

Behavioural phenotypes

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J R Soc Med 2000;93:618-620

RSM FORUM ON LEARNING DISABILITY AND THE SOCIETY FOR THE STUDY OF BEHAVIOURAL PHENOTYPES, 11 MAY 2000

How much of behaviour is genetic? Putting it another way, to what extent can genetic study ultimately reveal the determinants of human behaviour? This vexed question is being increasingly explored in many quarters—in the scientific and clinical press as well as the popular media, often in emotive terms. While no single field of research has the capacity to address all the issues, one area of enquiry that has contributed substantially to the debate is the study of behavioural phenotypes. So what are behavioural phenotypes? Broadly speaking, they are patterns or sets of behaviours that are dependent on genotype¹. The essence of the behavioural phenotype concept, as it is most often applied, was captured at the first meeting of the Society for the Study of Behavioural Phenotypes²—‘that some of the behaviours exhibited by children with biologically based mentally handicapping disorders are organically determined’. Notably, when a given syndrome or cause of mental retardation is described as featuring a certain behavioural phenotype, it is not proposed that all individuals affected by the condition in question will have all the behaviours in question. This is crucially important. In this definition it is emphasized that many other influences—developmental, biological, and environmental—operate in complex interactions with, and constantly shape, those behaviours³.

BACKGROUND

When we trace the history of the behavioural phenotype concept, certain strands come together. First, it is noteworthy that the term behavioural phenotype comes from animal-based ethological research, where it has long been employed to describe the behavioural characteristics of different genetic strains in laboratory animals. This origin tends to lend something of a reductionist feel to the term, and leans towards an implication of a single genotype/phenotype relationship. Certainly, when the term behavioural phenotypes was incorporated into clinical paediatrics, it was in respect of what was clearly and manifestly the occurrence of a well-defined, biologically driven behaviour in a specific genetic disorder—namely, the compulsive self-mutilation of the Lesch–Nyhan syndrome⁴.

More often, however, behavioural phenotypes are less dramatic, and their associations with causal syndromes of learning disabilities are less clear-cut. For, although undergraduates have long been taught that ‘the phenotype is the expression of the genotype’, the emphasis is on expression—developing and changing over time, not as a simple or direct result. Consequently, we have the longstanding set of observations that the phenotypes of the different major causal syndromes of learning disability are discernible not only in their respective dysmorphology, predisposition to medical problems and typical ranges of intellectual disability, but also in their patterns of behaviour and temperament. Here, when Langdon Down described his eponymous syndrome, he stressed that a ‘typical character’ was found in these individuals⁴. One key element of Down’s account is now rightly and thoroughly discredited; his notion of ‘Mongoloid idiocy’ was based on fallacious eugenic thinking. But the broader message which endures is that certain genetic syndromes of learning disability do result in relatively specific patterns of behaviour and related attributes. This theme has been revisited and energetically elaborated most recently by a group that has given immense impetus to the delineation and study of behavioural phenotypes—not physicians but parent support groups. As parent support groups have blossomed to advocate and raise awareness of major issues, such as the dementia of Down syndrome⁵, the movement disorder of Rett syndrome⁶ and the over-eating of Prader–Willi syndrome⁷, a notable phenomenon has occurred. Over the course of their meetings and discussions, participating families have found that they have been describing to each other similar sets of behavioural problems, and that these problems are often unfamiliar to the clinicians with whom the families have been dealing⁸. This in turn has led many of these groups to take the initiative in promoting research to explore the validity of their observations. Lately, family and parental observation and pressure have been pivotal in elaborating the phenotypes. But claims and counterclaims continue to bedevil the subject.

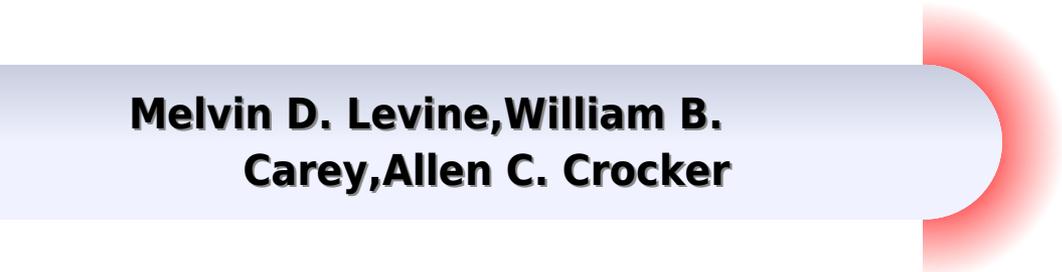
CONTROVERSIES IN BEHAVIOURAL PHENOTYPES

Let me illustrate the controversies by reference to two major causal syndromes of learning disability—XYY

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Behavioural Phenotypes

**Melvin D. Levine, William B.
Carey, Allen C. Crocker**



Behavioural Phenotypes:

Behavioural Phenotypes in Clinical Practice Gregory O'Brien, 2002-01-21 Clinics in Developmental Medicine No 157 Clinicians educators and other specialists who work with young people with intellectual disabilities are increasingly aware of the extent to which their clients behaviours are shaped by the respective causal syndrome This book is a practical response to the need for interventions and ongoing care programmes to take account of this within the context of coordinated multimodal case planning An international team of experts drawn from child health special education psychology psychiatry and related disciplines explores general principles of case management in addition to giving consideration to a large number of individual syndromes resulting in a comprehensive review of the subject All of the authors have been involved in original research on the themes explored and in the development of coherent service responses to the challenges posed by behavioural phenotypes This will be essential reading for all professionals engaged in the care and management of people with intellectual disabilities

