

X linked recessive inheritance

X-linked recessive inheritance is a mode of inheritance in which a mutation in a gene on the X chromosome causes the phenotype to be expressed in males (who are necessarily hemizygous for the gene mutation because they have one X and one Y chromosome) and in females who are homozygous for the gene mutation, see zygosity.

X-linked inheritance means that the gene causing the trait or the disorder is located on the X chromosome. Females have two X chromosomes, while males have one X and one Y chromosome. Carrier females who have only one copy of the mutation do not usually express the phenotype, although differences in X chromosome inactivation can lead to varying degrees of clinical expression in carrier females since some cells will express one X allele and some will express the other. The current estimate of sequenced X-linked genes is 499 and the total including vaguely defined traits is 983.

Some scholars have suggested discontinuing the terms dominant and recessive when referring to X-linked inheritance due to the multiple mechanisms that can result in the expression of X-linked traits in females, which include cell autonomous expression, skewed X-inactivation, clonal expansion, and somatic mosaicism.

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