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DNA Diversity, Disease and Diagnosis

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There is a considerable similarity in the human genome between different individuals. There are also small variations in the DNA sequence which have been associated with differences in disease susceptibility and prognosis. The preponderance of a particular variant of a gene in a group of individuals with a disease may provide a clue to predict predisposition to a specific disease. However susceptibility to complex traits such as mental retardation could be related to the cummulative effects of several genes, with one or more variants of small effect size occuring with a specific frequency within the genome, combined with environmental factors. This implies that the genes responsible for disorders such as learning disabilities may represent the quantitative extreme of the same genetic and environmental factors that cause variation throughout the normal distribution. This lecture covers the main areas of variations in the human genome and their association with disease. Specific reference would be made to the penetrance and expressibility of genes in selected disorders including those with a neurological aetiology. The approach to clinical genetic screening and the diagnostic strategies for these diseases are also reviewed.