

Behavioural phenotypes

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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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