The human genome, the disabled person. What's in it for me?

Editorial

No piece of biological research has attracted such widespread media attention as the news that the Human Genome Project is complete (although as I understand it, it is not quite finished). Nevertheless, the heads of state of the USA and the UK appeared on television to proclaim the importance of the event, mutually admiring the contribution that scientists all around the world, and particularly in their own countries, had made to the project. I seem to remember the unravelling of the chromosome structure by Watson–Crick attracted some press coverage, but if my memory serves me right, the appearance of their paper preceded any hype. Darwin would surely have been pleased when he received his copy of 'On The Origin of The Species' but I doubt if any reporters started calling on him until Bishop Wilberforce launched his famous attack. Darwin's peace of mind would have been rudely shattered by today's media attention.

The news is that the exploitation of the logging of the human genome is likely to be in the hands of commercial companies whose main aim will be to make money from the products they will provide to alleviate a condition. With profit as their incentive, they are likely to make products for more common disorders. Therefore those with congenital disorders, which may or may not have a genetic basis, are likely to be at the bottom of the pile, particularly if their condition is rare. The average parent and young person with a disability may well have read or heard all this information with the sinking feeling that, in the short term at least, little benefit from this dramatic piece of work is going to come to them.

I was amused to be told recently that doctors who fail to follow the latest in genetic research are likely to become second-class citizens. That is as it may be, but it is also the case that the genetic researcher who fails to keep up with the latest developments in neurodisability is equally likely to become out of touch with information which may be vital to him/her as a genetic researcher.

Bearing this in mind, it is worth drawing attention to the remarkable importance of Nyhan's discovery of the behavioural phenotype, which he developed out of his study of children with Lesch–Nyhan disease and their strikingly characteristic pattern of self-mutilation. The importance of this discovery was not only that it was one of the first to link a clear genetic syndrome, now known to be encoded by a single gene on the X chromosome (mapped to Xq26–27), but also because it suggested that it would be profitable to look at simpler levels of behaviour than had previously been examined by behavioural geneticists. For example, there are lots of studies of intelligence which attempt to sort general from specific factors. The notion that finger-biting might be a genetic behaviour was indeed novel. It is interesting how long a latency there was between that study and the beginning, not really until the eighties, of substantive work looking for 'a characteristic pattern of motor, cognitive, linguistic, and social abnormalities which is consistently associated with a biological disorder'. Such work has yielded most interesting observations of behaviours which are so associated, for example, the hyperacusis or hypersensitivity to particular sounds found in Williams syndrome [2]; the unusual greeting behaviours consistently recorded in children with fragile X syndrome [3]; or the extraordinary pattern of feeding behaviour in Prader-Willi syndrome, with early failure to thrive and difficulty in feeding, leading on later to 'relentless foraging, stealing, and hoarding of food'[4].

In my view, the importance of these observations is not only that behaviours are found in specific syndromes which are frequently rare, but also the future possibility of looking at a genetic component in a whole range of behaviours in less severely damaged children who may have a near normal physical phenotype. In this way, an understanding of some of their psychological and behavioural characteristics may be gained by investigating aspects of their genetic make up. Another feature of these studies is how the type of human behaviour seen to have a genetic basis is similar to other species: both Conrad Lorenz and Niko Tinbergen found systematic patterns of behaviour in greeting, feeding, mating, etc. in the animals and birds they examined.

These thoughts and speculations on the genetics of human behaviour may seem a far cry for the parent looking for help for their child. But it does suggest that we are now in a position to ask the geneticists to look for a relationship between a difficulty we perceive in a child and a gene, and then ask our commercial colleagues to begin to develop possible interventions to help children with a vast range of disorders. As I said at the outset, it is unlikely that the big commercial producers will initially be interested in these topics and, as I see it, parents once again will need to battle for the rights for their children to have effective interventions.

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https://doi.org/10.1017/S0012162200000967 Published online by Cambridge University Press