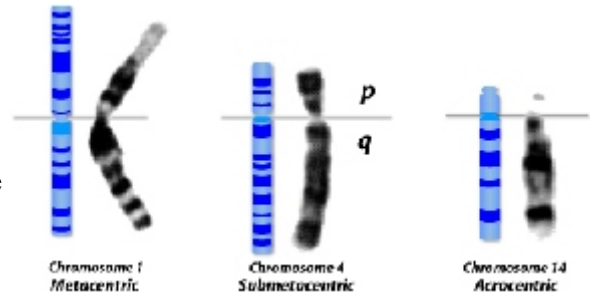


# CHROMOSOME MUTATION: CHANGES IN STRUCTURE & NUMBER

2/3/92, 2/1/95, 1/29/96, 1/31/97, 1/30/98, 28 Jan 00, 29 Jan 01, 3 Feb 03, 3 Feb 06, 1 Feb 08, 4 Feb 09, 29Jan10  
gmslg (7th) p. 523-576, 9<sup>th</sup>: 555-588

Cytogenetics: centromere position: telocentric at end  
acrocentric near end  
metacentric near middle  
acentric without centromere



**heterochromatin:** densely stained regions of chromosomes, vs **euchromatin**, poorly stained

In *Diptera*, certain tissues replicate chromosomes many times, hold homologs together = **polytene**  
Giant chromosomes formed by **endomitosis**, cells never divide

**chromocenter** (coalescence of heterochromatic area around centromeres of all four)

Banding patterns (each band = more than single gene): puffed Balbiani rings: regions of great activity (576)  
Ends of chromosomes very reactive, attach to ends of other broken ends (not telomeres)

**CHANGES IN NUMBER:** p. 557:

**euploid:** multiples of ploidy: haploid, polyploid, monoploid, diploid, triploid etc

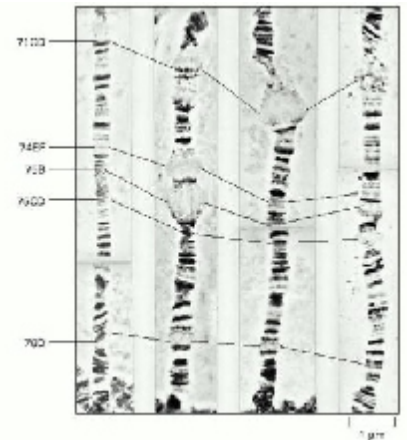
559: **colchicine** (isolated fr autumn crocus) inhibits formation of mitotic spindle

558: Form **polyploid** plants, also double haploid to make diploid

**monoploid** plants derived fr anther tissue, used to generate new genotypes, 2x to become fertile

**autopolyploid:** multiple sets of chromosomes from same species

**allopolyploid:** different chromosomes from related plants, often by cell fusion (p 562 for *Brassicac*)



**Aneuploid:** extra of one or more chromosomes [SEE TABLE, P 570 in 7<sup>th</sup> ed]

**nondisjunction** can produce trisomy at first or second meiotic division (p 566)

Trisomic:  $2n + 1$ :

Trisomy 21: Down's syndrome (see curve on p 569) 1/3rd die before 10, severely mentally retarded 1/700 all births, 1/2000 early years, 1/15 between 45-50, 1/7 over 49.

Sex Chromosome:	freq:	signs:
Turner's XO p 567	1/3000	pathognomic: lymphedema of dorsal hands&feet, neck, sterile, normal IQ
triple X XXX	1/1000	little effect some times, may be sterile, may be retarded ('superfemale')
Klinefelter's XXY p 568	1/700	tall, eunuchoid, sterile, a few are retarded
super male XYY	1/1000	usually fertile, tall, severe acne, language dysfunction (debated: 1/15 [1:50?] in prisons for violent crimes)

**CHANGES IN STRUCTURE:** (p 556, 573)

Variety of changes can be seen in *meiotic or polytene* chromosomes:

**deletions:** show loop in meiotic chromosome (*cri du chat* in humans, del tip of chromo 5) (p 575)

**duplication** can be tandem, or reverse also forms loop, but with same banding pattern as adjacent (p 530)

**inversion**, depending on where centromere is, forms several kinds of loops:

**paracentric** (centromere not in loop: can tie centromeres together in meiosis, breakage, and lost acentric fragments, 4 dif products (p 581)

**pericentric:** crossovers produce four different products, each with centromere (p 583)

**translocation:** form big X in synapsis from two chromosomes (p 584)