

Chapter 8

Variation in Chromosome Number and Arrangement

Euploid -- complete haploid sets of chromosomes are present.

Aneuploid -- organism gains or loses one or more chromosomes but not a complete haploid set.

Polyploid -- 3 or more complete haploid sets of chromosomes (triploid = $3n$, tetraploid = $4n$, pentaploid = $5n$, etc.)

In general, variation in sex-chromosome number has less dramatic phenotypic effect than does variation in autosomal number.

Variation in chromosome number results from nondisjunction (Chapter 2).

***Monosomy* -- loss of a single chromosome ($2n - 1$)**

With the exception of Turner syndrome (45, X), these are NOT tolerated in animals.

Human monosomies are conceived but none survive embryonic and fetal development.

Monosomy exposes recessive lethal alleles.

There also is a delicate balance in expression of genetic material early in development.

CRI-DU-CHAT SYNDROME --

--partial monosomy in humans.

--only a part of a chromosome is lost -- referred to as a *Segmental Deletion*.

--loss of 1/2 of the short arm of chromosome 5

--chromosomal designation = 46, -5p

- anatomical malformations including cardiac and gastrointestinal.**
- often mentally retarded.**
- abnormal development of glottis and larynx results in a characteristic sound of a meowing cat.**
- frequency = 1 in 50,000 live births.**
- size of deletion influences the physical, psychomotor, and mental skills.**
- many individuals achieve a level of social development in a trainable range.**
- those who receive home care and early special schooling are ambulatory, develop self-care skills, and learn to communicate verbally.**

***Trisomy* -- addition of a chromosome to a diploid complement.**

In general, the effects of trisomy ($2n + 1$) parallel monosomy, but the addition of an extra chromosome produces somewhat more variable individuals.

DOWN SYNDROME

- trisomy 21 = 47, 21+**
- usually due to nondisjunction during ovum formation.**

| Mother's Age | Frequency |
|---------------------|---------------------|
| 20 | < 1/1000 |
| 30 | ca 1/1000 |
| 40 | 1/100 |
| 45 | 1/50 -- 1/12 |

- prominent epicanthic fold in corner of eye; characteristically short; flat, round heads, protruding, furrowed tongues.**
- characteristic palm and fingerprints.**
- physical, psychomotor, and mental development is retarded.**
- shortened life expectancy, although many live until their 50s.**
- prone to respiratory disease, heart malformations, leukemia, and Alzheimer's disease.**

PATAU SYNDROME

- trisomy 13 = 47, 13+**
- not mentally alert, harelip, cleft palate, and polydactyly.**
- congenital malformations of most organ systems -- characteristic of abnormal development as early as 5 - 6 weeks of gestation.**
- average survival is about 3 months.**
- frequency = 1/19,000 live births.**
- average maternal and paternal age when a Patau child is born = 32.**

EDWARD SYNDROME

- trisomy 18 = 47, 18+**
- smaller than average newborn with skulls elongated in anterior-posterior direction.**
- webbed neck, congenital dislocation of hips, receding chin.**
- average survival time is 4 months with death usually caused by pneumonia or heart failure.**
- frequency = 1/8000 live births and more common in female offspring.**

Polyploidy -- more than 2 complete complements of haploid genome.

- Uncommon in mammals and birds**
- seen in lizards, amphibians, fish**
- very common in plants**

Autopolyploid -- each additional set of chromosomes is identical to parent species.

AAA = autotriploid

AAAA = autotetraploid

Produce autopolyploids by:

- 1. heat/cold shock during meiosis**
- 2. apply colchicine to somatic cells undergoing mitosis.**

- colchicine interferes with spindle formation.**
- replicated chromosomes do not enter anaphase**
- remove colchicine -- cells return to interphase with double the number of chromosomes.**

***Autotriploids* -- AAA -- several potatoes, Winesap apples, commercial bananas, seedless watermelons.**

***Autotetraploids* -- AAAA -- alfalfa, coffee, peanuts, McIntosh apples.**

***Autooctoploids* -- AAAAAAAAAA -- commercial strawberries.**

***Allopolyploids* -- polyploids that result from hybridization of two closely related species.**

hybrids may be sterile due to the failure of some or all chromosomes to synapse.

Figure 8.11 -- development of autopolyploid.

Upland Cotton -- Gossypium

- 26 pairs of chromosomes
- allotetraploid -- AADD
- hybrid between American and Asian cotton (A genome - 13 pairs of chromosomes) and Old World cotton (D genome - 13 pairs of chromosomes).

Radish ($n = 9$) X Cabbage ($n = 9$)

Hybrid (9R + 9B)
Most Sterile



18R + 18B



roots --> Cabbage
shoots --> Radish

Endopolyploidy -- cells in normal diploid organism becomes polyploid through the process of *Endomitosis*.

--vertebrate liver -- $2n$, $4n$, $8n$, or $16n$

--waterstrider $2n = 22$

-- considerable variation in chromosome number in different tissues.

-- salivary glands -- 1,024 - 2,048 copies of each chromosome.

-- nuclei may contain over 40,000 chromosomes.

WHY?

Variation in Chromosomal Structure and Arrangement