

Chapter 15: Chromosomal Abnormalities

1. Define:
 - nondisjunction
 - polyploidy
 - aneuploidy
 - trisomy
 - monosomy
2. Describe each of the aneuploidies that can be found in an appreciable number of human adults (chromosomal abnormality, common name of the syndrome if it has one, phenotypes)
3. Draw an inversion, a deletion, a duplication, and a reciprocal translocation.
4. Describe trinucleotide repeat disorders.

1. Abnormalities in chromosomal number

A. How does it happen? nondisjunction

1. **nondisjunction** - mistake in cell division where chromosomes do not separate properly in anaphase
 - usually in meiosis, although in mitosis occasionally; in meiosis, can occur in anaphase I or II
2. **polyploidy** - complete extra sets (3n, etc.) - fatal in humans, most animals
3. **aneuploidy** - missing one copy or have an extra copy of a single chromosome
 - three copies of a chromosome in your somatic cells: **trisomy**
 - one copy of a chromosome in your somatic cells: **monosomy**
 - most trisomies and monosomies are lethal well before birth in humans; exceptions covered below
 - generally, autosomal aneuploids tend to be spontaneously aborted
 - over 1/3 of human pregnancies are lost spontaneously after implantation (probably closer to 1/3)
 - chromosomal abnormalities are the leading known cause of pregnancy loss
 - data indicate that minimum 10-15% of conceptions have a chromosomal abnormality
 - at least 95% of these conceptions spontaneously abort (often without being noticed)

B. aneuploidy in human sex chromosomes

1. **X₀ female (Turner syndrome)**
 - short stature; sterile (immature sex organs); often reduced mental abilities; about 1 in 2500 human female births
2. **XNY male (Klinefelter syndrome)**
 - often not detected until puberty, when female body characteristics develop; sterile; sometimes reduced mental abilities; testosterone shots can be used as a partial treatment; about 1 in 500 human male births
3. **XYY male (XYY syndrome)**
 - usually tall, with heavy acne; some correlation with mild mental retardation and with aggressiveness; usually still fertile; about 1 in 1000 human male births
4. **XXX female (triple X syndrome)**
 - usually just like XX females, except for having 2 Barr bodies in somatic cells; HOWEVER, more likely to be sterile, and if fertile, more likely to have XXY and XXX children; about 1 in 1000 human female births

C. aneuploidy in human autosomes

1. **autosomic monosomy** appears to be invariably fatal, usually very early in pregnancy
2. **most autosomic trisomy** is fatal, but sometimes individuals trisomic for autosomes 13, 15, 18, 21, or 22 survive to birth and even beyond
 - chromosome number reflects size; bigger number = smaller size, and usually fewer genes
 - extra 13, 15, or 18 leads to multiple defects and usually death well before 1 year of age
 - extra 22 is much like extra 21 (Down syndrome, covered below), but usually more severe, with shorter life expectancy

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David G. Myers, Richard O. Straub



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