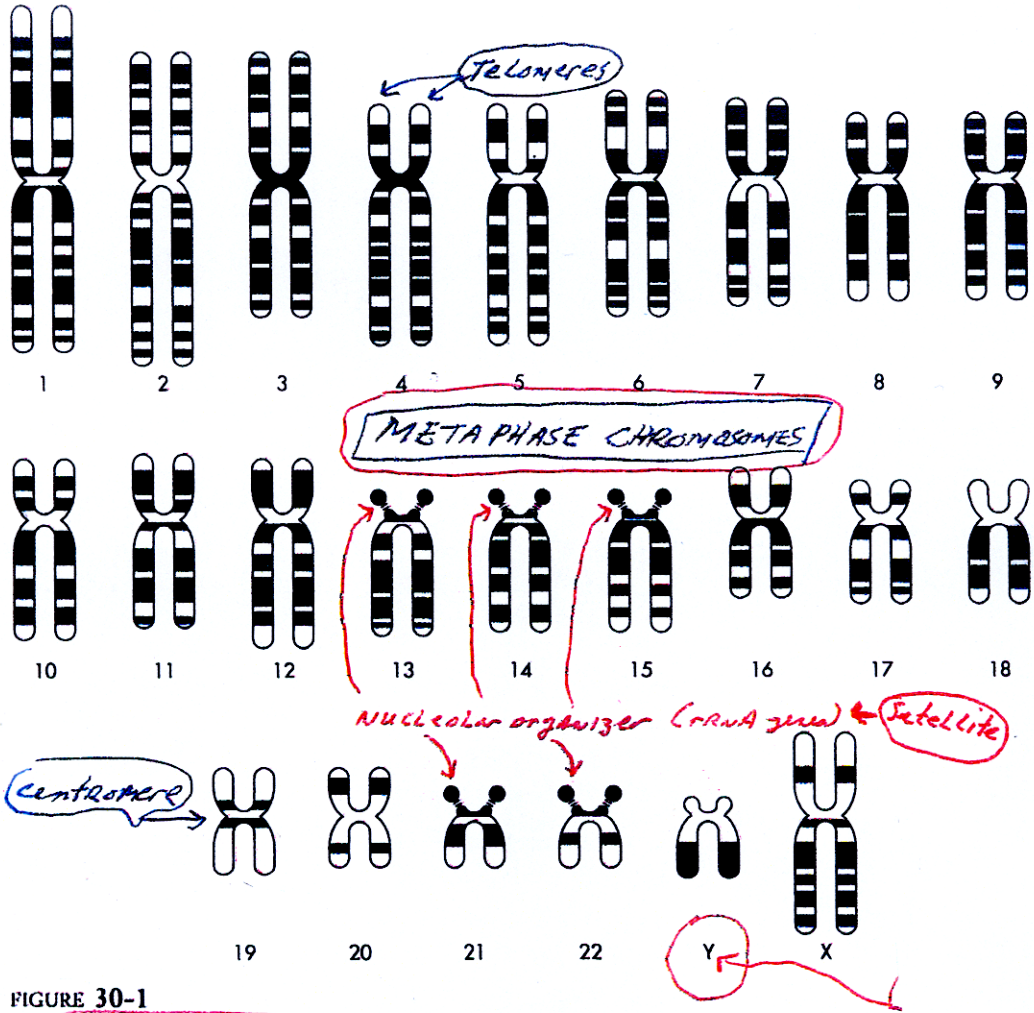
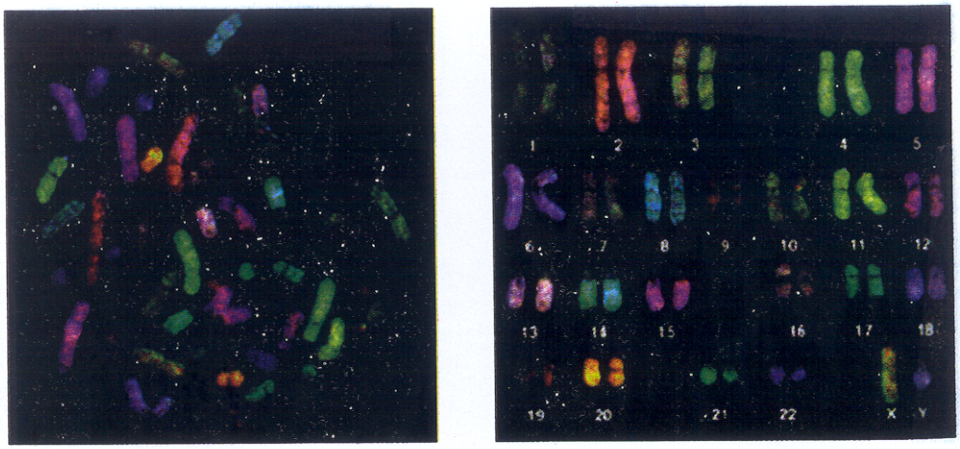


FIGURE 30-1
The haploid human genome. This is a schematic drawing of 1 of each of the 23 human chromosomes, showing the pattern of staining seen with the Giemsa banding method. Chromosomes are first treated with trypsin and then stained with Giemsa. The patterns of light and dark bands are characteristic for each chromosome; and translocations, deletions, and other structural abnormalities can be identified. Typically 400 bands can be seen per haploid genome, and each band represents on average 7.5×10^6 bp, or twice as many base pairs as in the entire *E. coli* genome! Chromosome 1 constitutes 8.4 percent, and the Y chromosome about 2.0 percent, of the human genome. Taking the *E. coli* genome as a unit of genome size, a cytogenetic band is 2 genome units, and the Y chromosome is 15 genome units.

