SECOND EDITION

NURSING CARE OF CHILDREN AND YOUNG PEOPLE WITH LONG-TERM CONDITIONS



EDITED BY
MANDY BRIMBLE | PETER McNEE

WILEY Blackwell

Nursing Care of Children and Young People with Long-Term Conditions

Second Edition

This book is dedicated to all children and young people with long-term conditions and their families

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Introduction

Mandy Brimble and Peter McNee

Currently, there are limited books available which analyse the context, theory and practice of nursing children and young people with long-term conditions. This second edition provides a comprehensive and fully updated resource for nursing students and post-registration children's nurses on assessing health needs and delivering care and services holistically within and across a variety of care settings in order to meet the changing needs of children and young people with long-term conditions and their families.

Although each chapter can be read independently, the book is designed to provide a comprehensive insight into the changing health care needs of children and young people with long-term conditions and the implications for delivering nursing care and services to children and young people of several age groups, cultural backgrounds, with differing conditions and in a variety of care settings.

In each of the chapters, individualised case studies and reader activities are used to apply theoretical principles and current evidence to nursing practice. In addition, readers are able to gain a greater understanding of the clinical conditions featured in the case studies, both in relation to development issues and associated care needs.

Chapter 1 revisits the aetiology of long-term illness, examining the genetic basis of children and young people's long-term conditions and certain disabilities as a consequence of hereditary influence, providing an overview of chromosomal anomalies and genetic pathways of inheritance. The latter half of this chapter explores the differing onsets of long-term conditions, considering prenatal, neonatal and late onset, and their implications for practitioners and care delivery.

Chapter 2 examines some of the current political, economic and social policies that are shaping the context and service delivery for children and young people with long-term illness, and the issues and challenges these bring to managers, practitioners and service users. Particular points discussed include workforce changes, patient engagement and commissioning. Examples of service models and nursing roles are analysed to apply these issues and challenges to nursing practice and demonstrate the changing boundaries of clinical practice, multidisciplinary working and service delivery.

Chapter 3 provides a theoretical basis for the impact of long-term illness on the child and parents, examining in detail some classic and contemporary theories relating to grief, loss, coping and adaptation. Suggestions are made concerning effective care strategies and practices to support and help parents adapt to their child's diagnosis of long-term illness. A clinical case scenario of a girl with type 1 diabetes is used to apply the key principles outlined in the chapter.

Chapter 4 is new for this second edition and specifically examines the impact of having a sibling with a long term condition. Contemporary thinking on the rights and needs of siblings is addressed together with impact of becoming a carer for a member of your family. A case study is used to examine these issues in relation to a baby with cystic fibrosis.

Chapter 5 explores these issues further by examining the particular care needs of a girl with eczema, focusing on the implications for children, young people and their

families in their adaptation to long-term illness and addressing the practical implications of assessing and meeting their physical, psychological and social needs. Interesting discussions include issues around ethnicity, culture, spirituality, social isolation and the use of complementary therapies.

Chapter 6 provides insights into the general principles for the need to inform, educate and promote health to children and young people with long-term conditions and their families as an effective means of empowering them to be 'experts' in their care. Using an asthma case scenario, challenges that may arise due to the receptiveness of children, young people and their families, or their intellectual or resource ability to change behaviour, are considered.

Chapter 7 reviews ethical, legal and professional aspects of nursing children and young people with long-term conditions. Scenarios from other chapters are analysed within a framework of ethical principles to identify potential ethical debates and difficult decision-making that practitioners may encounter. The ethical discussions are applied to the practice situation.

Chapter 8 presents a partnership approach between theory and practice, examining changing service boundaries, nursing roles and relationships with parents in the provision of continuing care for children and young people with long-term conditions and their families in the community. To explore this from a practice perspective, multidisciplinary working, discharge planning and respite care are considered using the case scenario of a Welsh-speaking rurally isolated family with a child with the neuromuscular disorder of Batten's disease. The contemporary issue of blended diet administration via gastrostomy is covered in this chapter.

