Re-thinking the brain: a frog leaps

The notion that one was, when one had completed one's studies (first the biological sciences and then medicine itself), going to be able to understand how people thought and how they behaved, was undoubtedly one of the factors that kept me struggling on with the load of medicine. And somehow at that time the mystique of my teachers and seniors suggested that they did indeed have that knowledge. Also of course at the simplest level it was clear that the nervous system could be stimulated and that stimulation of a nerve in the frog did cause contraction of the muscle. It seemed that people knew how frogs leapt. When I asked my distinguished tutor what I should read about thinking and behaviour in the human from the neurophysiological point of view he recommended Eccles' Neurophysiological Basis of Mind¹ – a struggle. More realistically, one was being encouraged to read the works of Sherrington, and elegant they were to read but somewhat disappointing in a way. What remained in one's mind was how reflexes structured certain fairly simple motor activities of the body, but it seemed a long way off from any real understanding neuropathologically of the processes of learning and control.

How naive this was rapidly became clear when one actually started clinical work in a defined area, such as that of neurodisability in childhood, and one became conscious of the extraordinary limitations of the neurological background which would allow one to explain, understand and, most importantly, to ameliorate the difficulties one saw in children.

Looking at domains of behaviour one quickly realized that the motor domain was in many ways the easiest to study because its output side was reasonably easy to measure. One noted too, when one started medical training, that the markers of disability were physical (height or facies) and less commonly neurological. Hypotonicity was the only 'neurological marker' in Down syndrome. There were difficulties in trying to develop other markers which extended to the individual's behaviour and cognitive function. The demeaning label of the 'epileptic personality' had been discarded about the time I heard of it. This danger of stigmatization meant that people were cautious in identifying personality characteristics which were regularly occurring features of a condition.

It is not perhaps surprising therefore that the field of behavioural phenotypes was initiated with the identification of the extreme behaviour in Lesch Nyhan syndrome, ascribed to one initially bio-chemically identified syndrome, with its genetic features described later. This preamble is to defend the Press' continued interest in the field of behavioural phenotypes. The danger of stigmatization is not forgotten. There are 'cons' as well as 'pros' in this field, all fully described by O'Brien in the new book *Behavioural Phenotypes in Clinical Practice*.² It is thirty years since Nyhan coined the term behavioural phenotype – one is now able to produce a book on clinical approaches to treatment.

The meeting of the Society of the Study of Behavioural Phenotypes (SSBP) in Whistler, Canada in November nevertheless returns to basics under the heading 'Re-thinking the Brain'. In order to make any analysis which looks to a genetic basis one has to question the accuracy and effectiveness of measurements, in all aspects of neurological functioning and one needs some understanding of key signaling events. Indeed, such analysis needs a lot more understanding (see www.ssbp.co.uk). From a research point of view, the field has had some very important spin-offs from approaches in the study of behavioural phenotypes. One is that to get accurate taxonomies of behaviours one must have a close working relationship with the parents. That relationship is now firmly established. The second obvious one is that the detailed analysis of behaviour and its infrastructures, such as the studies of the biochemical pathway between the gene and behaviour, opens up our understanding of the mechanisms that underlie behaviour. Some of these phenotypes are quite unusual, for example, the hyperacusis in Williams syndrome. Knowledge of such mechanisms enriches our understanding of how, for example, sound is analyzed and responded to by the brain, and thus it leads to understanding of normal functioning.

We are shortly to issue a new edition of Gage's classic work on gait analysis, leading to a real diagnostic approach to the motor problems of the child and to some real advances in management. Similar advances in the O'Brien text suggest it is actually worth trying to find out how a frog leaps.

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References

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- 2. O'Brien G, editor. (2002) Behavioural Phenotypes in Clinical Practice. Clinics in Developmental Medicine No. 157. London: Mac Keith Press