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Behavioural Phenotypes Study Group Symposium*

T. P. BERNEY, Consultant in the Child Psychiatry of Handicap, Prudhoe Hospital, Prudhoe, Northumberland NE42 5NT

Apart from the degree of mental handicap, the traditional description of a genetic syndrome is limited to somatic characteristics, although occasionally noting some quirk of personality or behaviour. However, these subjective stereotypes become a memorable part of our concept of the syndrome. Although at times startlingly perceptive, more often they are misleading, particularly where there has been only a superficial contact. Latterly more systematic scrutiny, albeit using idiosyncratic rating scales, selected populations, and differing diagnostic standards, has highlighted earlier inadequacies. Consequently the last three years have seen the formation of an informal, multidisciplinary group interested in behavioural phenotypes: an initiative greatly helped by contact with parent groups. Their intention is to develop research and, by introducing some consistency to the observations, to allow comparisons across studies. From this has grown the more public and formal Society for the Study of Behavioural Phenotypes† which was launched at this symposium.

The meeting began by reviewing descriptive studies for a wide variety of syndromes. A large amount of information was presented in a breathless series of brief lectures. This was made possible by the small size of the meeting (there were less than 50 participants), efficient organisation, the use of extended abstracts and a favourable venue.

The emphasis quickly turned to the mechanisms which might link genotype and phenotype. For example, why should there be such variation in phenotypic expression for a given genotype? As suggested in other areas of developmental disturbance, the timing of events might be all-important, with critical windows occurring in the course of foetal development (Susan Huson). On the other hand, how do we explain the narrow range of features, notably those of autism, associated with a wide range of aetiologies. This may be the result of a dual restriction on the range of normal variation; first, the general limitations imposed by the mental handicap and, second, the more specific ones (such as a diminution in 'theory of mind') which accompany certain syndromes (Christopher Gillberg).

Is a behavioural phenotype the product of a given syndrome or is it the result of other factors held in common? For example is autism a feature of tuberous sclerosis or does the link merely reflect their joint association with the combination of mental handicap and severe epilepsy (Jennifer Dennis, Anne Hunt and Paolo Curatolo)? Similarly does congenital blindness give rise to autism (and if so, how) or do they share a common cause (Robert Goodman)?

Where a link exists, how specific and inevitable is it? Self-injury is a disorder which is probably based on innate behaviours, inappropriately revealed by a neurotransmitter disturbance, but then governed and shaped by environmental factors. The balance of these contributions is likely to vary and may be reflected in the phenotype: the predominantly innate perhaps being characterised by a preference for imposed restraint, the environmental by a more ready response to behavioural management (Chris Oliver).

The nature and varied aetiology of autism was a prominent theme throughout. The concept now combines a primary disorder with a secondary syndrome embracing a wide variety of aetiologies: the reality of secondary autism was affirmed by the description of a post-encephalitic case (Christina Gillberg). There was debate about the diagnostic lumping of subjects into categories and subtypes. Alternatively subjects might be characterised by the dimension and balance of their various symptoms; classified by autistic feature rather than by autism. The outcome, particularly when based on the symptom of sociability, might only be to gerrymander a fresh group of autisms. However, a more precise description of symptoms previously obscured by too coarse a labelling, may yet reveal the grail; behavioural clusters characteristic of differing aetiologies (Ken Aitken, Christopher Gillberg).

The meeting was thought-provoking and sufficiently fruitful for the Society to hold a similar event next year. Together with the Durham conferences on autism, which follow a similar format, it should engender and guide a substantial body of research.

A report of the conference held in Welshpool on 22–24 November 1990.

†Further information about the society may be obtained from the secretary, Dr Jennifer Dennis, The Park Hospital for Children, Headington, Oxford OX3 7LQ.