Helping parents with the diagnosis of disability

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Abstract

The impact on parents and the wider family of caring for a disabled child depends on many factors, including the child's behaviours, available resources and the families' coping strategies. Optimal care requires careful management of every stage from diagnosis onwards. How the diagnosis of neurodevelopmental disability is communicated to the family has long-lasting effects. When clarification of the diagnosis takes a long time, this is particularly hard for child health professionals to manage well, and a joint understanding of the process with the family is important. This short review offers guidance for the conduct of the diagnostic consultation referring to the current best evidence and drawing from personal experience. It also explains how current theory and evidence can facilitate better understanding of families' reactions to a diagnosis, moving beyond a simple bereavement adjustment model towards a nuanced appreciation of parental reaction.

Keywords Adaptation; clinical decision-making/ethics; continuity of patient care; disabled children; methods; parenting; patient care; psychological; quality of life

Introduction

The overall prevalence of neurodevelopmental disability in childhood is difficult to ascertain. This is due to the variety of ways in which disability has been defined and measured. Further, as subtler problems may only be diagnosed in later childhood the precise rates increase with age. Nonetheless, rates of neurodevelopmental disability are increasing. Part of this increase is a result of improved survival of premature infants, and better medical care. In the UK, in common with many developed healthcare systems, there is a substantial requirement for health services to support children and young people with complex medical and neurodevelopmental needs. In recent decades, both professional and carer understanding of the impact of impairment and subsequent disability has evolved from a largely medical model to an appreciation of the importance of maximizing young people's quality of life and social participation, regardless of body structure and function.

Guidelines regarding how to discuss the diagnosis of neurodevelopmental disability with the family are available; however the child and family journey through recognition of impairment through diagnosis and subsequent adaptation is complex. This review will discuss the factors affecting resolution and coping strategies, in order to offer further guidance for child health clinicians in communicating effectively with families before, during, and after the time of diagnosis.

In 2007, the adapted version of the International Classification of Functioning, Disability and Health for children and youth (ICF-CY) was published, and participation and quality of life in young people with disability are now the focus of multi-national health research, and service delivery. This shift in emphasis to include both a medical and social construct of any diagnosis is the key to ensure optimal professional support for families. Parents and carers look to professionals for accurate information about the characteristics and aetiology of their child's identified impairments. At the same time, their understanding of their child's needs and their desire to achieve a good outcome, allows joint development of the meaning of the child's diagnosis (Figure 1).

Before diagnosis

In infants where there is an immediately recognized medical abnormality or need for intensive care, issues regarding survival and medical intervention are paramount; however, parents may not appreciate the possibility that their infant may survive with long-term impairment. The need to discuss possible outcomes in the neonatal period is not uncommon and where possible the consultant or most senior member of the team should be available. Junior medical staff may, however, have the initial contact with the family, particularly out of hours. Training in generic communication skills and specific scenarios (e.g., discussion of Down syndrome, or impending premature delivery) is an essential component of both undergraduate and postgraduate medical curricula. Nevertheless, adherence to guidelines and best practice in communication skills may not be universal.

A more protracted process of recognition before diagnosis is likely in the case of other developmental disabilities such as cerebral palsy, neurometabolic, neurogenetic, and pervasive developmental disorders. Although concerns may be identified early, there may be a prolonged period of investigation or assessment. With significant recent advances in diagnostic investigations, particularly with genetic investigations (initially with CGH array and now with established clinically available single gene panels for recognised clinical phenotypes, and more detailed research-based techniques such as whole genome and exome sequencing), there may be results of uncertain significance, leading to further anxiety for parents. In many cases, a

