Chapter 1
Terms, definitions and concepts
Colin Kennedy

Key messages
- The precautionary principle, first do no harm, was established 2500 years. Justification for the use of a treatment remains the responsibility of the treating physician.
- For many centuries ‘ecologies of care’ rather than definition of illness was the predominant paradigm of medical practice and this continues to be relevant to the young because the relationships between a child, the family and the wider environment remain important determinants of health outcomes, especially in infancy.
- Discussion of management of disease is greatly facilitated by internationally agreed definitions of disease and these are available as the ICD-10 (www.who.int/classifications/apps/icd/icd10online).
- Evidence-based medicine provides an objective method for the systematic evaluation of the evidence of benefit and harm of medical interventions.

Common errors
- Use of imprecise terms for which international agreement is lacking, for example raised intracranial pressure, hydrocephalus syndrome, myotonic syndrome, hyperexcitability syndrome.
- Imprecise or incorrect use of terms for which precise definitions exist, for example perinatal encephalopathy, epilepsy.
- Generalization of uncommon conditions to common clinical situations, for example attributing trembling of the chin, or feeding problems, or excessive crying, or febrile seizures to neurological disorders.
Part 1: Principles of child neurology in infancy

When to worry
- Separation of infants from their families (one should facilitate bonding between an infant and their main carer).
- Use of poorly evaluated, potentially harmful, interventions in many infants to treat rare neurological problems (one should use common sense, and look at international recommendations of good practice).
- Resistance to evaluation of the benefits and harms of current treatments (one should consider all interventions for potential benefits and risks).

The basis of medical practice
This book offers knowledge, only some of which is truly evidence-based, and a framework for incorporating evidence into the clinical care of infants in whom there is concern about neurological function or developmental progress. Historically, the starting point was myth and wise myths will continue to have their place in medical practice. According to the ancient Greeks, Apollo was the god of healing and Asclepius, his son, was rescued by Apollo from the womb of his dying human mother, Coronis. Asclepius’ daughters were Hygeia, the goddess of health, and Panacea, the goddess of cures. Asclepius also had sons and Hippocrates, according to myth, was a descendant of one of those sons. Hippocrates was a practising physician nearly 2500 years ago and author of the Hippocratic Oath (www.pbs.org/wgbh/nova/body/hippocratic-oath-today.html), the most famous text in Western medicine.

The most widely quoted section of that oath states: ‘I will use treatments for the benefit of the ill in accordance with my ability and my judgement, but from what is to their harm and injustice I will keep them.’ In addition to this statement of the precautionary principle (i.e. ‘first do no harm’), other sections of the oath bind the practitioner to resist all temptations that their privileged position as physician offers, to acknowledge the limits of their competence and refer to specialist practitioners when necessary, to leave surgery to the surgeons, to respect patient confidentiality, to treat one’s professional teachers as one’s parents and to pass on the art of medicine to the next generation. Thus many of the issues of key importance to clinical practitioners and the health systems within which they work are identified within the oath.

Precautionary principles in the context of neurological problems in infancy
The precautionary principle is especially relevant in the assessment and management of neurological and neurodevelopmental problems in infancy when medical intervention may unwittingly hinder the role of the parents in the development, whether typical or impaired, of the child: hospitalization or other institutionalization should be avoided whenever possible (Chapters 2 and 4). Any system of medical activity that involves
surveillance of typically developing children should be based on explicit principles of screening (Chapter 6), including evidence that benefit of early intervention, whether special investigation (Chapters 7–9) or treatment (Chapters 10 and 11), outweighs the potential for harm. The range of ‘normal’, better termed ‘typical’, neurological development in infancy is broad. In cases of doubt, continuing clinical surveillance and support for normal parenting is needed. This has less potential for harm than enthusiastic separation into medical categories in the border zones of normality and the use of treatments for which benefit is not established or is outweighed by risk of harm. Any system of practice that categorizes more than a few per cent of infants as neurologically abnormal must itself be suspect. Such a system is incompatible with the epidemiology of neurological disorders in childhood and will, by definition, expose many normal children to the risk of being wrongly categorized as impaired. This is, in effect, a particular example of the need for screening, in this case secondary screening, to fulfil a number of criteria additional to those that apply to the treatment of illness (Chapter 6).

