



THE X AND Y CHROMOSOMES OF A PERSON REPRESENT THE BIOLOGICAL SEX OF A PERSON

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ANNOTATION

In humans and other mammals, the biological sex is represented by sex chromosomes: XY in males and XX in females. Genes located on the X chromosome are called X-linked. The genes attached to the X chromosome have a unique heritability, since their number is different in males (XY) and females (XX). X-linked genetic disorders in humans are more common in men than women due to X-linked inheritance.

KEYWORDS :X-linked inheritance is the chromosomal basis of sex determination. X- and Y-chromosomes, X-linked inheritance

Most chromosomes in humans are found in homologous pairs. Chromosomes in this homologous pair contain the same information, meaning that their genes are in the same order. But they have different variants of these genes. Are all your chromosomes in homologous pairs? The answer to this question depends on whether you are (chromosomally) male or female. Males have two sex chromosomes, X and Y. Unlike the 44 autosomal chromosomes (non-sex chromosomes), the X and Y chromosomes do not share the same genes and are therefore not considered homologous chromosomes. Instead of an X and a Y chromosome, women have two X chromosomes. These X chromosomes form a true homologous pair. Because sex chromosomes do not always come in homologous pairs, they have a unique heredity. contains genes.

The sex chromosomes in humans are X and Y, which represent a person's biological sex, ie XX is female and XY is male. Although the Y chromosome has very little similarity to the X chromosome, they can pair during meiosis. The Y chromosome is slightly shorter and has fewer genes. Compared to numbers, the X chromosome has 800-900 protein-coding genes with a wide range of functions, while the Y chromosome has 60-70 protein-coding



genes, and half of them are active only in the testes (organs that produce sperm). The Y chromosome in humans is an important factor in determining the sex of a developing embryo. This is often due to a gene called SRY ("Sex Determining Part of the Y Chromosome"). The SRY gene is located on the Y chromosome and synthesizes proteins necessary for the development of the male sex. XX-chromosomal embryos do not have the SRY gene, so they develop into females. Since embryos with XY chromosomes have the SRY gene, the male sex will develop from them. In rare cases, due to defects in meiosis, the SRY gene can be transferred from the Y chromosome to the X chromosome. If an X chromosome with the SRY gene fertilizes a normal egg, the result is an embryo that is chromosomally female (XX) but develops as a male. If a Y chromosome lacking the SRY gene fertilizes a normal egg, the result is an embryo that is chromosomally male (XY) but develops as a female. If genes linked to the X chromosome are located on the X chromosome rather than on the Y chromosome, it is called an X-linked gene. Genes attached to the X chromosome have a unique heredity compared to genes on non-sex chromosomes (autosomes). Because these genes have different numbers of copies in men and women. Because females have two X chromosomes, they have two copies of genes linked to the X chromosome. For example, in the fruit fly *Drosophila* (*Drosophilidae*) (XX female and XY male), the gene for white eye color is located on the X chromosome, and female flies have two copies of this gene. If the genes are present in two different alleles: (dominant, normal red eyes) and (recessive, white eyes), female flies can have three different genotypes. Male flies exhibit different genotypes than females. Because they have only one X chromosome (paired with a Y chromosome), there is only one copy of a gene attached to any given X chromosome. Taking eye color, for example, male flies can have two different genotypes: Any X-linked gene a male fly receives determines its appearance because it has no other copy of the gene, even if the female has the recessive allele. Instead of being heterozygous or homozygous for genes linked to the X chromosome, male flies are called hemizygous organisms. We cross a white-eyed female fly with a red-eyed male fly to see how sexing affects heredity.

If these genes were on non-sex (autosomal) chromosomes, we could say that all offspring would have red eyes, because red eyes are dominant. Here's what actually



happens: However, since this gene is X-linked and the female fly has a recessive genotype (white eyes), all male offspring inherit the X chromosome from the female (mother) fly and have white eyes. Female offspring are red-eyed because they get one of their two X chromosomes from the male (father) fly and the recessive gene from the female (mother) fly.

Genetic diseases linked to the X chromosome. The same patterns we've seen in fruit flies can be applied to human genetics. In humans, some traits (such as color blindness, hemophilia, and muscular dystrophy) are inherited on the X chromosome. These diseases are more common in men than in women due to the above hereditary characteristics. Why is this observed? Let's look at this in a mother who is heterozygous for a particular disease. Women who are heterozygous for this disease are carriers, and usually do not show symptoms of this disease. 50%, 50%, 50% of sons born to this mother will be born with the disease, but it is rare in daughters (only if their father has the disease) and they also have a 50%, 50%, 50% chance of being carriers. What else could be causing this? Recessive, X-linked traits are usually more common in males than females. If a male child receives a "bad" allele from his mother, he cannot receive a "good" allele that hides this bad trait from his father (because the father provides the Y chromosome). Female children are usually free of these diseases, having received normal alleles from their father. Hemophilia Let's look at the Punnett cell depicting hemophilia, a genetic disease inherited by attachment to the X chromosome. Patients with hemophilia do not have normal blood clotting and it is inherited in a recessive manner. In patients with this disease, even simple bleeding from an injury can be seriously life-threatening. Hemophilia is caused by a mutation in one of two genes located on the X chromosome. Both genes are responsible for the synthesis of proteins that help blood clotting. Let's look at one functional allele and another disease-causing allele of this gene.

In our example, a healthy, heterozygous genotype woman and a healthy, hemizygous man got married. Both parents have normal blood clotting, but if the woman is a carrier, what are the chances of having a son or daughter with hemophilia? Because the woman is a carrier, half of the children (both boys and girls) will have the hemophilia allele. None of the girls are born with hemophilia (no chance of the disease). In order to be born



with the disease, they had to receive the allele from both the father and the mother. Since the probability of girls inheriting alleles from their father is 0, the probability of having hemophilia in girls is also zero. Since boys receive a Y chromosome from their father instead of an X, the genes responsible for blood clotting are passed only from the mother. Since the mother is heterozygous, on average half of the sons receive the allele and develop hemophilia.

Using different genomic technologies, the two research groups mapped the Y chromosome in two separate sets of mammals, including more than 15 different species, including humans, chimpanzees, rhesus monkeys, bulls, marmosets, mice, rats, dogs, and opossums. independently analyzed its evolution.

Surprisingly, they found a small but stable group of important regulatory genes on the Y chromosome, which were preserved over long periods of evolution, even when the surrounding genes were destroyed. Notably, these genes play an important role in controlling the expression of other genes throughout the genome and can affect tissues throughout the human body. One of the reasons these Y-chromosome regulatory genes persist for so long is that they are "dosage-dependent," meaning that they require two copies to function normally. However, regulatory genes are often dosage-dependent and haplo-deficient, meaning that two copies of the gene are required, and having only one copy can lead to an abnormality or disease. In females, these regulatory genes escape X inactivation, so a copy of the second X chromosome is also expressed; in males with only one X chromosome, the maintenance of this group of regulatory genes on the Y chromosome is essential to ensure the second copy.

In general, this means that in addition to its role in determining sex and fertility, the Y chromosome also contains important genes that are important for male health and survival.

These findings have important implications for our understanding of differences in biology, health, and disease between men and women. Because the genes on the X and Y chromosomes have had independent histories of selection, there may be subtle functional differences that are a direct result of the genetic differences on the two chromosomes. Although these differences have yet to be studied in detail, more research on conserved Y-



chromosome genes will help us better understand the underlying biology and disease susceptibility differences in men and women, and better manage health.

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