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Review article

Phenotype–environment interactions in genetic syndromes associated with severe or profound intellectual disability

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ABSTRACT

The research literature notes both biological and operant theories of behavior disorder in individuals with intellectual disabilities. These two theories of genetic predisposition and operant reinforcement remain quite distinct; neither theory on its own is sufficient to explain challenging behavior in genetic syndromes and an integrated approach is required. This literature review integrates the two approaches by exploring how environmental factors can influence problem behavior in genetic syndromes associated with intellectual disability. Particular attention is paid to studies that describe evidence that problem behaviors in syndromes that are considered to be phenotypic are associated with other aspects of an established behavioral phenotype. The review highlights how the study of phenotype–environment interactions within syndromes can promote understanding of the aetiology of problem behaviors both within genetic syndromes and, ultimately, the wider population of individuals with severe intellectual disabilities. The review also evaluates the current status of research and the methods typically employed.

Implications for intervention, future research and extending existing causal models of challenging behavior are discussed.

Contents

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1. Introduction

Challenging behavior is a significant problem that can have an impact on the lives of those displaying such behaviors, as well as those who care for them (Hassiotis, Parkes, Jones, Fitzgerald, & Romeo, 2008; Hastings, 2002; Olsson & Hwang, 2001; Schwartz & Rabinovitz, 2003). There is growing evidence that challenging behavior is more common in some genetic syndromes than would be expected, given other characteristics such as degree of intellectual disability (e.g. Anderson & Ernst, 1994; Berney, Ireland, & Burn, 1999; Clarke & Boer, 1998; Collins & Cornish, 2002; Dykens & Clarke, 1997; Dykens & Smith, 1998; Holland, Whittington, Webb, Boer, & Clarke, 2003; Hyman, Oliver, & Hall, 2002; Symons, Clarke, Hatton, Skinner, & Bailey, 2003). The focus of this systematic review is the interaction between some of the established characteristics of the behavioral phenotype of genetic syndromes and environmental influences on challenging behavior.

A review of this empirical research is important in order to evaluate the current status of evidence for interactions that might inform more complete models of challenging behavior and highlight potentially productive areas for further work. There is emerging evidence in the literature to suggest that problem behaviors associated with genetic syndromes could be influenced by an interaction between an aspect of the behavioral phenotype and operant processes (e.g. O'Reilly, 1997; Oliver, Murphy, Crayton, & Corbett, 1993; Taylor & Oliver, 2008). Examination of specific forms of phenotype–environment interactions within syndromes will promote understanding of the aetiology of problem behaviors both within genetic syndromes and, ultimately, the wider population of individuals with severe intellectual disabilities and extend existing causal models.

Prior to the review, the seemingly opposing biological and environmental theories are described briefly to provide context. A systematic review follows with critique of methodology used in the study of problem behavior associated with genetic syndromes. Environmental influences on behaviors within genetic syndrome research are detailed and relationships between behavioral phenotypes and features of syndromes are discussed. Finally, the review will highlight the importance of functional analytic studies that incorporate facets of behavioral phenotypes to further understand the behavior of children and adults with genetic syndromes. Future research is discussed with particular reference to effective early intervention strategies.

1.1. Apparently opposing theories of challenging behavior

There is robust evidence from cohort studies that challenging behavior in people with intellectual disabilities is associated with a number of characteristics or risk markers such as a greater degree of intellectual disability, communication impairments, Autism Spectrum Disorder and the presence of stereotyped, compulsive and impulsive behaviors (Bodfish et al., 1995; Brylewski & Wiggs, 1999; Deb, Thomas, & Bright, 2001; McClintock, Hall, & Oliver, 2003; Powell, Bodfish, Parker, Crawford, & Lewis, 1996; Rojahn, Matson, Naglieri, & Mayville, 2004). In addition, genetic syndromes are a significant risk marker for the development of challenging behavior (Arron et al., 2006) and might thus be considered part of the behavioral phenotype for some syndromes.

A behavioral phenotype is defined by an increased probability of behavioral characteristics evident in those with a syndrome compared with individuals without the syndrome (Dykens, 1995). Evidence suggests that certain forms of self-injurious and aggressive behavior may constitute part of the behavioral phenotype of a number of genetic syndromes. Gene-behavior associations of varying specificity have been demonstrated repeatedly across a number of syndromes, for example, Cri du Chat, Cornelia de Lange, Lesch-Nyhan, Fragile-X, Smith-Magenis and Angelman syndromes (Finucane, Simon, & Dirrigl, 2001; Horsler & Oliver, 2006a; Nyhan, 1972; Symons et al., 2003).

In syndromes in which estimates of challenging behavior are consistently higher than might be expected, it has often been assumed that the behavior has strong biological determinants. One line of evidence in the literature concerns neurotransmitter systems, more specifically the dopamine, opioid and serotonin systems and how these may be abnormal. Much research over the last 20 years has focussed on the role of neurotransmitters in the expression of self-injury in some individuals. For example, in Lesch-Nyhan syndrome where self-injurious behavior (SIB) is observed in almost all individuals with the syndrome (Christie et al., 1982), the dopaminergic system has been implicated (Clarke, 1998). In brief, evidence arises from neuropathological, neuroimaging and neurochemical studies of individuals with Lesch-Nyhan syndrome (Schroeder et al., 2001). Functional loss of dopamine terminals has been found in positron-emission tomography studies of healthy individuals with Lesch-Nyhan syndrome and in post mortem studies. It has also been suggested that there is a super-sensitivity of postsynaptic dopamine receptors that results from the loss of dopamine terminals and this dopamine loss acts to mediate the self-injury (Casas-Bruge et al., 1985 cited by Ernst et al., 1996; Clarke, 1998; Turner & Lewis, 2002). Van Acker (1991) also suggested that abnormalities in the dopamine system might account for the hand stereotypies and loss of purposeful hand movements that are associated with Rett syndrome.

Other studies have examined a broader range of potential biological factors and identify brain regions that may be centrally involved in the expression of SIB. Several researchers have identified abnormalities of the basal ganglia as potentially associated with self-injury. The basal ganglia are made up of several structures, including the striatum and the globus pallidus. Lesions to the basal ganglia in humans have been associated with a variety of outcomes, including movement disorders, speech disorders, obsessive-compulsive behaviors and disinhibition (Bhatia & Marsden, 1994).