Chapter 9 recognises the importance of acute emergency care, resulting from illness or an unrelated admission, for children and young people with a long-term illness, and the need to ensure effective services and communication processes. Using an oncological haematological condition, current debates and care practices are explored including the need for alternative admission settings.

The last two chapters of the book are especially devoted to teenagers, an increasingly important issue for nurses to consider due to the increasing life expectancy of children with long-term conditions. Chapter 10 provides a critical analysis of the impact of long-term illness upon development transitions of adolescence and the possible health associated risks and longer-term consequences of these. The implications for practitioners in particular focus on communication, body image, compliance and resilience. Chapter 11 builds upon some of the themes raised in Chapter 10 by exploring further a number of aspects of adolescent development in relation to the planning and delivery of effective transition from child to adult services.

This edited book brings together contributions from a team of experienced academics and lecturers in the Children and Young People's Team at the School of Healthcare Sciences, Cardiff University, practitioners, a practice educator and a nurse consultant.

CHAPTER 1

The Definition and Aetiology of Long-Term Conditions

Siân Bill and Angharad Dwynwen Barklam

Introduction

The intention of this chapter is to help the reader further develop their knowledge and understanding of the genetic basis of children and young people's long-term conditions and certain disabilities as a consequence of hereditary influences. Following an overview of chromosomal anomalies, genetic pathways of inheritance will be defined and illustrated via examples of both sex-linked and autosomal recessive and dominant disorders. This chapter does not intend to provide an in-depth critique on the current ethical debates, research and practice controversies surrounding genetic engineering and modification. For this the reader is guided to websites such as www.bionews.org.uk.

The latter half of the chapter focuses on examining the differing onsets of long-term conditions, considering prenatal, neonatal and late onset. To provide the reader with a practice focus, case studies will be used as examples to examine the professional and care implications of nursing children, young people and their families whose long-term conditions have been diagnosed at various stages of their development. To allow these issues to be further developed and explored, the same case studies will be used in subsequent chapters.

Aim of the chapter

To enhance the genetic knowledge and understanding of nurses, including the aetiology of long-term conditions in children, and to examine how this genetic competence can be implemented in their practice to:

- Lead to a reduced risk of conditions occurring, or a reduction in severity for those where a condition has been identified.

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- Enable them to fully participate in the relevant debates and ethical discussions that can have implications for children, young people and their families.

Intended learning outcomes

- To examine the hereditary influences upon the genetic basis of long-term conditions in childhood
- To determine patterns of genetic inheritance
- To investigate the origins of long-term conditions
- To explore the role of the children's nurse during the period leading to, and at the time of, diagnosis

Genetic knowledge

This chapter is written on the assumption that the reader comprehends the basic foundations and principles of genetics. These being: the biology of chromosomes, the structure and role of deoxyribonucleic acid (DNA) in coding genetic information, its ability to replicate and the mechanisms for protein synthesis. In particular, knowledge of the nitrogenous bases and the mechanisms of transcription and translation are required. A good grasp of the cell cycle and its governing control system, along with knowledge of the distinct stages of mitosis and the two divisions, 1 and 2, of meiosis and their resulting products, is also assumed. It is important that the reader has knowledge and understanding of these basic units relating to normal DNA development and of the processes undertaken for the production of sperm and oocytes. Without this knowledge, the reader may find it difficult to comprehend how DNA mutations can cause disease and how errors within the processes of mitosis and meiosis can result in chromosomal abnormalities.

Test your knowledge

- What are the two major phases of a somatic cell cycle?
- What are the four stages of mitosis?
- What are the subdivisions of meiosis 1?
- What are the products of meiosis 2?
- What are the three parts of a DNA nucleotide?
- What are the four nitrogenous bases in DNA?
- In what way does RNA differ from DNA?
- Cells contain three different kinds of RNA. What are they and what is their function in carrying out the instructions encoded in DNA?
- Do you understand the following terms? Haploid and diploid germ cell, homologous chromosomes, allele, heterozygosity and homozygosity?

