

Learning from less common disorders

In my less experienced paediatric days, I had some distaste for my colleagues who seemed to enjoy collecting rare disorders, rather like stamps. 'I've got a case of X's syndrome on the ward' they would announce with glee, and I was taken to see a child with very lowly abilities and with often some quite distressing physical markers. I am grateful to one psychological research colleague for pointing out to me how insensitive she found the presentation in medical textbooks of this type of child. The photo was often taken of the child standing naked against the clinic wall with a caption drawing attention to all sorts of physical hazards the child was heir to. Rightly she insisted that, while we did use a clinical photo of the child, they were always presented as a human being first, and often in the arms of a parent, and displaying all their features and not just the damning ones the doctors sought.

A glance at any textbook of children's diseases going back more than 50 years often gives examples of this unfeeling approach. Foolishly, to some extent, I allowed this distaste to lead me to ignore what was being found out about the rare disorders, and it was only when I was carrying out an epidemiological study on young adults with physical disabilities¹ that I realized that within this limited group of the physical disorders, rare disorders were 'common', that is to say that when we looked at our samples although there were relatively large numbers of children with the common physical disorders (e.g. cerebral palsy, spina bifida) they were outnumbered by the number of children who had a single diagnosis. They deserved help and attention. But until the genetics developed for an increased number of these diagnoses, we made, perhaps, slow progress beyond the dysmorphic features recorded by physicians and observed in those photographs.

Now the ability to collect, often with the help and interest of the parents, a largish group of children with a rare disorder, hopefully we can learn to provide better service for those children, and we certainly learn a lot more about the nature of disease processes which we can apply much more widely. Now with well defined groups of patients, good detail of developing functions, both normal and abnormal, becomes available, and where there are abnormalities, so often the findings immediately become relevant to a much wider number of patients. The studies of the satiety problems of children with Prader Willi have given one much to think about in relation to many other children who have difficulties with feeding. Cass et al.² in this issue provide us with a host of material about Rett syndrome, building of course on the earlier work of Andreas Rett himself, Hagberg, and Witt Engerström, to mention but a few.

A first thought that comes in thinking about Rett syndrome is that it is not a degenerative disorder but it is a developmental disorder which shows a changing course in childhood and it shows changes in different functions. One is immediately reminded of the definition of the far commoner cerebral palsy, a

syndrome we still struggle to sort out, but where we talk about the non progressive but not unchanging nature of the condition. In Rett syndrome the developmental course is different, indeed unique, but again gives one great cause for thought, as one notes the changes that go on in the abnormal hand function from the greater attention to the movements towards the mouth, to be replaced by the hand wringing to be replaced by the acquisition of some, in a proportion, of purposeful, useful movements. An extended and different pattern to what one would see in cerebral palsy, obviously, but it makes one wonder whether it is simple motor tract difficulties which cause the problems in both these conditions. One can equally look at the pattern of disorder in the gross motor function and in the development of contractures and back problems and note again the 'not unchanging pattern' in the disease.

Another aspect of great interest is how the variables studied interact. As with other conditions (e.g. tuberous sclerosis), epilepsy is associated with worse scores in a number of variables such as breathing abnormalities and drooling. But in addition, most interestingly, it is associated with low height and weight, an affect presumably manifest before the active onset of epilepsy, often around school age. The possible mechanism of this is clearly a challenge to research.

Another area not perhaps so fully developed in this paper is the change in what one might describe as the functional factors: the distress, screaming, and sleep disturbance early on which later, in adult life, may change to laughter. Each specific area which one reviews, informs one to think about those activities and try to see how the pathology in Rett syndrome perhaps links up to and parallels that in other conditions. Time and more study of the whole genetic pathway from gene to function and behaviour will help us with that question. But for the time being, as Cass and her colleagues conclude, the study provides 'an increased delineation of the phenotypic characteristics of Rett syndrome' which will 'contribute to our clinical understanding of the complex neurodevelopmental condition.' That clinical understanding should lead us on to being able to begin to intervene effectively and improve the quality of life not only for the young women who have this condition but also for those with other 'rare disorders'.

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References

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