Neurological and developmental assessment of the infant is a practical skill of central importance (Chapter 5) that requires hands-on experience as well as knowledge. The importance of the physiological state of the infant (hungry or fed, wakeful or drowsy, contented or distressed), the need to rely on best performance and the extent to which clinical features are consistent over time are more important to bear in mind at this age than at any other. Almost any finding with respect to the deep tendon reflexes, other than complete absence of them, for example, is within the typical (i.e. normal) range in some physiological states or at some age within the first year.

Ecologies of care and categories of illness
On the foundations expressed by Hippocrates, the art of clinical practice in Western countries evolved in the pre-scientific era using a system of knowledge based on the eminence and experience of senior practitioners. For many centuries prior to the more modern description of categories of illness as the basis of medical practice, ecologies of care for maintenance of health and for the treatment of illness acknowledged the importance of the relationship between the patient and the wider environment and provided the predominant paradigm of care. In the case of the child patient, family relationships are of primary importance and are fundamental to the Head Start (USA) and Sure Start (UK) programmes for the improvement of the health and well-being of young children (Blair and DeBell, 2011). These issues are discussed in Chapters 2, 4 and 6 of this book.

The international classification of diseases
The World Health Organization was founded by international treaty in 1948 as a specialized agency of the United Nations with unique authority to establish global health standards and to secure international agreement on defining disease. The 193 member states of the WHO have agreed to use the International Classification of Diseases (ICD) and the updating of ICD-10 is expected to be completed and lead to
Part 1: Principles of child neurology in infancy

the publication of ICD-11 in 2014. Classification of mental disorders, which include neurological disorders, is complex and controversial both because underlying pathophysiology cannot be observed directly and because many symptoms are continuous with normal phenomena. Nowhere are these issues more relevant than in the neurology and neurodevelopment of infants. The ICD-10 classification is predominantly driven by the clinical utility and public health outcomes of the disease entities and, in spite of these controversies, is therefore an appropriate framework for clinical practice (Reed et al., 2011).

The ICD-10 provides the basis of the discussion for classifying the phenomena observed in the clinical contexts that are discussed in Chapters 12 to 24 of this book. A number of entities (e.g. brain tumours of infancy) are included in the differential diagnosis but not covered in detail in this volume for lack of space. Other entities do not appear because they are based on classifications of disease other than ICD-10. Such diagnostic classifications, including some listed in the paragraphs below, may claim to identify disease entities requiring active management in a substantial percentage of neonates or infants. In some cases, the criteria for making such diagnoses are vague, their relationships with disorders of later childhood unknown and the rationale for intervention is obscure.

International clinical guidelines depend upon this shared nosology and classification of illnesses and disease: the foundation for rational management requires knowledge of what treatments are of benefit and what are harmful which in turn requires specific disease definitions that are shared by all those involved in providing care.

Relevant terms and definitions
The brief discussion of terms below is intended to help the reader to navigate through later chapters of the book but is not intended to be exhaustive.

*Encephalopathy* is defined as ‘a disease in which the functioning of the brain is affected by some agent or condition’ (*New Oxford Dictionary*). Because this definition is so inclusive, it is of little practical value in clinical medicine. While ‘acute’ means ‘of short duration’ or ‘experienced to an intense or severe degree’ (*New Oxford Dictionary*), the medical definition of *acute encephalopathy* includes alteration in conscious level as an essential criterion (Chapter 14) and to that extent is more clinically useful for the formation of a plan of investigation and management.

The term *perinatal encephalopathy* does not indicate either whether the observed effect on the functioning of the brain is of short duration or whether it involves an alteration of conscious level. Furthermore, the infant’s level of consciousness prior to birth is usually not known to the clinician. However the general term encephalopathy is no more useful in the perinatal period than it is generally, especially since there is often disagreement as to whether or not commonly observed neonatal or post-neonatal behaviour (e.g. tremor of the chin) indicates abnormal brain functioning. This carries
important potential for harm in exposing many infants, the vast majority of whom will have no known subsequent medical disorder, to the risks associated with medical diagnoses of doubtful validity. In practice, therefore, the definition of perinatal encephalopathy includes alteration of level of consciousness (Chapter 12). It is, in effect, the special case of ‘acute encephalopathy’ in an individual within the perinatal age group. Other more inclusive uses of the term perinatal encephalopathy are to be avoided and will not be further discussed here.

