

Comorbidities

Searching through medical dictionaries surprisingly revealed very few definitions of comorbidities until very recently. The word was suddenly thrown on us in medicine in the last 10 years. I did find a dictionary with 'comorbidity' in it and it said, fairly concisely, that the term pertains to 'a disease or other pathological process that occurs simultaneously with another'. That's 'comorbid' and 'comorbidity', the extent to which two diseases occur together in the same population. The word 'comorbid' seems with us to stay. Indeed in this number Steffenburg et al. produce a paper entitled 'Autism spectrum disorders in children with active epilepsy and learning disability: comorbidity, pre- and perinatal background, and seizure characteristics'. It is a good paper about, as it says, autism, epilepsy, and learning disability; but why the comorbidity in the title, and should the editor of 'Developmental Medicine and Child Neurology' have allowed it to stay there?

We look at usual clinical practice which describes processes and events in the past: premature birth, seizures etc. Symptoms, which as the history implies, change over time. Signs are what we find and see. More complicated perhaps than what the classical neurologist thinks of as physical signs, are behavioural signs which may prove to be specific to certain diseases, or at least more common with certain diseases.

The whole field of behavioural phenotypes has opened up this notion. But bear in mind that initially we thought of an exclusivity of certain symptoms to specific phenotypes: of self-mutilation in Lesch-Nyhan syndrome, overeating and obesity in Prader-Willi, disturbed greeting behaviour in fragile X and so on. But as the field has developed we now know many of these behaviours occur not simply in one condition but in several. The triad of autism is social behaviour, speech-language difficulties, and unusual behaviours. These are all a signs, aren't they? (Or are they behaviours?) Then we think of aetiologies we know of, such as genetic, asphyxial, traumatic and maldevelopment. How often in neurodisability are there actual clear-cut aetiologies? Add infective causes to the list, with diseases like Rubella, a clear-cut disease in most people but complicated patterns if they occur in fetuses and infants. We see how complicated it is when one or another aspect of the clinical findings is mixed with aetiological postulates to produce 'comorbidity'. With learning disability we can start thinking about environmental factors as well as genetic, and lesional factors associated with damage to the foetus or infant, the implication is that there are three comorbidities: genetic, lesional, and environmental. Does the environment actually cause brain pathology?

I found shortly after Rett syndrome was described, my first case of it in a special school which I was visiting when I observed the hand behaviour. Noted too that the child was toe-walking with tight heel cords. I was interested to learn that the child had up until that point been diagnosed with cerebral palsy. Should the tightness of the heel cords (cerebral

palsy) and the Rett syndrome constitute a comorbidity?

Gillberg and Coleman in their *Biology of the Autistic Syndromes*,¹ list 27 conditions where you see autism as a comorbidity. They list mucopolysaccharides as one condition. They talk about the tuberous sclerosis complex. Each of us who has detailed knowledge of one of those syndromes can gloss the diagnosis. All these would pass muster on some autistic diagnostic questionnaire. Gillberg and Coleman also list some toxic syndromes: foetal alcohol, foetal valproate, lead poisoning, and some infectious syndromes. They list cerebral palsy as a disease entity; in all instances implying that two diseases are occurring together.

Steffenburg, in previous papers, has found autism in an epileptic clinic unbeknownst to the neurologists who were running it. Psychiatrists have been caught short when a neurologist has indicated to them that a child with autism also has epilepsy. One could equally think of us professionals failing to take proper histories, proper accounts of behaviours, proper study of the signs, a proper neurological exam, and whether these professional inadequacies don't lie behind use of the term 'comorbid'.

But there are real difficulties: what about the 'blind' child who you may begin thinking (as I have) has 'blindisms' associated with some delayed communication and surely some difficult social interaction with his visual loss; but in whom, some two or three years later, you're finding that you now recognise a diagnosis of autism. What about the boy with cerebral palsy you didn't notice for some time because you're concentrating on his motor disorder, who responds to you in an odd way? In these conditions do you think of genetic factors causing the autism? Do you think the lesional aspect of cerebral palsy has produced a pathology in the brain from the same asphyxial event and has damaged that part of the brain which is genetically damaged in 'pure' autism?

These deliberate ramblings will hopefully arouse your interest. Be careful when you use the word comorbid. Be careful to list out history, symptoms, signs and all these phenomena that arise. Start from a general term, 'neurodevelopmental disorder', and after your investigations, decide what pathologies you really are dealing with. The Castang Trust, in order to help with these issues, funded last year a workshop on comorbidities and next year we hope to produce some of their deliberations in a book on the topic. By that time, you will have worked it all out for yourself.

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Reference

- Gillberg C, Coleman M. (2000) *The Biology of the Autistic Syndromes. Clinics in Developmental Medicine. No153/4*. London: Mac Keith Press.