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X-Chromosome and Abnormalities

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Abstract: This research investigates the function of the X chromosome and its different abnormalities on humans, including data from comparative genome analysis of other organisms. The X chromosome has many fatures that are unique in the human genome. Females inherit an X chromosome. Gene expression on one of the female X chromosomes is silenced early in development by the process of X-chromosome inactivation (XCI), and this chromosome remains inactive in somatic tissues thereafter. In the female germ line, the inactive chromosome is reactivated and undergoes meitotic recombination with the second X chromosome. The male X chromosome fails to recombine along virtually its entire length during meiosis: instead, recombination is restricted to short regions at the tips of the X chromosome arms that recombine with equivalent segments on the Y chromosome. Genes inside these regions are shared between the sex chromosomes, and their behaviour is therefore described as 'pseudoautosomal'. Genes outside these regions of the X chromosome are strictly X-linked, and the vast majority are present in a single copy in the male genome. The unique properties of the X chromosome are a consequence of the evolution of sex chromosomes in mammals. The sex chromosomes have evolved from a pair of autosomes within the last 300 million years. In the process, the original, functional element have been conserved on the X chromosome, but the Y chromosome has lost almost all traces of the ancestral autosome, including the genes that were once shared with the X chromosome. The hemizygosity of males for almost all X chromosome genes exposes recessive phenotypes, yhus accounting for the large number of diseases that have been associated with the X chromosome.The biological consequences of the sex chromosome evolution account fort the intense interest in the human X chromosome in recent decades. However, evolutionary processes are likely to have shaped the behaviour and structure of the X chromosome in many the ways, influencing features such as repeat content, mutation rate, gene content and haplotype structure. The availability of the finished sequence of the human X chromosome, described here, now allows us to explore its evolution and unique properties at a new level.

Keywords: X chromosome, XCI, Y chromosom

Introduction

Identifying gens on each chromosome in an active area of genetic research. Buceuse researchers use different approaches to predict the number of genes on each chromosome, the estimated number of genes varies The X chromosomelikely contains 800 to 900 genes that provide instructions for making proteins. These proteins perform a variety of different roles in the body. The X chromosome in humans spans more than 153 million base pairs (the building material of DNA). It represents about 800 protein-coding genes compared to the Y chromosome containing about 70 genes, out of 20,000-25,000 total genes in the human genome. Each person usually has one pair of sex chromosomes in each cell. Females have two X chromosomes, whereas males have one X an done Y chromosome. Both males and females retain one of their mother's X chromosomes, and females retain their second X chromosome from their father. Since the father retains his X chromosome from his mother, a human female has one X chromosome from her paternal grandmother (father's side), an one X chromosome from her mother (1). This inheritance pattern follows the Fibonaccci numbers at a given ancestral depth. Genetic disorders that are due to mutations in genes on the X chromosome are decribed as **X linked.** The X chromosome carries hundreds of genes but few, if any, of these have anything to do directly with sex

determination. Early in embryonic development in females, one of the two X chromosome is randomly and permanently inactivated in nearly all somatic cells (cells other than egg and sperm cells). This phenomenon is caled X-inactivation or iyonization, and creates a Barr body. If X-inactivation in the somatic cell meant a complete de-functionalizing of one of the X- chromosomes, it would ensure that females, like males, had only one fuctional copy of the X chromosome in each somatic cell (2,3). This was preciously assumed to be the case. However, recent research suggests that the Barr body may be more biologically active than was previously supposed. Genetic disorders that arise from missing, additional of malformed copies of the X chromosome are termed numerical disorders. Examples include Klinefelter's syndrome where a male has one or more extra copies; Triple X syndrome, where a female has one extra copy and Turner syndrome where a female has one normal X chromosome an done missing or abnormal one (4).

Structure of X Chromosomes

It is theorized by Ross et al. 2005 and Ohno 1967 that the X chromosome is at least partially derived from the autosomal (non-sex-related) genome of other mammals, evidenced from interspecies genomic sequence elignments. The X chromosome is notably larger and has a more active euchromatin refion than its Y chromosome counterpart. Futher comparison of the X and Y reveal regions of homology between the two.

However, the cerresponding region in the Y appears far shorter and lacks regions that are conserved in the X throughout primate speciens, implying a genetix degeneration for Y in that region. Because males have only one X chromosome, they are more likely to have an X chromosome-related disease.

It is estimated that about 10% of the genes encoded by the X chromosome are associated with a family of ''CT'' genes, so named because they encode for markers found in both tumor cells (in cancer patients) as well as in the human testis (in healthy patients) (5).