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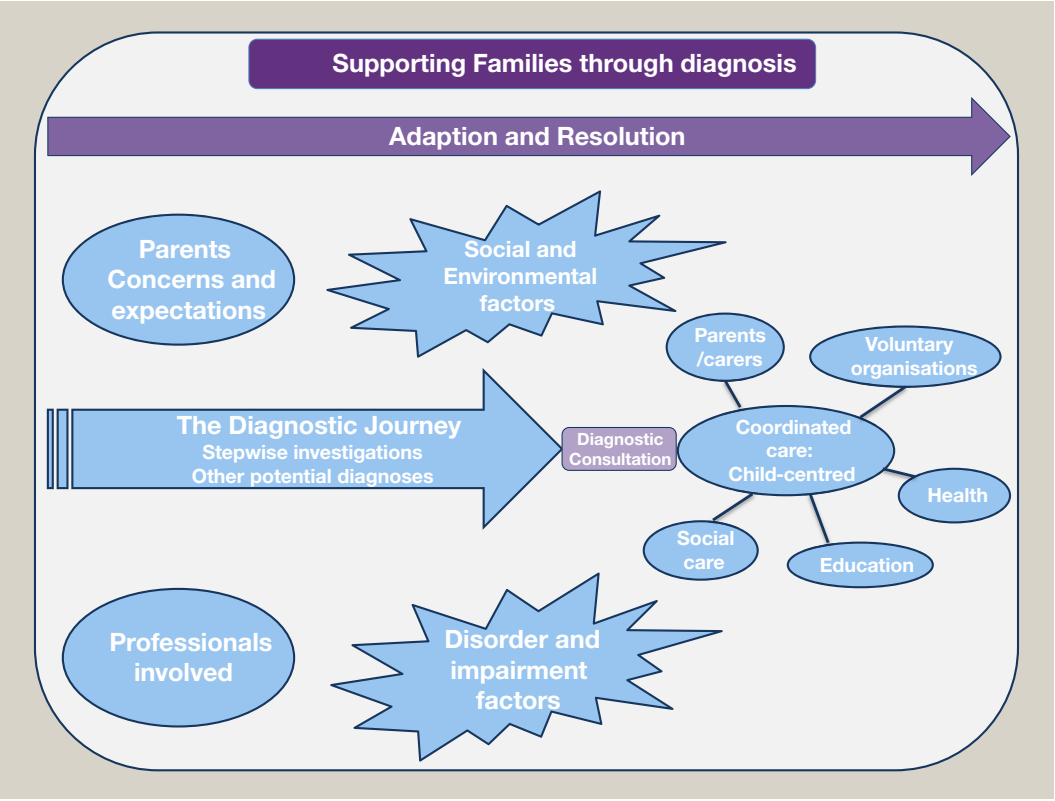

Figure 1 Supporting families through diagnosis.

definitive diagnosis may still remain elusive. In these situations, parental adaptation and reaction to the impairment (and thus opportunities to help parents cope) may have already commenced and there may be significant clinical and social support established with or without a diagnosis. On the other hand, some parents report that they feel isolated in their concerns, or that there are challenges to accessing the support they need (e.g., administrative barriers, service provision issues).

Diagnostic consultation

The adaptation and resolution of a family with a disabled child are dependent on many factors; however, a commonly reported significant factor affecting this is the manner and circumstances of the diagnostic consultation. The diagnostic consultation presents an interesting dynamic: the parent may feel overwhelmed by emotion, and the clinician is primarily focused on the information they are providing. These two perspectives can be difficult to reconcile. Qualitative studies have identified 'see-sawing' in negotiation between carers and professionals, where judgement is made regarding how to use language, and information and prognosis given according to perceived parental reactions. For example, where a parent is despairing the clinician may hold out more hope, whereas when parents are optimistic, the clinician may be more blunt and pessimistic.

How, when and what then?

Studies demonstrate that the empathy and language of the person who disclosed the diagnosis, the arrangements made for the consultation, and subsequent follow up are the key components. Parental dissatisfaction with the diagnostic process include lack of sufficient and understandable information, dismissal of worries, lack of empathy, delay in diagnosis (and the belief that clinicians know before disclosing), lack of opportunity to ask questions, and unapproachable clinicians.

The following guidelines suggest an approach to diagnostic consultation (whether early or later in a child's life), and are based on published evidence from research in health and social sciences and guidance from the Department of Health and other organisations, including parent led organisations (e.g., National Network of Parent Carer Forums, Cerebra). Guidelines provide guidance only and should not be used as a 'rulebook'. Professionals should remember that each child, their family, their cultural and environmental situation are different and adapt their approach accordingly.

Guidelines for diagnostic consultation

Preparatory information

- It is important to know the family dynamics, environment and level of understanding of the child. Wherever possible clinician(s) should access the shared knowledge of the whole multidisciplinary team.
- The clinician(s) who will be disclosing the diagnosis must have appropriate generic training in communication skills.
- Clinician(s) should have full information and confidence in the diagnosis.