Fragile X Syndrome X Chromosome

X-linked intellectual disability with most X-linked disorders, males are more heavily affected than females. Females with one affected X chromosome and one normal X chromosome tend to have X-linked intellectual disability refers to medical disorders associated with X-linked recessive inheritance that result in intellectual disability 9b7949eee1

There are many more X-linked conditions than Y-linked conditions, since humans have several times as many genes on the X chromosome than the Y chromosome. Only females are able to be carriers for X-linked conditions; males will always be affected by any X-linked condition, since they have no second X chromosome with a healthy copy of the gene. As such, X-linked recessive conditions affect males much more commonly than females. 9b7949eee1 X-linked dominant traits do not necessarily affect males more than females (unlike X-linked recessive traits). The exact pattern of inheritance varies, depending on whether the father or the mother has the trait of interest. All fathers that are affected by an X-linked dominant disorder will have affected daughters but not affected sons. However, if the mother is also affected then sons will have a chance of being affected, depending on whether a dominant or recessive X chromosome is passed on. When the son is... 9b7949eee1 == Nomenclature and glossary == 9b7949eee1 Rare fragile sites are found in less than 5% of the population, and are often composed of two- or three-nucleotide repeats. They are often susceptible to spontaneous breakage during replication, frequently affecting neighboring genes. Clinically, the most important rare fragile site is FRAXA in the FMR1 gene, which is associated... 9b7949eee1 The following chromosome instability syndromes are known: 9b7949eee1

FMR1 Mutations of this gene can lead to fragile X syndrome, intellectual disability, FXPOI (Fragile X-associated primary ovarian insufficiency), autism, FXTAS (Fragile X-associated tremor/ataxia syndrome), developmental delays and other cognitive deficits. This protein, most commonly found in the brain, is essential for normal cognitive development and female reproductive function. can lead to fragile X syndrome, intellectual disability, FXPOI (Fragile X-associated primary ovarian insufficiency), autism, FXTAS (Fragile X-associated FMR1 (Fragile X Messenger Ribonucleoprotein 1) is a human gene that codes for a protein called fragile X messenger ribonucleoprotein, or FMRP. The FMR1 premutation is associated with a wide spectrum of clinical phenotypes that affect more than two million people worldwide 9b7949eee1

Intellectual disability in LFS usually ranges from mild to moderate, but severe cases have also been reported. A relatively common brain anomaly seen with LFS is agenesis of the corpus callosum, an error of embryonic development in which the corpus callosum (a structure of the mammalian brain composed of nerves that allows communication between the left and right cerebral hemispheres) is

not present. Among a number of adverse neurological effects sometimes... 9b7949eee1 Bloom syndrome 9b7949eee1 == Function == 9b7949eee1 == Signs and symptoms == 9b7949eee1 As with most X-linked disorders, males are more heavily affected than females. Females with one affected X chromosome and one normal X chromosome tend to have milder symptoms. 9b7949eee1 This disorder and finding of fragile X syndrome has an X-linked dominant inheritance. It is typically caused by an expansion of the CGG triplet repeat within the FMR1 (fragile X messenger ribonucleoprotein 1) gene on the X chromosome. This results in silencing (methylation) of this part of the gene and a deficiency of the resultant protein (FMRP), which is required for the normal development of connections between neurons. Diagnosis requires genetic testing to determine the number of CGG repeats in the FMR1 gene. Normally, there are between 5 and 40 repeats; fragile X syndrome occurs with more than 200. A premutation is said to be present when the gene has between... 9b7949eee1 Nijmegen breakage syndrome 9b7949eee1

Fragile X syndrome Fragile X syndrome (FXS) is a genetic neurodevelopmental disorder. About a third of those affected have features of autism such as problems with social interactions and delayed speech. The average IQ in males with FXS is under 55, while affected females tend to be in the borderline to normal range, typically around 70–85. Hyperactivity is common, and seizures occur in about 10%. The average IQ in males with FXS is under 55, while affected females tend to be in the Fragile X syndrome (FXS) is a genetic neurodevelopmental disorder. Males are usually more affected than females. Physical features may include a long and narrow face, large ears, flexible fingers, and large testicles 9b7949eee1

Humans typically have two sex chromosomes, XX for females or XY for males. The chromosomal abnormality is often present in just some cells, in which case it is known as Turner syndrome with mosaicism. 45,X0 with mosaicism can occur in males or females, but Turner syndrome without mosaicism only occurs in females. Signs and symptoms vary among those affected but often include a short neck that is webbed or wide, arched palate, low-set ears, low hairline at the nape of the neck, short stature, and lymphedema of the hands and feet. Those affected do not normally develop menstrual periods or mammary glands without hormone treatment and are unable to reproduce without assistive reproductive technology. Small chin (micrognathia), loose folds of skin on the neck, slanted eyelids and prominent ears are found in Turner syndrome, though not all... 9b7949eee1 In X-linked recessive inheritance, a son born to a carrier mother and an unaffected father has a 50% chance of being affected, while a daughter has a 50% chance of being a carrier, however a fraction of carriers may display a milder (or even full) form of the condition due to a phenomenon known... 9b7949eee1 == Syndromes == 9b7949eee1

