# 22 - 8. Polymorphisms

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© SPMM Course Multifactorial inheritance It is a complex inheritance in which multiple genes are involved jointly with environmental influences. Most common psychiatric disorders such as schizophrenia do not show a Mendelian pattern of inheritance. But these disorders are categorically defined as present or absent hence cannot be regarded as continuous variables too. But these conditions could be regarded as quasi-continuous in that those who are affected can be graded along a continuum of severity. So we can also assume that there is an underlying liability to develop the disorder, which is continuously distributed in the population. Those who pass a certain threshold manifest the condition. This is known as the liability/threshold model. If the underlying liability to develop the disorder is inherited in a multifactorial fashion, one can assume that the distribution will be approximately distributed along a normal distribution curve. But compared to the normal population, the genetic liability of relatives of affected individuals will be increased, and their liability distribution will be shifted to the right. Thus, the proportion of relatives above the disease threshold will be greater compared with the general population. If we know the proportion of affected relatives of probands and the proportion of those affected in the general population, it is possible to calculate the correlation in liability between pairs of relatives using this model. Recurrence risks to relatives for multifactorial disorders are influenced by the disease severity, the degree of relationship to the index case, the number of affected close relatives and, if there is a higher incidence in one particular sex, the sex of the index case. Polygenic inheritance Polygenic inheritance is again a complex inheritance in which multiple genes but no environmental factors are involved. Both polygenic and multifactorial inheritances defy normal Mendelian principles. The additive effects of many genes, i.e. polygenic inheritance, probably cause characteristics such as height and intelligence, which show a normally distributed continuous distribution in the general population. 8. Polymorphisms Polymorphism refers to variations in genetic make-up at a particular locus noted in general, apparently healthy population. To be defined as polymorphism the variant must occur in at least 1% of the total population and must be associated with normal but varied (not disease causing) expression of final phenotype. This excludes spontaneous mutations that are random and so cannot simultaneously occur in such significant (1%) proportion of total population. ABO blood groups are good examples of polymorphism expressed in protein products of genes.  Restriction fragment length polymorphisms are variations that change the sites at which restriction enzymes can act on a DNA molecule, rendering differences in the final ‘restricted’ or cleaved DNA when these enzymes are applied in vitro (Southern Blotting).  If polymorphisms are due to changes in single nucleotide in a sequence, then these are called SNPs or single nucleotide polymorphisms. These single-base polymorphisms can be assayed by DNA sequencing or through the use of DNA chips.  If the variations are due to changes in length of the genetic sequence, these are termed length polymorphisms.

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