Ave band size = 7.5 Mb or 7.5×10^6 bp
larger than size of E. coli Genome!

HUMAN CHROMOSOMES CAN ALSO BE DISTINGUISHED BY THEIR SEQUENCES

How are these chromosomes "painted"?



(A) (B)

Figure 9.1 Human chromosome painting, in which each pair of chromosomes is labeled by hybridization with a different fluorescent probe. (A) Metaphase spread showing the chromosomes in a random arrangement as they were squashed onto the slide. (B) A karyotype, in which the chomosomes have been grouped in pairs and arranged in conventional order. Chromosomes 1–20 are arranged in order of decreasing size, but for historical reasons, chromosome 21 precedes chromosome 22, even though chromosome 21 is smaller. [Courtesy of Johannes Wienberg and Thomas Ried.]

Table 7.2: DNA content of human chromosomes[a]

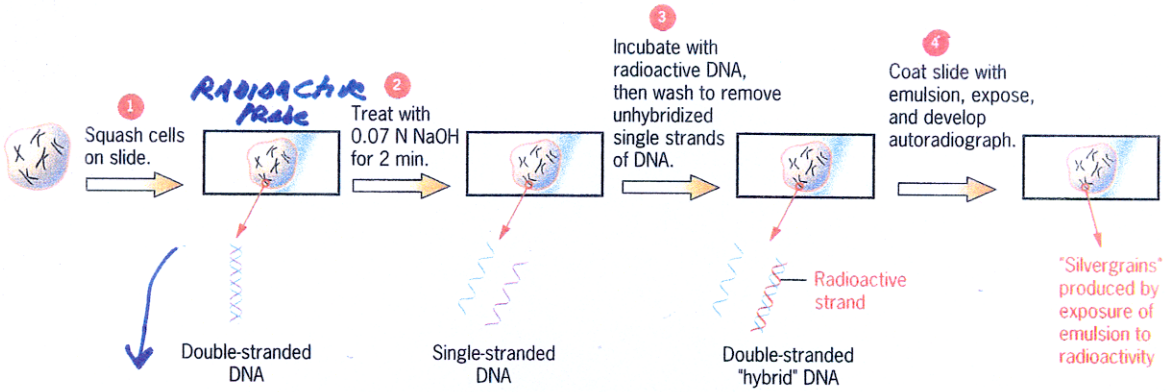
Chromosome	Amount of DNA (Mb)	Chromosome	Amount of DNA (Mb)
1	263	13	114
2	255	14	109
3	214	15	106
4	203	16	98
5	194	17	92
6	183	18	85
7	171	19	67
8	155	20	72
9	145	21	50
10	144	22	56
11	144	X	164
12	143	Y	59

[a] The DNA content is given for chromosomes prior to entering the S (DNA replication) phase of cell division (see *Figure 2.2*). Data abstracted from electronic reference 1.

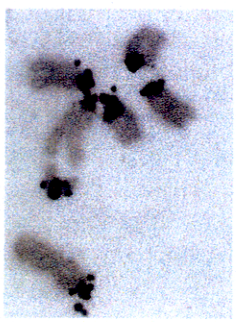
AND AMOUNT OF DNA!

FROM SEQUENCE DATA!

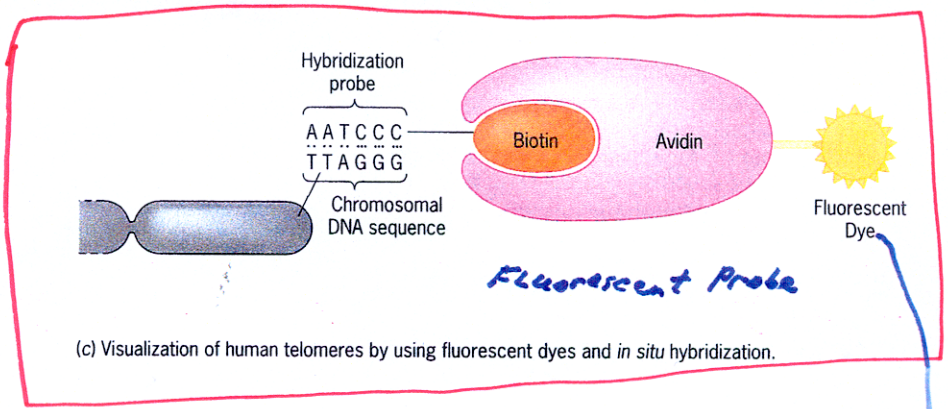
IN SITU HYBRIDIZATION WITH FLUORESCENT PROBES CAN IDENTIFY GENES & CHROMOSOMES



(a) Steps in performing *in situ* hybridization.



(b) Autoradiograph showing chromosomal locations of mouse satellite DNA sequences.