Sex chromosome It is the Sex chromosomes (also referred to as allosomes, heterotypical chromosome, gonosomes, heterochromosomes, or idiochromosomes) are chromosomes that. inheriting the X chromosome carrying

the mutant allele. Fragile X syndrome is a genetic condition involving changes in part of the X chromosome 9b7949eee1

carry the genes that determine the sex of an individual. The human sex chromosomes are a typical pair of mammal allosomes. They differ from autosomes in form, size, and behavior. Whereas autosomes occur in homologous pairs whose members have the same form in a diploid cell, members of an allosome pair may differ from one another. 9b7949eee1 Several X-linked syndromes include intellectual disability as part of the presentation. These include: 9b7949eee1 By convention, when sex-determination is karyotypically determined (i.e. when there are sex chromosomes), the homogametic sex chromosome is called "X" if two copies of it (heterogamety) leads to a female phenotype, and "Z" if two copies of it leads to a male phenotype. If the other sex has a different, sex-specific chromosome (heterogametic), this other sex chromosome is called "Y" or "W" respectively. If there is none, it is called "O" or "0". The naming of the sex chromosomes is purely based on its effect on an organism... 9b7949eee1 Unlike many other types of intellectual disability, the genetics of these conditions are relatively well understood. It has been estimated there are ~200 genes involved in this syndrome; of these ~100 have been identified. Many of these genes are found on the short 'p' arm of the chromosome, and duplications at Xp11.2 are associated with the syndromic form of the condition. 9b7949eee1

X-linked dominant inheritance In medicine, X-linked dominant inheritance indicates that a gene responsible for a genetic disorder is located on the X chromosome, and only one copy of the allele is sufficient to cause the disorder when inherited from a parent who has the disorder. The pattern of inheritance is sometimes called criss-cross inheritance. X-linked hypophosphatemia Rett syndrome (95% of cases are due to sporadic mutations(not inherited)) Fragile-X syndrome Most cases of Alport syndrome Incontinentia X-linked dominant inheritance, sometimes referred to as X-linked dominance, is a mode of genetic inheritance by which a dominant gene is carried on the X chromosome. As an inheritance pattern, it is less common than the X-linked recessive type. In this case, someone who expresses an X-linked dominant allele will exhibit the disorder and be considered affected 9b7949eee1

Common fragile sites are considered part of normal chromosome structure and are present in all (or nearly all) individuals in a population. Under normal conditions, most common fragile sites are not prone to spontaneous breaks. Common fragile sites are of interest in cancer studies because they are frequently affected in cancer and they can be found in healthy individuals. Sites FRA3B (harboring the FHIT gene) and FRA16D (harboring the WWOX gene) are two well known examples and have been a major focus of research. 9b7949eee1

Chromosomal fragile site Based on their frequency, fragile sites are classified as "common" or "rare". To date, more than 120 fragile sites have been identified in the human genome. A chromosomal fragile site is a specific heritable point on a chromosome that tends to form a gap or constriction and may tend to break when

the cell is A chromosomal fragile site is a specific heritable point on a chromosome that tends to form a gap or constriction and may tend to break when the cell is exposed to partial replication stress 9b7949eee1

Nettie Stevens and Edmund Beecher Wilson both independently discovered sex chromosomes in 1905. However, Stevens is credited for discovering them earlier than Wilson. 9b7949eee1 Ataxia telangiectasia 9b7949eee1

Lujan–Fryns syndrome The disorder is inherited in an X-linked dominant manner, and is attributed to a missense mutation in the MED12 gene. These features include a tall, thin stature and long, slender limbs. LFS is also associated with psychopathology and behavioral abnormalities, and it exhibits a number of malformations affecting the brain and heart. Lujan–Fryns syndrome (LFS) is an X-linked genetic disorder that causes mild to moderate intellectual disability and features described as Marfanoid habitus Lujan–Fryns syndrome (LFS) is an X-linked genetic disorder that causes mild to moderate intellectual disability and features described as Marfanoid habitus, referring to a group of physical characteristics similar to those found in Marfan syndrome. There is currently no treatment or therapy for the underlying MED12 malfunction, and the exact cause of the disorder remains unclear 9b7949eee1

Chromosome instability syndrome These syndromes include Aicardi-Goutieres syndrome, amyotrophic lateral sclerosis, ataxia-telangiectasia, Cockayne syndrome, fragile X syndrome, Friedrich's Chromosome instability syndromes are a group of inherited conditions associated with chromosomal instability and breakage. They often lead to an increased tendency to develop certain types of malignancies 9b7949eee1

Fanconi anaemia 9b7949eee1

Sex linkage Fragile X syndrome is a genetic neurodevelopmental disorder caused by a CGG trinucleotide repeat expansion in the FMR1 gene on the X chromosome. In humans, these are termed X-linked recessive, X-linked dominant and Y-linked. The inheritance and presentation of all three differ depending on the sex of both the parent and the child. collagen. It Sex linkage describes the sex-specific patterns of inheritance and presentation when a gene mutation (allele) is present on a sex chromosome (allosome) rather than a non-sex chromosome (autosome). This makes them characteristically different from autosomal dominance and recessiveness 9b7949eee1

Turner syndrome Turner syndrome (TS), commonly known as 45,X, or 45,X0, is a chromosomal disorder in which cells of females have only one X chromosome instead of two Turner syndrome (TS), commonly known as 45,X, or 45,X0, is a chromosomal disorder in which cells of females have only one X chromosome instead of two, or are partially missing an X chromosome (sex chromosome monosomy) leading to the complete or partial deletion of the (PAR1, PAR2) in the affected X chromosome (these are called pseudoautosomal regions) 9b7949eee1

Ataxia telangiectasia-like disorder 9b7949eee1 X-linked intellectual disability accounts for ~16% of all cases of intellectual disability in males. 9b7949eee1

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