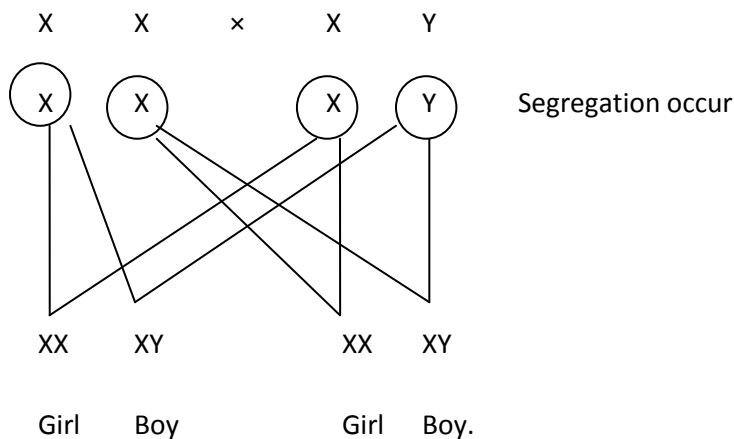


BIO 301: GENETICS 2

SEX DETERMINATION IN HUMANS

It is the tiny Y chromosome that determines maleness in human being. When the Y is absent, the individual is female. In spite of the fact that they are physically dissimilar, the X and Y chromosomes segregate as a homologous pair during gametogenesis. Following spermatogenesis, half the mature sperm therefore contain the Y chromosome and the half carries the X chromosome. Since the mature ovum or X cell can have only the X chromosome. The sperm which fertilizes an egg will determine the sex of the child conceived.

The Y bearing sperm determines that the child will be a boy and the X bearing sperm determines that the child will be a girl. This means that the father is the parent whose germ cells determine the sex of the children.



X- LINKED TRAITS

In women, there are genes identified as been on the X chromosome which is referred to as X- linked and which are referred to in the past as sex linked trait. Since there is a true pair of homologous sex chromosomes in human females, the Mendel law of dominant and recessiveness apply to trait determination by X-linked genes in woman in the same manner as to traits determined by genes on the autosomes. For this reason, X-linked traits are more frequently expressed in man than in woman.

When a man carries a recessive mutation on his X-chromosome such as genes for colour blindness, he will express the traits i.e. he will be colour blinded. A woman can carry the same recessive gene on her X-chromosome but because she is likely to have a normal dominant allele on her other X-chromosome,

she will be phenotypically normal but she will be a carrier (note that except for its capacity to determine maleness in higher animals, the Y-chromosome is genetically inactive).

Another well known X-linked trait in woman is hemophilia the so called bleeders' disease in which the clotting mechanism in affected person's blood is defective. Although hemophilia and colour blindness are determined by gene on the X-chromosome, the trait obviously are related to sex determination.

SEX INFLUENCED AND SEX LIMITED TRAITS

There are some traits found predominantly in man and not in woman such as baldness can resemble Y-linked in their transmission merger. This trait however are actually determined by autosomal gene but their expression is influenced by sex hormone. The gene for baldness for example behave as an autosomal dominant in males but an autosomal recessive in females such traits are said to be sex influenced. There are also traits which are expressed only in one or the other sex. For example in Deer, antlers are part of the male genotype and even though the production of milk is controlled by hormones. Only in female can be induced to lactate, such traits are commonly referred to as sex limited traits.

Although these genes are expressed in only one sex, they are transmitted on both sexes. For example, some bulls are selected only for high milk yield to their female offspring.

MUTAGENESIS

Chromosomal mutation: mutation can involve either the entire chromosomes or large portion of chromosomes and are known as **gross or chromosomal mutations**. Or, they may change the sequence of nucleotide within the individual genes and are then known as **point mutation**.

ABBERATIONS OR CHROMOSOMAL MUTATION.

Numerical aberration of chromosomes may involve the entire set a condition called euploidy or individual chromosome called aneuploidy. Aberrations involving individual chromosomes are indicated by the suffix 'somy'. Thus, the presence of an extra chromosome on 21 is called Trisomy 21. An individual possessing only one rather than a pair of 21-chromosomes will be monosomic.