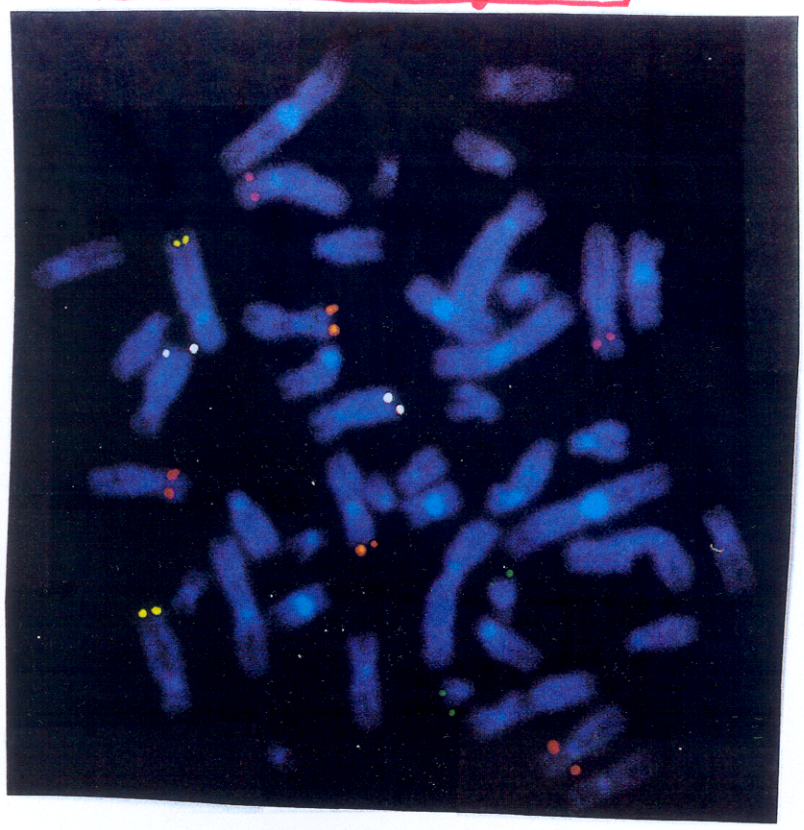
(c) Visualization of human telomeres by using fluorescent dyes and *in situ* hybridization.

Figure 11.15 Localization of repeated DNA sequences in chromosomes by *in situ* hybridization performed with radioactive probes (*a* and *b*) or fluorescent probes (*c* and *d*). The *in situ* hybridization procedure developed by Pardue and Gall is shown in (*a*), and one of their autoradiographs demonstrating the presence of the mouse satellite DNA sequence in centromeric heterochromatin is shown in (*b*). Use of fluorescent dyes to localize the TTAGGG repeat sequence to the telomeres of human chromosomes is illustrated in (*c*), and a photomicrograph demonstrating its telomeric location is shown in (*d*).

VISIBLE COLOR IN MICROSCOPE @ SPECIFIC WAVELENGTH

In situ hybridization

Mapping genes to chromosomes and specific regions



How correlate gene to chromosome position & band?

