Inbreeding, Consanguinity and Inherited Diseases

Inbreeding

Inbreeding describes the mating between two blood relative (consanguineous relatives), the degree of inbreeding is normally measure Coefficient of Inbreeding. The value of this coefficient is equal to the probability that an individual will have inherit 2 alleles of a gene that are "identical by descent" (the same form of an allele inherited from a single common ancestor). The coefficient is of medical significant when considering autosomal recessive conditions and genetic load. The phenomenon by which the number of individuals affected by autosomal recessive disorders increases due to inbreeding is referred to as inbreeding depression and results in the reduction of the overall fitness of a population.

Inbreeding is used to create inbred strains of rats and mice (normally requires 20 generations of inbreeding) which are genetically identical. Members of the inbred stain can be genetically altered normally by knocking out or altering a single gene to observe its effect. They have provided a large deal of information about the function of specific genes.

Consanguinity

A consanguineous marriage/mating is considered as being between two individuals who are 1^{st} to 4^{th} degree relatives, when considering 1^{st} degree relatives the term incestuous marriage/mating (or simply incest) is often used. The degree to which two people are related is usual measured via the Coefficient of Relatedness which is also referred to as the coefficient of consanguinity. The value generated represents the proportion of identical by descent alleles that two individuals will share.

Degree of Relationship	Example of relative	Coefficient of Relatedness
1^{st}	Brother, Sister, Mother, Father, Daughter, Son	1/2
2^{nd}	Grandfather, Grandmother, Granddaughter, Grandson, Nephew, Uncle	1/4
3^{rd}	Great Grandfather, Great Granddaughter, Cousins, Great Nephew	1/8
4^{th}	Great Great Grandfather, Great Great Grandson, Great Great nephew	1/16

In a normal population only about 1% of marriages will be consanguineous however in some isolated small populations this level can increase to in excess of 40%. This level of inbreeding also occurs after events that cause large reductions in a number of individuals in a population, such events are referred to as bottle neck events.

Inherited Diseases

Mendel identified two patterns of inheritance of traits which he explained as being due to some alleles being dominant while others are recessive, explained by his Laws of Inheritance. The patterns of inheritance were seen to only be accurate for autosomes and not genes carried on the sex chromosomes therefore diseases inherited in a manner true to either of the patterns are referred to as autosomal dominant or autosomal recessive. In addition to being inherited in a mendelian fashion some disease display Extrachromosomal and Non-Mendelian Inheritance.

Autosomal Recessive	Autosomal Dominant	X linked Recessive	X linked Dominant	Imprinting	Mitochondrial	Multifactorial
Albanism	Osteogenesis imperfecta	Hemophilia A	Vitamin D Resistant Rickets	Angelman Syndrome	Leber's hereditary optic neuropathy	Crohn's Disease
Sickle Cell Anemia	Achondroplasia	Hemophilia B	Incontinentia Pigmenti	Prader-Willi Syndrome	Diabetes mellitus and deafness	Autism
Cystic Fibrosis	Huntington's Disease	X linked Colour Blindness	Rett Syndrome			Diabetes Mellitus
Phenylketonuria	Familial Hypercholesterolemia	Duchenne Muscular Dystrophy				

Links

Related Articles

- Multifactorial Inheritance
- Laws of Inheritance
- Coefficient of Relatedness
- Coefficient of Inbreeding
- The Molecular Basis of Genetic Diseases

- Multifactorial Inheritance, Heritability

Bibliography

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