1. **Down's syndrome (Mongoloid idiosy)**: one of the most familiar conditions in humans resulting an euploidy is trisomy 21 originally studied by Langdon Down in 1856. It has been termed mongoloid idiosy or Mongolia because individuals with trisomy21 were considered to have certain facial characteristics that resembled the oriental features(Asians).

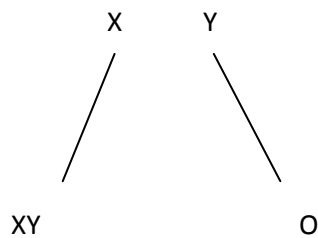
The presence of an extra copy of one of the smallest chromosomes in trisomy 21 result in a wide variety of abnormalities in every area of the body differing in concentration and degree of severity from parent to parent.

NON DISJUNCTION

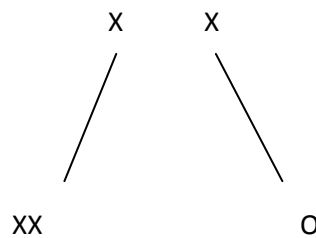
This can occur before the first or second meiotic division. The basic event in non disjunction is that the chromosomes do not separate or they do not disjoint. This result in an euploidy in the daughter cells. Gametes containing the extra chromosomes will produce an individual with Down's syndrome.

SEX CHROMOSOME ABBERATIONS

1. **Turner's syndrome;** the monosomic condition depicted as XO is called Turner's syndrome such individuals are usually phenotypically female but are sterile due to abnormal gonads. Thus, another term for Turner's syndrome is gonadal dsygenesis. The patient tends to be short and secondary sexual characteristics in adult such as breast development are slight.



Non disjunction in male



Non disjunction in female

In both cases if the O or nullisomic gamete were involved in the fertilization with an X pairing gamete from the other parent. It results in gamete XO.

2. **Klinefelter's syndrome:** if a sperm from non disjunction were to fertilize a normal X bearing ovum and if an XX ovum rising from non disjunction during oogenesis were to be fertilize a normal Y bearing sperm.

This imbalance in the number of sex chromosome leads to series of abnormalities collectively called Kline felter's syndrome. Such individuals have 47 chromosomes including a Y and two X chromosomes; they are generally developed gonads and tend towards breast development.

3. **XXX Super females:** fertilization of XX germ cell by XY sperm cells results in XXX individual. there is no characteristic phenotype associated with this aneuploidy condition. As the level of intelligence and development of the reproductive structures vary from normal to sub normal.

STRUCTURAL ABNORMALITIES OF CHROMOSOMES

1. **Translocation:**
2. **Reciprocal and non reciprocal translocation:**
3. **Robertsonian translocation:**
4. **Duplication; .**
- 5 **Deletion;**
- 6 **Inversion;**

PLEIOTROPY

The phenomenon of a single gene determining a number of distinct and seemingly unrelated characteristics is known as pleiotropy. Since geneticists now know that each gene determines a specific protein and that each protein can have a cascade of effects on an organism, we can understand how pleiotropy arises. Among the aboriginal Maori people of New Zealand, for example, many of the men develop frequent respiratory problems and are also sterile. Researchers have found that the fault lies with the recessive allele of a single gene. The gene's normal dominant allele specifies a protein necessary for the action of cilia and flagella, both of which are hair like structures extending from the surface of some cells. In men who are homozygous for the recessive allele, however, cilia that normally clear the airways fail to work effectively and flagella that normally propel sperm fail to do their job. Thus, one gene determines a protein that indirectly affects both respiratory function and reproduction.

GENES

The gene is the unit of inheritance. Each gene is a nucleic acid sequence that carries the information representing a particular polypeptide. A gene is a portion of a DNA. It is a stable entity but can suffer a change in sequence. Such a change is called a mutation. When a mutation occurs, the new form of the gene is inherited in an unstable manner just like the previous form. The average gene called a cistron consist of about 600 consecutive base pairs. Some genes however, contain fewer consecutive base pairs. A gene may exist in alternate forms that determine the expression of a particular characteristic.