Pre-sequence approach

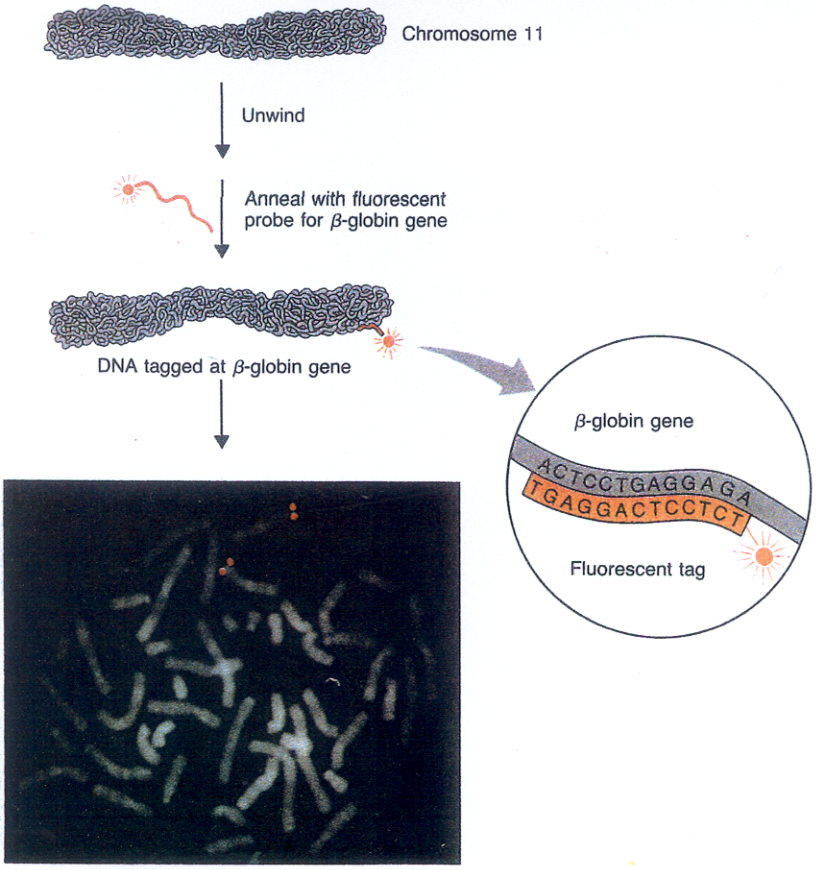
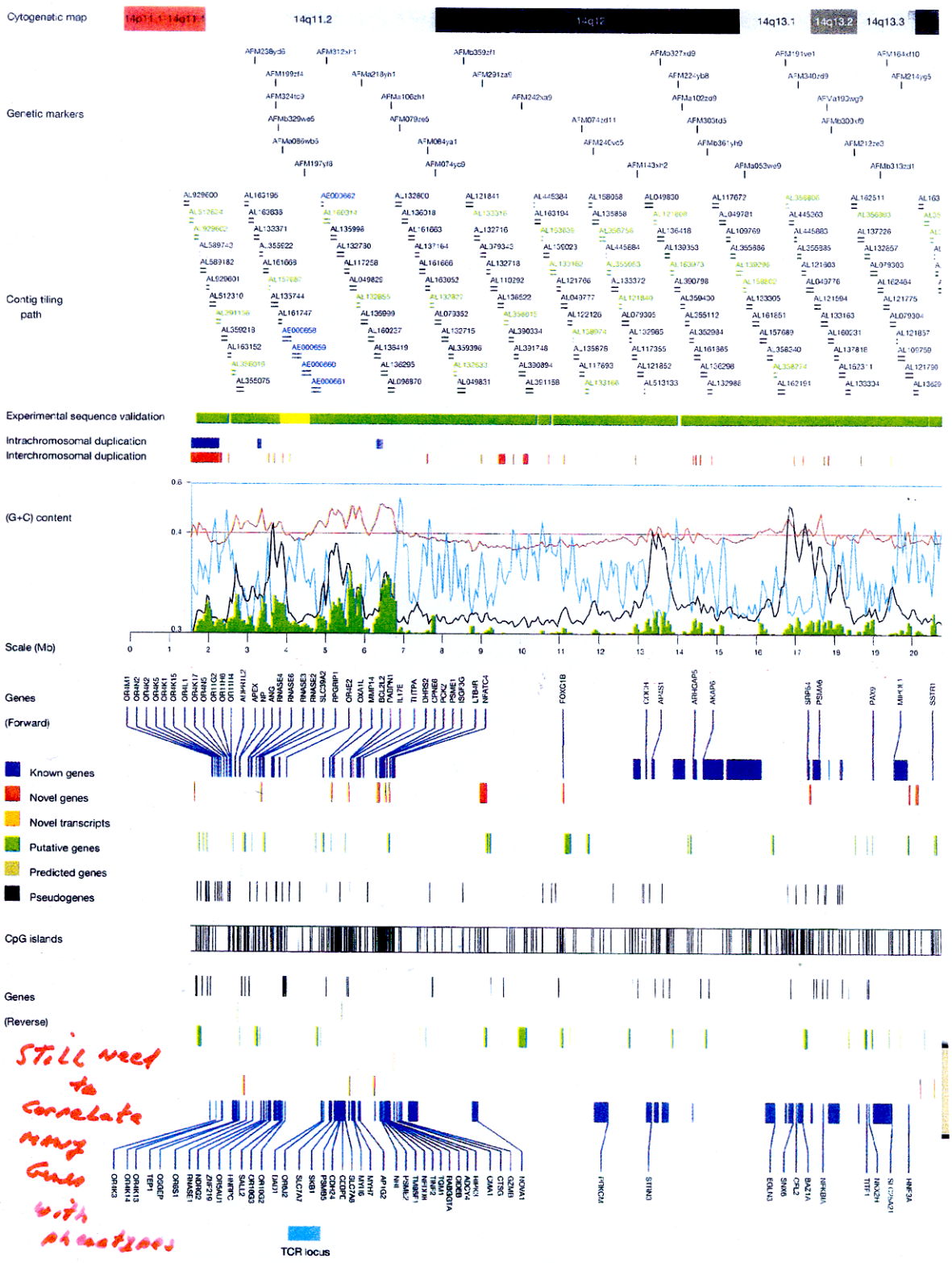


Figure 7.5 Locating the position of the β-globin gene on human chromosome 11.

