Commentary

Health care professionals, for long accustomed to thinking in terms of pathological entities rather than functional significance, have had difficulty in grasping the full implications of the distinctions between impairment, disability and handicap, as defined by the WHO. Even now, nearly 20 years after this classification was introduced, the terms are still frequently misused. It could be argued that we should aim for better understanding and more appropriate usage of the existing terms, rather than redefining them; but there are several reasons why change is needed.

Firstly, the international disability movements have generally rejected the WHO definitions in favour of two basic concepts related to the social model of disability: 'impairment' meaning the loss or abnormality plus the effect on function; and 'disability/handicap', the disadvantage or restriction of activity caused by social factors which take little or no account of people who have impairments and thus exclude them from the mainstream of social activities.[1] This simpler classification has the additional advantage of being easier to translate for non-English speakers, as few other languages recognise the subtle distinction between disability and handicap. Furthermore, we should refer to 'disabled people', not to 'the disabled', or 'the handicapped'. The latter terms are offensive because they classify people on the basis of a single characteristic and do not acknowledge their individuality.

Is this another example of 'political correctness'? Perhaps – but being PC is no bad thing. By adopting PC terminology we not only acknowledge that there may be some justification for the new terms, we also help to bring about change. Phrases which our generation regards as 'PC' and uses tongue in cheek could become the accepted language and attitude of the next generation.

Articulate people with physical disabilities rightly point out that they are disadvantaged by the attitudes of society more than by their loss of function – a view elegantly presented in the advertising campaigns of the Spastics Society, now known as Scope ('our biggest handicap is poor communication – yours'). The weaknesses of this 'social model' of disability are all too obvious to the parent or carer whose life is devoted to providing the total care needed by their profoundly multiply disabled adult offspring. Hutchison is right to try and bridge the gap between these differing perspectives.

Secondly, better classification, definition, and registration of impairments would have many scientific and practical benefits. Although the terminology used in the OPCS survey probably came closer to the ideal than any other attempt so far, its application in routine service has proved difficult. Ways of measuring quality of life would be helpful[2] and might make registration more acceptable. At present, some people actively avoid being registered, because they do not feel disabled and do not wish to be defined as such – whatever the professionals might think.

Thirdly, a change in terminology might facilitate provision of services for people with disabilities, without having to apply the dreadful 1948 definitions of disability. The dilemma is a real one. A person has an impairment which, without equipment and resources for better living conditions, will render him disabled. In order to minimise his disability, he must first submit to being classified as disabled. Surely we can do better than this.

We should accept the challenge to re-think our attitudes and our terminology, but we must learn the lessons of the past 20 years and make sure the new terms are acceptable and understandable. Change must be driven by the real needs of disabled people. Complex human problems must not be forced into the crude and often arbitrary categories demanded by our still primitive computer software.

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1 Coleridge P. *Disability, liberation and development.* Oxford: Oxfam Publications, 1993: 100–1.
2 Lindström B. Quality of life for children and disabled children based on health as a resource concept. *J Epidemiol Community Health* 1994; **48:** 529–30.

Investigation of mitochondrial disease

Mitochondrial diseases are now recognised as a significant group of neurometabolic disorders in childhood[1] and may be caused by mutations in mitochondrial DNA (mtDNA). The major energy generating reactions in a cell are concentrated in the mitochondrion where the pathways of carbohydrate, fatty acid, and amino acid oxidation converge and feed into the tricarboxylic acid cycle and thence the electron transport chain. This review will use the term 'mitochondrial disorder' to refer to diseases of the electron transport chain and associated central pathways, many of which are associated with lactic acidosis and/or caused by mtDNA mutations.

Diagnosis of mitochondrial diseases is frequently difficult because of the wide variety of clinical presentations which are possible particularly in the paediatric age range. While there have been major advances in recent years, a molecular diagnosis is considerably less likely in paediatric than in adult practice. In this review we will first discuss the general investigation of these patients. We will then introduce the basic biology of mtDNA in order to classify mtDNA diseases by mode of inheritance (table 1) and the specific investigation of some of the commoner clinical syndromes (table 2).

Which patients warrant investigation?

Patients may present with a recognised clinical syndrome or a suggestive constellation of symptoms. Mitochondrial studies are clearly indicated in patients with Leber's hereditary optic neuropathy (LHON) (in which patients present in adolescence with acute loss of vision),[2–4] Kearns-Sayre syndrome (in which patients present with proximal myopathy, chronic progressive external ophthalmoplegia, and retinopathy), mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes (MELAS)[5 6] and myoclonic epilepsy and ragged red fibres (MERRF)[7] (table 2). Because mtDNA diseases

Table 1 Mode of inheritance of mtDNA disease

1.	Sporadic	Major rearrangements (Kearns-Sayre syndrome, CPEO, Pearson's)
2.	Mitochondrial	Point mutations (MELAS, MERRF, NARP, LHON, aminoglycoside induced deafness) Familial duplications
3.	Autosomal dominant	Variable deletions
4.	Autosomal recessive	Depletion Chaperonin deficiency
5.	X linked	?Susceptibility in LHON

CPEO=chronic progressive external ophthalmoplegia.