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Detection and Characterisation of Genetic Material

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The current approach to the understanding and diagnosis of disease involves identification and characterisation of the candidate disease genes. Various molecular diagnostic methods are used to establish the specific disease risk profiles for several genes implicated in many genetic disorders. These procedures apply the information from the human genome sequence to screen and test for gene aberrations. Given that a sizable majority of the sequenced human genes are expressed either exclusively or preferentially in the brain, neuroscience stands to benefit extensively from this transition to a molecular diagnostic discipline. This lecture highlights the application of molecular biology tools in neurogenetics. It provides an outline of the currently applied "single gene" diagnostic strategies for DNA characterisation and extends to include the significance of the application of some high throughput procedures, in measuring the abundance of DNA and RNA, gene expression analysis and functional polymorphism, for genome analysis of complex traits and common disorders.