This TASK IS NOW COMPLETE WITH THE COMPLETION OF THE HUMAN GENOME SEQUENCE

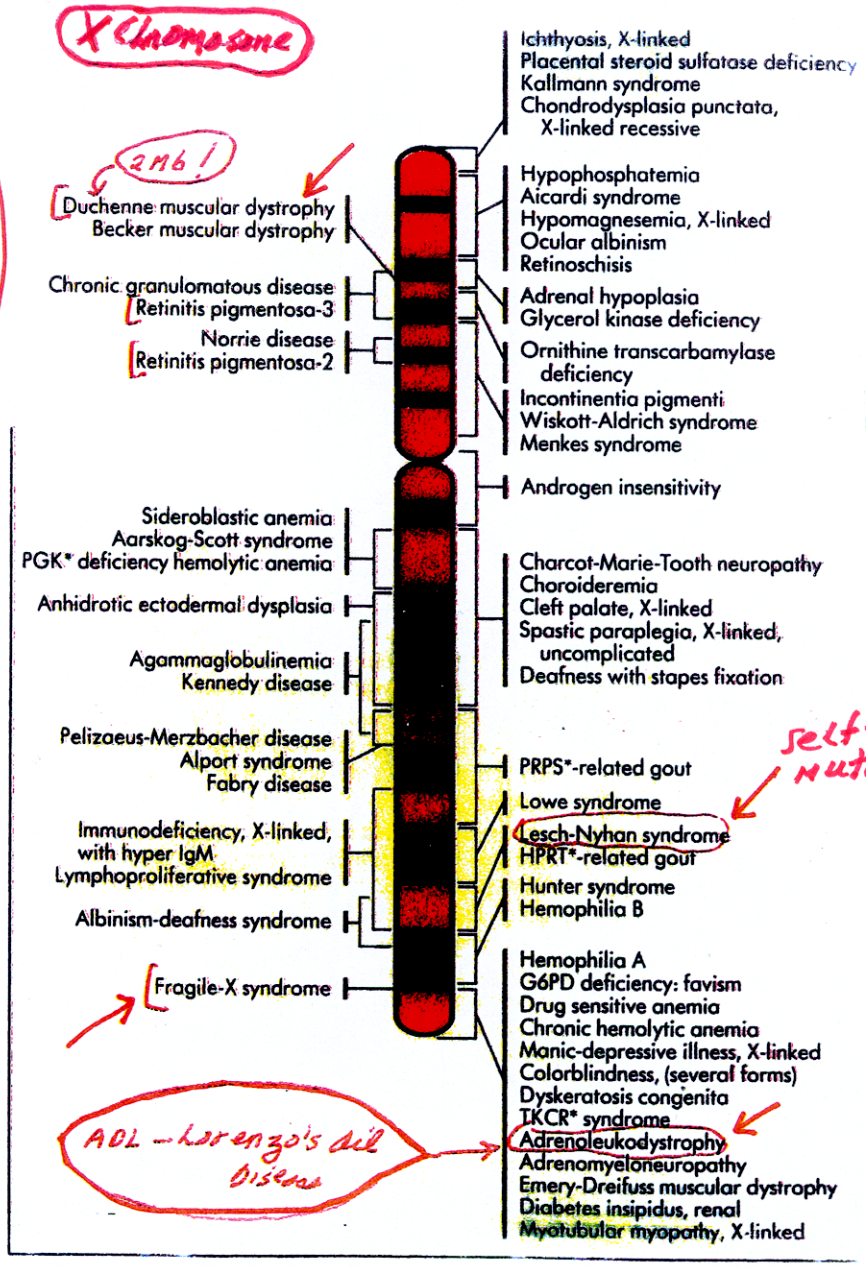


STILL need to correlate many Genes with phenotypes

GENES CAN BE MAPPED TO SPECIFIC BANDS OF EACH CHROMOSOME

X Chromosome

How locate these Genes if no Probe or Sequence?



self-mutilation

ADL – Lorenzo's oil Disease

FIGURE 12-22
The human X-chromosome gene map. Over 59 diseases have now been traced to specific segments of the X-chromosome. Many of these disorders are also influenced by genes on other chromosomes. *KEY: *PGK*, phosphoglycerate kinase; *PRPS*, phosphoribosyl pyrophosphate synthetase; *HPRT*, hypoxanthine phosphorbibosyl transferase; *TKCR*, torticollis, keloids, cryptorchidism, and renal dysplasia