If the reader wishes to refresh their knowledge on these areas following this test, they are advised to refer to a nursing anatomy and physiology text such as *Wong's Nursing Care of Children* (Hockenberry *et al*., 2019) or Brown (2011) *Introduction to Genetics: A Molecular Approach*. Additionally, the CPD article by Davies and Meimaridou (2020) would be a valuable learning resource.

The need for genetic knowledge

Several authors have argued that children's nurses increasingly need genetic/genomic knowledge to maintain currency of practice (Skirton *et al*., 2010; Botkin *et al*., 2015; Prows, 2019). This is regarded as essential if they are to provide appropriate information and advice to families and be able to engage in policy decisions and relevant genetic debates. The genetics White Paper *Our Inheritance, Our Future* (DoH, 2003) supports this premise, emphasising that education for health professionals is vital to enable advances in genetics to be translated and applied to everyday clinical practice. In response, a genetics educational framework was developed for nurses and midwives by an expert panel and endorsed by the Nursing and Midwifery Council (NMC) (Kirk *et al*., 2013). Since the implementation of the framework in 2003, advances in genetics/genomic has been far more significant than predicted (House of Lords Science and Technology Committee, 2009). The House of Lords Inquiry into genomic medicine (2009) foresaw that as the requests for genetic testing would increase, so would the need for education and training in genetics and genomics across the breadth of the health care professions. The Inquiry urged the NMC to set standards on genetic and genomics across the nursing curriculum for both pre-registration and post-registration education. However, the NMC fell short of this recommendation in their revised pre-registration nurse training requirements in 2010 (Kirk *et al*., 2013). It was therefore seen as timely to review the framework (Kirk *et al*., 2013). Following the inquiry, the Human Genomics Strategy Group (HGSG) was established in 2010. In their report (2012), they continue to emphasise the vital importance of genomic education so that health care professionals can respond to new challenges. A review of training and education included the establishment of Health Education England, who developed core educational standards for genomics which evolve in response to advances made within the field, as well as monitor outcomes. They predict that genomics will have some degree of impact on almost every role in all clinical fields. Therefore, there is a requirement to build an awareness, knowledge and understanding of genomics across the whole of the NHS. The Progress Educational Trust (PET) have also produced a guide to genetics and epigenetics, which provides a good basic overview of genetics (Pembrey, 2012).

For children's nurses, this genetic education would be required to impart several key areas of practice when delivering care and education to children and young people with long-term conditions and their families. Sex education and genetic advice may be required for the teenager with a genetic long-term condition, for example sickle cell anaemia, who may be considering commencing a sexual relationship. Alternatively, parents may require support, advice and guidance following the diagnosis of their child with a genetic disorder. Parents who already have a child with a genetic condition and are considering future pregnancies may also require genetic counselling and advice.

If children's nurses are to deliver sensitive, informed, evidence-based information, education and support to children, young people and their families, they must ensure that they have a current knowledge base upon which to draw. They must also be professionally aware of their limitations in this field and have a good knowledge of, and guide their patients to, local resources and expertise. This could be a hospital's local genetic department or a genetic specialist nurse.

The ethical, legal and social implications in the screening, testing and recording of genetic information

Along with technological advances, our enhanced knowledge and understanding of the human genome and the role of genes in body processes has enabled the mechanisms for genetic screening and testing to be realised for a number of genetic disorders. This new ability to predict the potential for, or to identify, disease-related genes in individuals long before they can be clinically detected, has brought both positive advantages and some practice challenges. For example, knowing from birth that a child has Duchenne muscular dystrophy provides the opportunity for prophylactic treatment regimes and health education strategies to commence immediately. This potentially reduces the complications that can negatively impact upon a child's quality of life. However, this new knowledge has also resulted in some ethical dilemmas and debates that need to be considered; for example, issues such as consent, confidentiality, and the management of situations where the child or young person, their family and the professional's views are not in unison.

