

Poster presentation

Behavioural phenotypes in genetic syndromes: a window on to the biology of behaviour

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Background

A behavioral phenotype (BP) is the characteristic cognitive, personality, behavioral, and psychiatric pattern that typifies a disorder. A number of genetic syndromes have been identified as having this type of distinctive and consistent behaviour pattern.

Materials and methods

In this article we aim to illustrate information currently available in the field of behaviour phenotypes. Also described are the brain-behavior relations, and gaining understanding of the genetic bases of behavior.

Results

They were organized by dividing them into three groups according to the biological characteristics recognized to date: a) BP associated to genetic diseases with an identified biological basis (syndromes such as Lesch Nyhan, Rett, fragile X, tuberous sclerosis complex, Noonan, Sotos, Aicardi, Angel man, Prader Willi, Williams, Down, Smith Magenis, Di George, Pallister Killian and Turner, among others; b) BP associated to a genetic disease with an unidentified biological basis (Cornelia de Lange syndrome); and, c) BP with an as yet unidentified biological basis associated to diverse causations (autism). Because of the rarity of many of the syndromes, and the complexity of their genetic basis, there are great difficulties in establishing the validity of the association between syndrome and behavioural phenotype. The combination of molecular genetics, neuroimaging, and behavioral research has advanced our understanding of the linkages between genetic variables, neurobiological measures, IQ, and behaviour.

Discussion

Although neither the genome nor the environment can be manipulated in research on human behaviour, some of the new tools of molecular genetics can be brought to bear on human behavioural disorders. In addition, because they are the consequences of known genetic alterations, behavioral phenotypes can be potentially highly valuable clues to the identification of genes in the population that are important to determination of cognitive skills or deficits, personality determinants, behavioral abnormalities, or psychiatric disorders.