DISEASE GENES CAN BE LOCALIZED TO SPECIFIC CHROMOSOMES

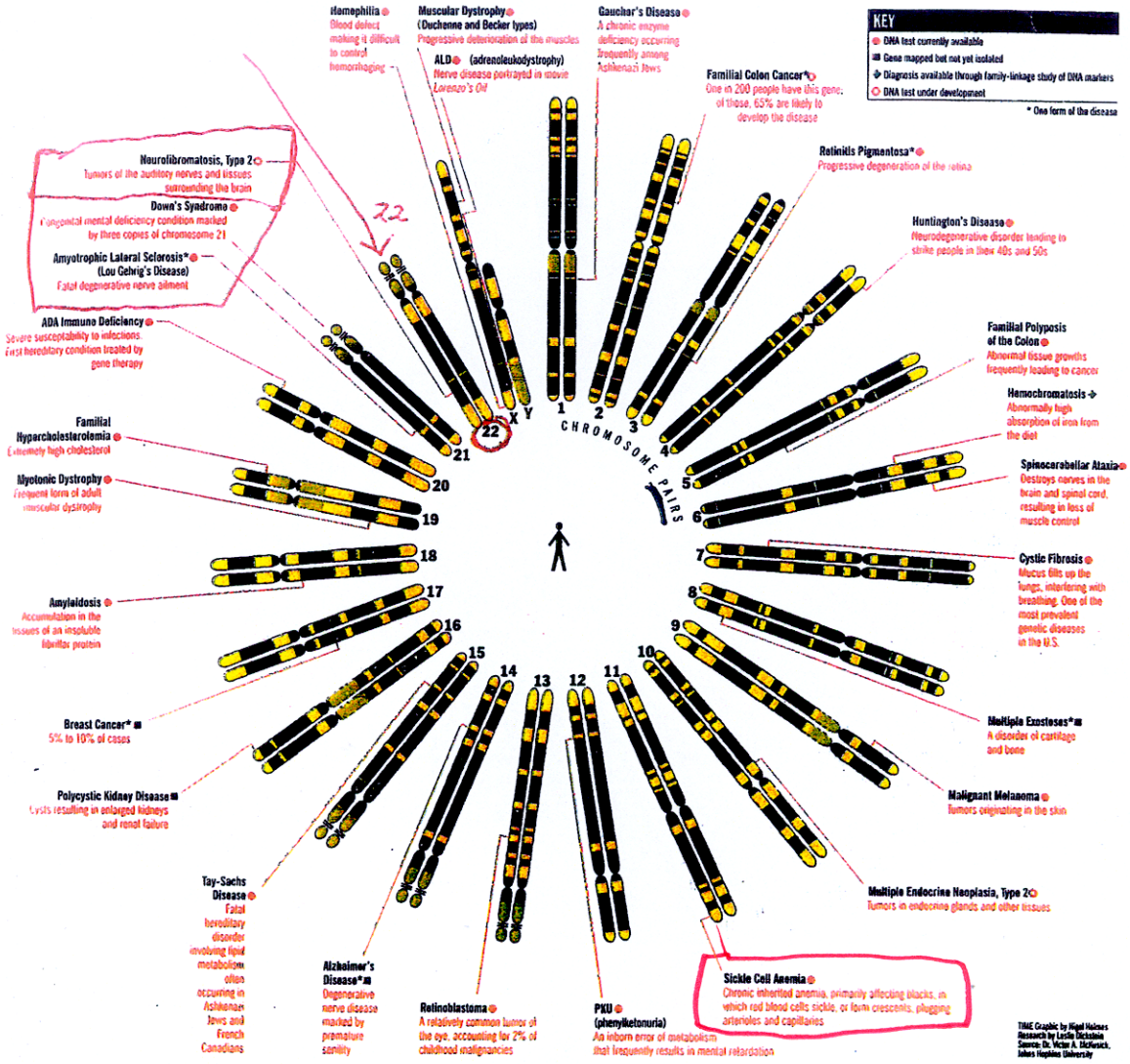


Figure 1-6 The 23 chromosomes of a human being, showing the positions of genes whose abnormal forms cause some of the better-known hereditary diseases. (*Time*)

BUT MANY MORE NEED to be correlated with disease! why difficult to do?

ORIGINS OF LETHAL POLYPLOID Zygotes / Embryos

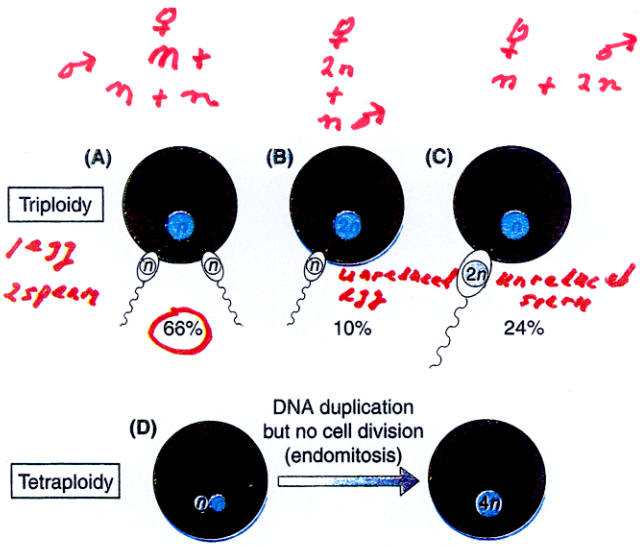


Figure 2.19: Origins of triploidy and tetraploidy.

About two-thirds of human triploids arise by fertilization of a single egg by two sperm (**A**). Other causes are a diploid egg (**B**) or sperm (**C**). Most human triploids abort spontaneously; very rarely they survive to term, but not beyond. Tetraploidy (**D**) results from failure of the first mitotic division after fertilization, and is incompatible with development.

What causes lethality with xtra genes/chromosomes?

What are the consequences of xtra Chromosomes & Chromosome Sets?

HOW CAN CHANGES OCCUR IN THE HUMAN GENOME?

LARGE GROSS CHANGES

TABLE 12.1 Chromosomal Rearrangements and Changes in Chromosome Number (or Ploidy).

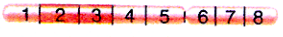
How detect?

Chromosomal Rearrangements

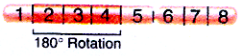
Before / After

Deletion: Removal of a segment of DNA

Duplication: Increase in the number of copies of a chromosomal region

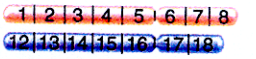


Inversion: Half-circle rotation of a chromosomal region

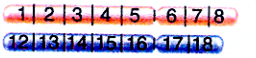


Translocations:

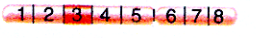
Nonreciprocal: Unequal exchanges between nonhomologous chromosomes



Reciprocal: Parts of two nonhomologous chromosomes trade places



Transposition: Movement of short DNA segments from one position in the genome to another



Chromosomal rearrangements → large changes in DNA

How detect?

How & when would these occur?

Changes in Chromosome Number or Ploidy

Euploidy: Cells that contain only complete sets of chromosomes

Diploidy (2*x*): Two copies of each homolog

← Somatic cells

Monoploidy (*x*): One copy of each homolog

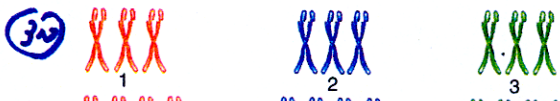
← Gametes

Polyploidy

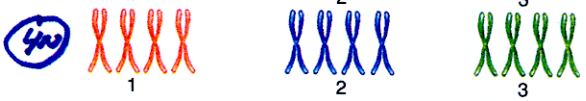
> 2n # of chromosome sets

Polyploidy: More than the normal diploid number of chromosome sets

Triploidy (3*x*): Three copies of each homolog



Tetraploidy (4*x*): Four copies of each homolog



Aneuploidy

Change in one or more chromosome #

Aneuploidy: Loss or gain of one or more chromosomes producing a chromosome number that is not an exact multiple of the haploid number

Monosomy (2*n* − 1)

Monosomic

Trisomy (2*n* + 1)

Trisomic

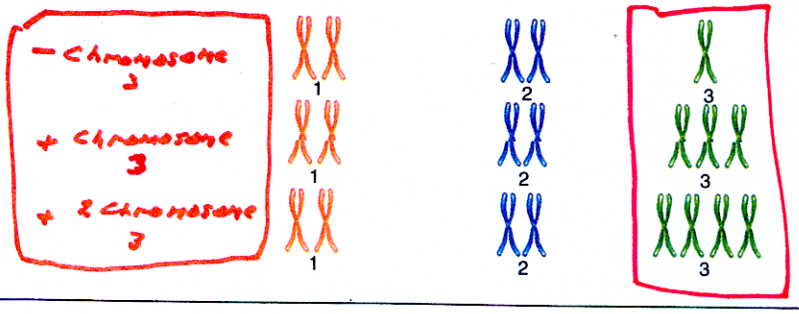
Tetrasomy (2*n* + 2)

Tetrasomic

− chromosome 3

+ chromosome 3

+ 2 chromosome 3



Note that it is more accurate to denote monoploids, triploids, and tetraploids as multiples of *x*, which represents the number of different chromosomes in a complete set, rather than as multiples of *n*, the number of chromosomes in the gametes. In this table, as throughout the chapter, nonhomologous chromosomes are drawn in different colors. Different shades of the same color highlight different regions of the same chromosome.

