

How do geneticists indicate the location of a gene?

Geneticists use maps to describe the location of a particular gene on a chromosome. One type of map uses the cytogenetic location to describe a gene's position. The cytogenetic location is based on a distinctive pattern of bands created when chromosomes are stained with certain chemicals. Another type of map uses the molecular location, which is a precise description of a gene's position on a chromosome. The molecular location is based on the sequence of DNA building blocks (nucleotides) that make up the chromosome.

Cytogenetic location

Geneticists use a standardized way of describing a gene's cytogenetic location. In most cases, the location describes the position of a particular band on a stained chromosome:

17q12

It can also be written as a range of bands, if less is known about the exact location:

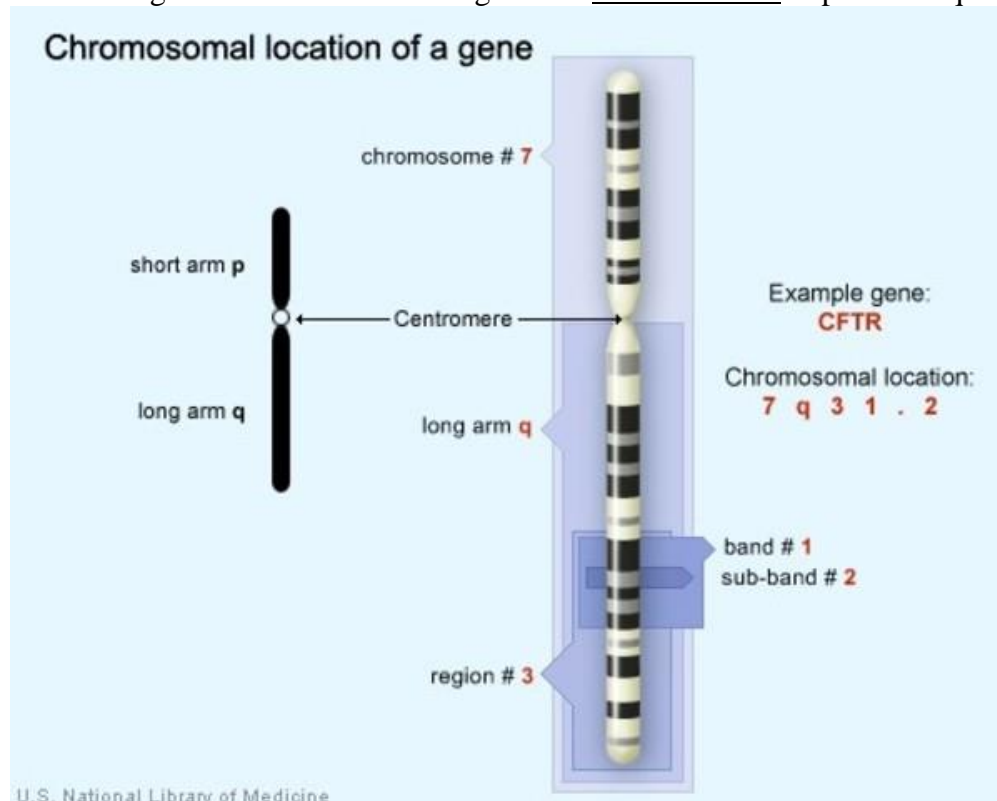
17q12-q21

The combination of numbers and letters provide a gene's "address" on a chromosome. This address is made up of several parts:

- The chromosome on which the gene can be found. The first number or letter used to describe a gene's location represents the chromosome. Chromosomes 1 through 22 (the autosomes) are designated by their chromosome number. The sex chromosomes are designated by X or Y.
- The arm of the chromosome. Each chromosome is divided into two sections (arms) based on the location of a narrowing (constriction) called the centromere. By convention, the shorter arm is called p, and the longer arm is called q. The chromosome arm is the second part of the gene's address. For example, 5q is the long arm of chromosome 5, and Xp is the short arm of the X chromosome.
- The position of the gene on the p or q arm. The position of a gene is based on a distinctive pattern of light and dark bands that appear when the chromosome is stained in a certain way. The position is usually designated by two digits (representing a region and a band), which are sometimes followed by a decimal point and one or more additional digits (representing sub-bands within a light or dark area). The number indicating the gene position increases with distance from the centromere. For example: 14q21 represents position 21 on the long arm of chromosome 14. 14q21 is closer to the centromere than 14q22.