Health conditions related to chromosomal changes

The following a few chromosomal conditions are associated with changes in the structure or number of copies of X chromosome

a) 46,XX testicular disorder of sex development

In most individuals with 46,XX testicular disorder of sex development, the condition results from an abnormal Exchange of genetix material between chromosomes (translocation). This Exchange occurs as a random event during the formation of sperm cells in the affected person's father. The translocation affects the gene responsible for development of a fetus into a male (the SRY gene). The SRY gene, which is normally found on the Y chromosome, is misplaced in this disorder, almost always onto an X chromosome. A fetus with an X chromosome that carries the SRY gene will develop as a male despite not having a Y chromosome.

b) Intestinal pseudo-obstruction

Intestinal pseudo-obstruction, a condition characterized by impairment of the muscle contractions that move food through the digestive tract (peristalsis), can be caused by genetic changes within the X chromosome. Some individuals with intestinal pseudo-obstruction have mutations, duplications, or deletions of genetic material in the X chromosome that affect the FLNA gene. Researchers believe that these genetix changes may impair the function of the filaminA protein , causing abromalities in the cytoskeleton of nevre cells (neurons) in the gastrointestinal tract. These abromalities result in impaired peristalsis, which causes abdominal pain and the other gastrointestinal symptoms on intestinal pseudo-obstruction. Deletions or duplications of genetix material that affect the FLNA gene can also include adjacent genes on the X chromosom. Changes in adjacent genes may account for some of the other signs and syptoms, sucg as neurological abromalities and unusual facial features, that ocur in some affected individuals.

c) Klinefelter syndrome

Klinefelter syndrome is caused by the presence of one or more exta copies of the Xchromosome in a male's cells. Exta genetic material from the X chromosome interferes with male sexual development, preventing the testes from functioning normally and reducing the levels of testosterone. A shortage of testosterone can lead to

delayed or incomplete puberty, genital abnormalities, breast enlargement (gynecomastia), reduced facial and body hair, and an inability to have biological children (infertility). Children with Klinefelter syndrome, and a shy and unassuming personality. Typically, people with Klinefelter syndrome have one exta copy of the X chromosome in each cell, for a total of two X chromosome and one Y chromosome (47,XXY). Less commonly, affected individuals may have two or three extra X chromosomes (48,XXXY or 49,XXXXY). As the number of extra sex chromosomes increases, so does the risk of learning problems, intellectual disability, birth defects, and other health issues. Some people witht features of Klinefelter syndrome have the extra X chromosome in only some of their cells; in these individuals, the condition is described as mosaic Klinefelter syndrome (46,XY/47,XXY). Individuals with mosaic Klinefelter syndrome may have milder signs and symptoms, depending on how many cells have an additional X chromosome.

d) Triple X syndrome

Triple X syndorme (also called 47,XXX or trisomy X) results from an exta copy of the X chromosome in each of a female's cells. Females with triple X syndrome have three X chromosomes, for a total of 47 chromosomes per cell. An exta copy of the chromosome is associated with tall stature, learning problems, and other features in some girls and women.
Some females with X syndrome have an extra X chromosome in only some of their cells. This phenomenon is called 46,XXX mosaicism.
Females with more than one extra copy of the C choromosome (48,XXXX or 49,XXXXX) have been identified, but these choromosomal changes are rare. As the number of extra sex choromosomes increases, so does the risk of learning problems, intellectual disability,birth defects,and other health issues.

e) Turner syndrome

Turner syndrome results when one normal X chromosome is present in a female's cells and the order sex choromosome is missing or structurally altered. The missing genetic material affects development before and after birth, leading to short stature, ovarian malfunction, and the other features of Turner syndrome. About half of individuals with Turner syndrome have monosomy X (45, X) , which means each cell in an individual's body has only one copy of the X chromosome instead of the usual two sex chromosomes. Turner syndrome can also ocur if one of the sex choromosomes is partially missing or rearranged rather than completely absent. Some women with Turner syndrome have a chromosomal change in only some of their cells, which is known as mosaicism.

Some cells have usual two sex choromosomes (either two X choromosomes or one X choromosome an done Y chromosome), and other cells have only one copy of the X choromosome. Women with Turner syndrome caused by X choromosome mosaicism (45,X/46, XX or 45 X/46, XY) are said to have mosaic Turner syndrome. Researchers have not determined which genes on the X chromosome are responsible for most of the features of Turner syndrome. They have, however, identified one gene called SHOX that is important for bone development and growth. The SHOX gene is located in the pseudoautosomal regions of the sex xhoromosomes. Missing one copy of this gene likely causes short stature and skeletal abnormalities in women with Turner syndrome (6,7).

Results and Discussion

The X chromosome has many genes related to growth and development. The Y chromosome is smaller and contains fewer genes. Females have two X chromosomes (XX), so when one copy of the gene on one X chromosome changes, the normal gene copy on the other X chromosome is sufficient for normal growth and development, briefly supporting the modified copy. In such a situation, the female individual is healthy but carrier for the disease caused by X. Carrier means that an altered copy of the gene is transported without the patient. In some cases, females may show some signs of the disease. Although scientists have known this phenomenon, called X chromosome inactivation, for more than fifty years, little is still known about how it occurred or how it developed. At the same time, every copy of the X chromosome contains a variety of genes that are not in another. So having two X chromosomes gives females greater genetic diversity than males with one X chromosome. For this reason, ideas have been discussed in recent years as females have a genetic diversity that scientists have just begun to understand. We think that detailed cytogenetic studies should be done on this subject.

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