↳ Lead to phenotypic changes / embryo death

How are these changes detected?

HUMAN GENETICS SIDELIGHT

Amniocentesis and Chorionic Biopsy: Procedures to Detect Aneuploidy in Human Fetuses

The Andersons, a couple living in Minneapolis, were expecting their first baby. Neither Donald nor Laura Anderson knew of any genetic abnormalities in their families, but because of Laura's age—38—they decided to have the fetus checked for aneuploidy.

Laura's physician performed a procedure called **amniocentesis.** A small amount of fluid was removed from the cavity surrounding the developing fetus by inserting a needle into Laura's abdomen (Figure 1). This cavity, called the amnionic sac, is enclosed by a membrane. To prevent discomfort during the procedure, Laura was given a local anesthetic. The needle was guided into position by following an ultrasound scan, and some of the amnionic fluid was drawn out. Because this fluid contains nucleated cells sloughed off from the fetus, it is possible to determine the fetus's karyotype (Figure 2). Usually the fetal cells are purified from the amnionic fluid by centrifugation, and then the cells are cultured for several days to a few weeks. Cytological analysis of these cells will reveal if the fetus is aneuploid. Additional tests may be performed on the fluid recovered from the amnionic sac to detect other sorts of abnormalities, including neural tube defects and some kinds of mutations. The results of all these tests may take up to three weeks. In Laura's case, no abnormalities of any sort were detected, and 20 weeks after the amniocentesis, she gave birth to a healthy baby girl.

Chorionic biopsy provides another way of detecting chromosomal abnormalities in the fetus. The chorion is a fetal membrane that interdigitates with the uterine wall, eventually forming the placenta. The minute chorionic projections into the uterine tissue are called *villi* (singular, villus). At 10–11 weeks of gestation, before the placenta has developed, a sample of chorionic villi can be obtained by passing a hollow plastic tube into the uterus through the cervix. This tube can be guided by an ultrasound scan, and when it is in place, a tiny bit of material can be drawn up into the tube by aspiration. The recovered material usually consists of a mixture of maternal and fetal tissue. After these tissues are separated by dissection, the fetal cells can be analyzed for chromosome abnormalities.

Chorionic biopsy can be performed earlier than amniocentesis (10–11 weeks gestation versus 14–16 weeks), but it is not as reliable. In addition, it seems to be associated with a slightly greater chance of miscarriage than amniocentesis, perhaps 2 to 3 percent. For these reasons, it tends to be used only in pregnancies where there is a strong reason to expect a genetic abnormality. In routine pregnancies, such as Laura Anderson's, amniocentesis is the preferred procedure.

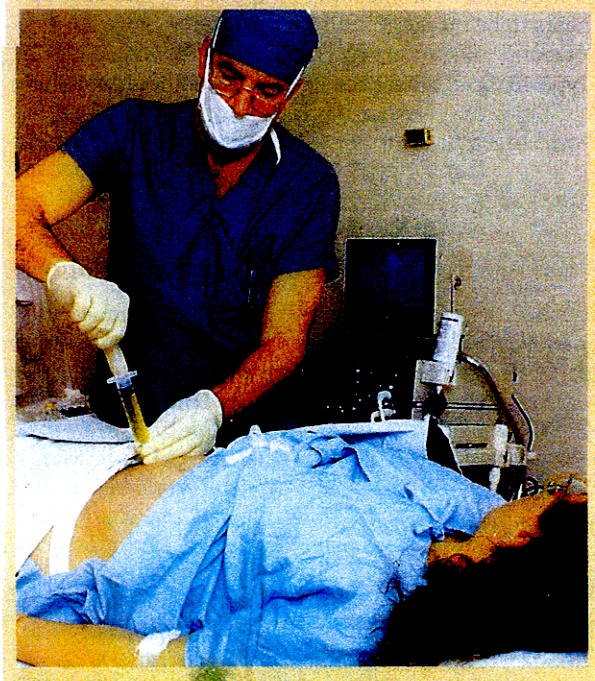


Figure 1 A physician taking a sample of fluid from the amniotic sac of a pregnant woman for prenatal diagnosis of a chromosomal or biochemical abnormality.