Other areas of debate and controversy include who should be tested? What should be the availability of testing? Is mandatory prenatal testing and neonatal screening required or ethical? What are the predictive values of the genetic test and the appropriateness of testing for diseases where there is no treatment or intervention available, as in the case of Huntington's chorea? For those children and families that are tested, there are concerns about possible stigmatisation or discrimination (Williams *et al.*, 2010; Prows, 2019; World Health Organization, 2019) and the role of family counselling within this process (Craufurd *et al.*, 2015; Henneman *et al.*, 2016). A document published by the European Society of Human Genetics recommends that psychosocial support and information should be given pre and post screening and post-screening counselling should be available to all carrier couples (Hennman *et al.*, 2016). Prows (2019) suggests that the most important area for nursing practice is conveying clear information and teaching, making them a valuable resource to help families make the best decision and to reduce anxiety. Ashtiani *et al.* (2014) support this point when discussing parents' experience of receiving their child's diagnosis. In their study, parental experiences were far more positive when they felt that they had active participation in discussions rather than the medical team being dominant and using jargon-heavy language. They required sufficient emotional support and counselling as well as being provided with hope and perspective and a clear plan for follow-ups. It is also suggested that the emotional impact of a diagnosis was less significant when parents

were prepared for the diagnosis and they were better able to receive and retain the information without feeling overwhelmed.

This chapter, however, wishes only to draw the reader's attention to these growing ethical dilemmas and the legal and social issues related to genetic screening and the identification of a genetic disease. Although there is no absolute guide to good action, there are frameworks and models for resolving ethical decision-making. For further information regarding these ethical frameworks, the reader is directed to Chapter 7, where ethical frameworks are used to guide the reader through decisions.

Key points

Children's nurses need genetic competence to implement this knowledge and understanding into their practice in order to:

- Lead to a reduction of risk of conditions occurring, or a reduction in severity for those where a condition has been identified.
- Enable them to fully participate in the relevant debates and ethical discussions that can have implications for children, young people and their families.

The determinants of genetic disease

Due to the intricate nature of DNA formation that occurs during embryological and foetal development, chance mutations or damage can easily alter DNA, producing abnormal sequences of base molecules. There are natural processes within the cell to monitor, recognise and repair defects produced in DNA base sequencing. However, if these internal mechanisms do not detect or repair this damage, expression of the dysfunctional gene can either cause a congenital problem in that child or become part of the genome to be passed on to future generations (hereditary).

Environmental insults to DNA material caused by chemical (carcinogenic), physical (heat) and ionising radiation (X-ray) processes may also produce damage to the genetic material (Martin and Fry, 2018). Damage to somatic cells by radiation, carcinogenic chemicals and ultraviolet light may cause mutations, particularly in cells that are constantly regenerating, and can lead to tumour growth in that individual (Parsa, 2012). However, damage to the sex cells that go on to produce the gametes for fertilisation means that the mutation will not affect the individual but could be passed on to future generations.

The term 'multifactorial inheritance' is used to describe the origin of diseases where there are multiple genetic and environmental factors involved in determining the phenotype, such as leukaemia, where there is familial clustering, and asthma. Some writers believe, however, that the environment has a role to play in all genetic conditions. Although most diseases are influenced by a genetic/genomic predisposition, they can be activated, modified or suppressed by environmental factors (Prows, 2019).

Later in the chapter, prenatal onsets of genetic disorders are discussed in more detail including potential permanent effects caused to the developing foetus by the prenatal intrauterine environment.

Time out

- Before you get to that section write a list of teratogens – agents that cause birth defects.