Behavioural Phenotypes Gregory O'Brien, William Yule, 1995 Increasing interest over recent years in the study of the influences of environment and genetic factors on behavioural disorder has come from a wide range of disciplines These studies have subsequently been focused through the foundation of the Society for the Study of Behavioural Phenotypes which forms the basis for assimilating new information and coordinating future research in this field This volume from founder members of the society presents a distillation of thinking and reviews appropriate measurement schedules Including research findings explanation of concepts genetic scientific techniques and methodological issues this work will be welcomed by those with an interest in behavioural disorder at every level

Developmental Disability and Behaviour Christopher Gillberg, Gregory O'Brien, 2000-01-18 This book reviews clinical models for working with developmental disability and behavioural problems The first section explores the causes and nature of behaviour problems amongst people with physical learning language and sensory disabilities and some key specific conditions The second section describes the assessment approaches that clinicians will find most useful in evaluating behaviour The third section covers treatment approaches emphasising the importance of a broad eclectic approach The contributors all acknowledged experts in their fields of paediatrics psychology and psychiatry provide a comprehensive overview of this set of major challenges emphasising the importance of auditory detection understanding measurement and treatment

Child Psychology and Psychiatry David H. Skuse, 2003-09

Measuring Behaviour in Developmental Disability, 2001

Readings in Animal Behavior Thomas E. McGill, 1973

Developmental Medicine and Child Neurology, 2000

Genetic Approaches to Behaviour, 1986

Behavioural Ecology Etienne Danchin, Luc-Alain Giraldeau, Frank Cézilly, 2008-02-14 Behavioural Ecology gives a fresh contemporary account of the evolutionary and ecological processes that underpin animal behaviour Contributions from subject experts and meticulous editing yield a text with all the qualities of a multi author book but without the potential drawbacks

Behavioural Biology Abstracts, 1980

The Behaviour of Domestic Animals

Elsayed Saad Eldin Hafez,1975 **Handbook of Aggressive Behavior Research** Caitriona Quin,Scott Tawse,2009

Aggression is a complex social behaviour with multiple causes In psychology as well as other social and behavioural sciences aggression refers to behaviour between members of the same species that is intended to cause pain or harm Aggression takes a variety of forms among humans and can be physical mental or verbal Aggression should not be confused with assertiveness however although the terms are often used interchangeably There are two broad categories of aggression These include hostile affective or retaliatory aggression and instrumental predatory or goal oriented aggression Empirical research indicates that there is a critical difference between the two both psychologically and physiologically Some research indicates that people with tendencies toward affective aggression have lower IQs than those with tendencies toward predatory aggression If only considering physical aggression males tend to be more aggressive than females This new book gathers the latest research from around the world in this field *Developmental Neuropsychiatry: Assessment, diagnosis, and treatment of development disorders* James C. Harris,1995 James Harris s two volume work on developmental neuropsychiatry sets the agenda for this emerging clinical specialty Written by an individual with the developmental expertise of a pediatrician the behavioral sophistication of an adult and child psychiatrist and a deep appreciation of neuroscience these two books offer an integrated yet comprehensive approach to developmental neuropsychiatry Grounded in neuroscience but enriched by clinical realities Volume II provides a comprehensive review of the developmental neuropsychiatric disorders Throughout the text current DSM IV diagnostic criteria are provided Part I outlines the diagnostic process and the genetic history provides details on the conduct of neuropsychological testing and offers a detailed review of brain imaging techniques moving from CT and MRI scanning to the most recent developments in functional MRI and PET scanning Part II discusses mental retardation cerebral palsy the learning disorders the pervasive developmental disorders and traumatic brain injury Part III describes behavioral phenotypes in cytogenetic and other genetic disorders genetic metabolic disorders and disorders that result from gestational substance abuse Part IV is devoted to developmental psychopathology and includes Attention Deficit Hyperactivity disorder schizophrenia Tourette s disorder sleep disorders and the syndromes of aggression and self injury primarily occurring in mentally retarded persons Part V covers treatment and includes detailed descriptions of psychotherapy behavior therapy pharmacological interventions genetic counseling and gene therapy Finally Part VI deals with legal and ethical issues as they pertain to developmentally disabled persons *Physiology and Ecology Japan* Peter J. Jarman,Andrew Rossiter,1994 *Annals of the Academy of Medicine, Singapore Academy of Medicine (Singapore)*,1995 **Proceedings** ,2009 **American Journal of Mental Retardation** ,2008 Includes the association s conference proceedings and addresses **Coenoses** ,1996 **Developmental-behavioral Pediatrics** Melvin D. Levine,William B. Carey,Allen C. Crocker,1999 Completely updated and revised the 3rd Edition of this respected resource offers specific guidelines for the diagnosis and treatment of the full spectrum of problematic behaviors Expert authorities

including many new contributors emphasize formulated reasoning and organized responses to developmental behavioral issues as presented in a clinical environment Offers recommendations for evaluating symptoms and specific suggestions for counseling medical treatment and follow up Many photographs charts and line drawings have been added to enhance this edition

Memory in Animals and Humans Andrew Richard Mayes, 1983

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