Sometimes, the abbreviations “cen” or “ter” are also used to describe a gene's cytogenetic location. “Cen” indicates that the gene is very close to the centromere. For example, 16pcen refers to the short arm of chromosome 16 near the centromere. “Ter” stands for terminus, which indicates that the gene is very close to the end of the p or q arm. For example, 14qter refers to the tip of the long arm, or the very end, of chromosome 14.

The *CFTR* gene is located on the long arm of chromosome 7 at position 7q31.2.



Molecular location

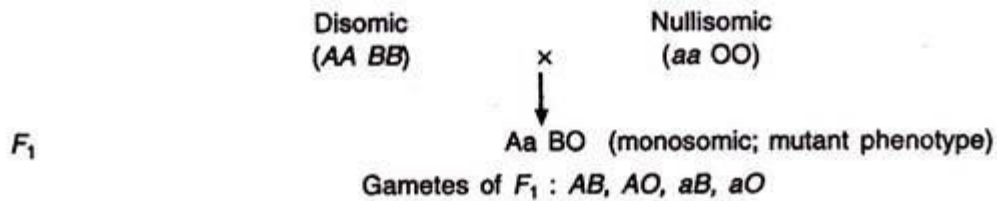
The Human Genome Project, an international research effort completed in 2003, determined the sequence of nucleotides for each human chromosome. This sequence information allows researchers to provide a more specific address than the cytogenetic location for many genes. A gene's molecular address pinpoints the location of that gene in terms of nucleotides. It describes the gene's precise position on a chromosome and indicates the size of the gene. Knowing the molecular location also allows researchers to determine exactly how far a gene is from other genes on the same chromosome.

Different groups of researchers often present slightly different values for a gene's molecular location. Researchers interpret the sequence of the human genome using a variety of methods, which can result in small differences in a gene's molecular address.

Use of nullisomics in locating genes on chromosomes

Nullisomics are used to assign genes to particular chromosomes. If the dominant mutant gene (A) to be located is present in the chromosome of a disomic parent, then a cross between this disomic line (AABB) and nullisomic line for gene (B) but disomic for gene A will produce allmonosomic F_1 plants. The F_1 monosomic plants are selfed and the segregation for the concerned gene (A) is studied in F_2 .

If the concerned gene (A) is located in the chromosome of a nullisomic parent (in other words, if the nullisomic parent is disomic for the concerned mutant gene), then in F_2 both (Fig. 16.8) mutant and nonmutant phenotypes will appear in all the types of F_2 plants, viz., disomic, monosomic and nullisomic plants. In case of nullisomics, the frequency of mutant and nonmutant phenotypes will be 3: 1.



F_2

♀ \ ♂		AB	AO	aB	aO
AB	AABB	AABO	AaBB	AaBO	
AO	AABO	AAOO	AaBO	AaOO	
aB	AaBB	AaBO	aaBB	aaBO	
aO	AaBO	AaOO	aaBO	aaOO	

- (i) Ratio of mutant (A) and nonmutant (a) phenotypes = 3 : 1.
- (ii) Both mutant and nonmutant phenotypes found in disomics, monosomics and nullisomics.
- (iii) Nullisomics show mutant phenotype in higher frequency than nonmutant phenotype.

Fig. 16.8. F_2 segregation for a dominant mutant gene (A) located in the chromosome for which the plant is disomic. Genes A and B are located in nonhomologous chromosomes; the aneuploid plant is nullisomic for the chromosome carrying the gene B.

On the other hand, if the dominant mutant gene is located in the chromosome of a disomic parent when crossed to parent nullisomic for the concerned gene (A) (That is, the nullisomic line lack the concerned dominant gene but disomic for the other gene) (Fig. 16.9), all the nullisomics will show the non-mutant phenotype in the F_2 , while all the other types of plants, viz., disomics and monosomics will exhibit the mutant phenotype.

Disomic (AABB) × Nullisomic (OOBB)

F_1 AOBB (Monosomic ; mutant phenotype)

Gametes : AB, OB

F_2

♀ \ ♂	AB	OB
AB	AABB	AOBB
OB	AOBB	OOBB

- (i) All the plants, except the nullisomics, have the mutant phenotype.
- (ii) All the nullisomics have the nonmutant phenotype.

Fig. 16.9. Segregation in F_2 for a dominant gene A located in the chromosome for which the aneuploid parent is nullisomic. Genes A and B are located in nonhomologous chromosomes; gene A located in the critical chromosome for which the aneuploid stock is nullisomic.