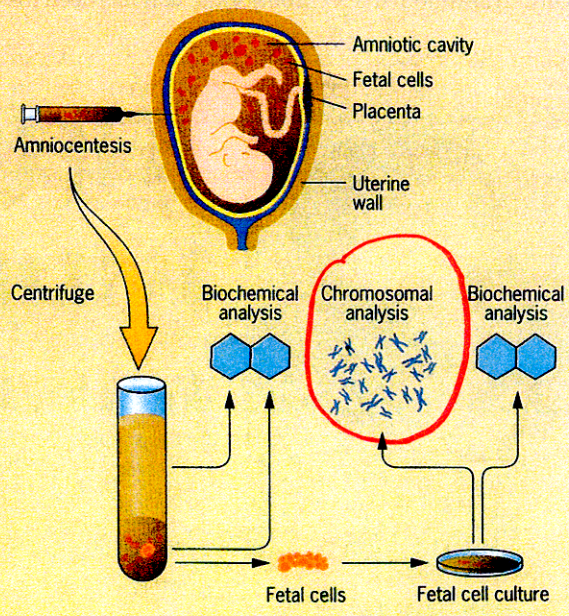


Figure 2 Amniocentesis and procedures for prenatal diagnosis of chromosomal and biochemical abnormalities.

PRENATAL Detection of CHROMOSOMAL ABNORMALITIES

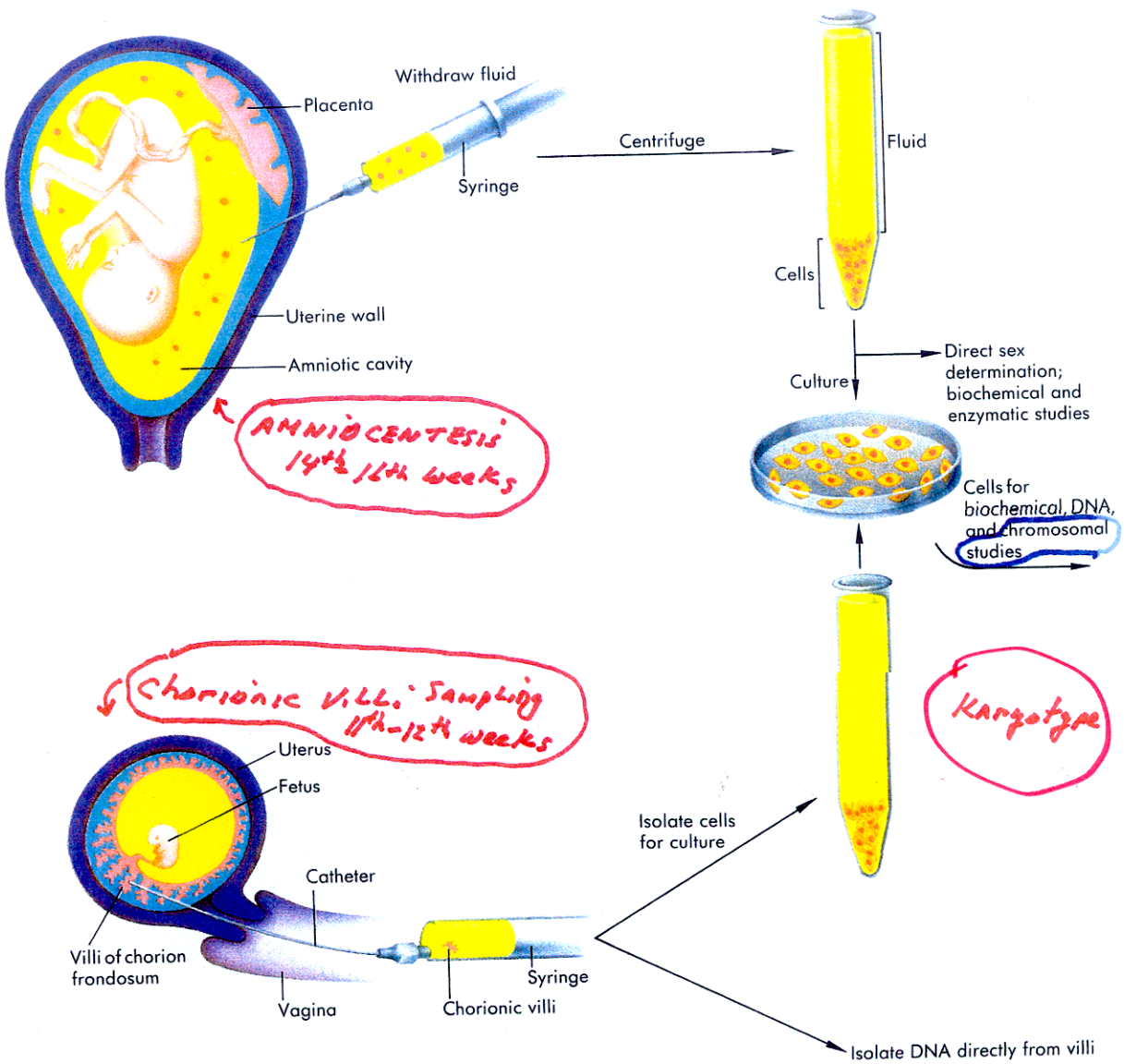


FIGURE 27-1
Amniocentesis and chorionic villus sampling. (a) A sample of amniotic fluid (mostly fetal urine and other secretions) is taken by inserting a needle into the amniotic cavity during or around the sixteenth week of gestation. The fetal cells are separated from the fluid by centrifugation. The cells can be used immediately, or more usually they are cultured so that a number of biochemical, enzymatic, and chromosomal analyses can be made. The cultured cells can also be a source of DNA. (b) Chorionic villus sampling is performed between the eighth and twelfth weeks of gestation. A catheter is introduced through the vagina or transabdominally, and a small sample of chorionic villi is drawn into the syringe. DNA can be isolated directly from the tissue, or cell cultures can be established. Note that the various elements of this figure are not drawn to scale.

ALSO CAN BE USED FOR DNA Testing

RFLP or SNP analysis