Hydrocephalus is used to mean an excess of cerebrospinal fluid within the head but excluding those situations where that situation has arisen purely from atrophy or failure to develop of the brain substance (sometimes called hydrocephalus ex vacuo). The presence of hydrocephalus cannot be confirmed or excluded from consideration, in isolation, of the dimensions of the third cerebral ventricle. See Chapter 18 for further discussion.

Hydrocephalus and raised intracranial pressure frequently co-exist and specific clinical signs, often including disturbed consciousness, can be combined with cranial imaging to provide evidence for the presence of both entities (see especially Chapters 14 and 18) but there is no internationally recognized entity of 'hydrocephalus-intracranial-hypertension syndrome'.

A seizure may be epileptic (Chapter 16) or non-epileptic (Chapter 17). Epilepsy is defined as recurrent unprovoked epileptic seizures. Febrile seizures are provoked by a rising fever and are not conventionally regarded as falling within the above definition of an epilepsy. Chapters 16 and 17 provide further discussion of these definitions and of the syndromes that constitute disease entities within them.

Myotonia is defined as the inability of muscle fibres to relax after muscle contraction and can be demonstrated by myotonic discharges on electromyography. This is a very rare phenomenon in infancy and even in an infant with congenital myotonic dystrophy (Chapter 20), myotonia is usually only demonstrable in an affected parent. The term 'myotonic syndrome', in which abnormality of muscle tone is the dominant feature, is not a recognized diagnostic entity in infants.

'Hyperexcitability syndrome' is not a generally accepted diagnostic disease entity term in infancy and internationally accepted criteria for its definition are lacking. Associations between problems with crying, sleeping and/or feeding in infancy and long-term behavioural outcomes in childhood, including attention-deficit-hyperactivity disorder, exist. Meta-analysis suggests that these so-called early regulatory problems can have an adverse effect on behavioural or cognitive development but findings have been inconsistent. The risk is highest in those with multiple regulatory problems in infancy in multiple-risk families. These problems are attributable partly to biological predisposition in the infant, partly to parenting behaviours and partly to interactions between the two. Interventions that alter parenting behaviour are followed by a reduction in regulatory problems (Hemmi et al., 2011).
Part 1: Principles of child neurology in infancy

Twenty per cent of infants show early regulatory problems and their predictive value for behavioural problems later in childhood is low. Most cases may therefore be regarded as falling within the spectrum of typical development and need not be conceptualized as indicating an underlying neurological disorder. In a minority, an underlying psychiatric, neurological or other medical disorder could, of course, present with early regulatory problems. Support to parents to help prevent or reduce early regulatory problems is therefore typically given in the context of general paediatric nursing or medical assessment of the infant and advice to families rather than as treatment of a disease entity.

Evidence-based medicine
The accumulated wisdom of previous generations of medical practitioners, has since the nineteenth century, been progressively supplanted by the concept of ‘evidence’, of greater or lesser quality, to support the use of treatments. Myth has been progressively replaced by evidence although the process has sometimes been hindered by political interference (McKee, 2007). Hopefully the value of certain myths, starting with Hygeia and Panacea, will continue to be recognized.

Evidence-based medicine has only emerged within the second half of the 20th century and has been an increasing influence on medical practice in the 21st century. It is, as described in Chapter 3, the systematic construction of a body of knowledge about interventions for medical illnesses with explicit, objective criteria for rating the quality of the evidence upon which that knowledge is based. The great strength of evidence-based medicine lies in its capacity for constant improvement as new information comes to light and without ad personam arguments about the authority of the individuals advocating any particular treatment, which had dominated previous medical thought since Hippocrates, sometimes referred to as ‘eminence-based medicine’.

Unfortunately, the quality of much of the evidence upon which we must currently rely for guidance in the treatment of neurological disorders in infancy is poor. Furthermore the traditional measures of quality of evidence are sometimes difficult to apply when studying rehabilitation including physiotherapeutic interventions (Rosenbaum, 2010; Autti-Ramo, 2011). The methodology of evidence-based medicine can also help us to identify those situations where evidence is lacking and serve to remind us that justification is always required for medical intervention, especially in an infant, and the responsibility for this rests with the physician.

References
Chapter 1 Terms, definitions and concepts


Resources


ICD-10 codes, www.who.int/classifications/apps/icd/icd10online