Editorial

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Is Heterogeneity in the Nature of the Mutation?

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Description

As a result, there are heterozygous carriers, who are phenotypically normal. The offspring of two carriers of a recessive trait have a 25% chance of developing the disease phenotype, through being homozygous for the recessive allele. Similarly, 25% are be homozygous for the type allele and 50% will become heterozygous carriers. When one parent has an autosomal recessive disorder, all children are heterozygous carriers they will all inherit one disease allele. In cases where the other parent is also a carrier for the same disease, pseudo dominance will occur 50% of the children will be homozygous for the mutation and the other 50% will heterozygous carriers. Often, recessive diseases result from mutations in genes, leading to a loss of function of the encoded protein. Many recessive mutations occur in genes encoding enzymes. Where the functional 50% of the enzyme derived from the type allele is sufficient to compensate for the non-functional allele. There may be heterogeneity in the nature of the mutation causing autosomal recessive disorders.

Locus heterogeneity refers to the fact that an inherited disorder can be caused by mutations in more than one gene. This occurs in deafness, which can be caused by mutation of one of many different genes. As a result of schooling and interactions in the deaf community. Deaf people often have children with other deaf people. Their children may be phenotypically normal: they will be heterozygotes for two different genes, at different genomic locations.

Mutational heterogeneity results from the fact that more than one mutation exists that can result in a non-functional protein product. A variety of alleles may be associated with a specific disease. Most autosomal recessive disorders are result from compound heterozygous alleles two different mutations at the same locus have been inherited. It Is unlikely that an individual is truly heterozygous (where both alleles have the same mutation) unless there is some consanguinity the disease causing allele having been inherited from a common ancestor. Disorders associated with mutations at loci on the X chromosome show a particular inheritance pattern. Only males are usually affected. Because they possess a single X chromosome: the mutation cannot be masked by a type allele. Females carrying the mutation are almost always heterozygous carriers with a normal phenotype. Although daughters of afflicted fathers are obligate carriers of the condition (they must inherit the affected X chromosome). Some conditions may be predominantly transmitted by the mother because affected males may not survive to reproductive age. Genome, whereas Liz twins share only 50% of genes similar to the siblings traits where there is a large inherited component are be much more frequent. Traits where the heritable component is low are show similar inheritance in MZ and DZ twins.

Susceptibility genes it is possible to identify specific loci in polygenic diseases that increase the likelihood of developing the disease. Many diseases show a familial clustering of inheritance, although there is obviously no form of mendelian inheritance. Type I diabetes represents the prototypic polygenic disease, and many of the 'susceptibility loci' fur the condition have been identified.