Chromosomal abnormalities

In humans, each cell, except the germ cells (ova in girls, sperm in boys), contains 46 chromosomes, which are further classified as 22 pairs of autosomes and one pair of sex chromosomes (XX in girls, XY in boys). Located throughout the chromosome are genes, intricate chemical units made up of DNA. As chromosomes are inherited from both parents, individuals have a copy of genes from both the maternal and paternal line. In homologous chromosomes, each gene sequence inherited from the father will have a corresponding gene sequence inherited from the mother (Coleman, 2015). Depending on inheritance factors, the gene sequences may be identical or different.

Chromosomes are numbered according to size and centromere position. Chromosome number 1, for instance, is the largest pair of chromosomes and number 22 the smallest pair of autosomes. The centromere, a constriction on the chromosome either in the centre or close to one end, divides the chromosome into a shorter arm (p) and a longer arm (q). The relative centromeric position allows the morphological classification of chromosomes: metacentric (p and q in equal lengths), submetacentric (q slightly greater than p), acrocentric (q much greater than p), or telocentric (the centromere terminal).

Where a chromosomal anomaly is detected, it can be present in all or just a certain set of cells within the body, demonstrating what is termed a 'mosaic pattern'. Chromosomal anomalies are usually categorised into three discrete areas:

1. Numerical abnormalities, where there is an excess or deficit in the normal complement of 46 chromosomes.
2. Structural abnormalities of the chromosomes.
3. Uniparental disomy, caused through non-disjunction of a chromosome pair.

Numerical abnormalities

If a haploid gamete or a diploid cell lacks the expected number of chromosomes, aneuploidy exists. Monosomy is the term used to depict where there is a deficit in the expected chromosomal numbers. Although autosomal monosomy is usually lethal (e.g. 45XY) in Turner syndrome, monosomy (45X) is not always lethal.

The term 'trisomy' identifies the presence of an additional chromosome. Autosomal trisomy usually occurs as a result of meiotic non-disjunction, with the most common autosomal trisomy being Trisomy 21 (Down syndrome). Other common trisomy syndromes include Trisomy 18 (Edwards syndrome) and Trisomy 13 (Patau

syndrome). The term 'polysomy' is frequently applied if the additional chromosome is a sex chromosome, for example 47XXY (Klinefelter syndrome).

The most common reason for abnormalities in chromosome number is a process called non-disjunction during cell division. Non-disjunction is a failure of separation of the homologous chromosomes during meiosis 1, or of sister chromosomes during meiosis 2. If non-disjunction occurs at meiosis 1, the gamete will have too few chromosomes, or too many if non-disjunction occurs at meiosis 2. Non-disjunction can involve both autosomes and sex chromosomes.

Translocations

Translocations are structural abnormalities where one or more chromosomes break and there is an exchange of genetic material between two or more chromosomes. Translocations are classified into two main types: a Reciprocal translocation and a Robertsonian translocation. In a Reciprocal translocation, the broken fragments of two different chromosomes exchange places. A Robertsonian translocation, however, occurs in acrocentric chromosomes where the centromere is situated near one end, with one arm much longer than the other. Acrocentric chromosomes are Group D (13, 14 and 15) and Group G (21, 22). In these translocations two whole chromosomes merge together through the fusion of their centromeres. One of the most important Robertsonian translocations involves chromosomes 14 and 21.

Translocations are important in heredity, disability and long-term conditions depending on whether they are balanced or unbalanced. Where infants are phenotypically normal and the translocation is referred to as balanced, it is assumed that during the translocation no genetic material was lost or gained and infants are not themselves affected. However, as they are carriers, in adulthood they should carefully consider their decision to have children, as their children could inherit what is termed an unbalanced form of the translocation. However, if infants are phenotypically abnormal, an unbalanced arrangement, either deficiency or duplication of genetic material, is assumed and the translocation is referred to as unbalanced (Chen *et al*., 2011; Chang *et al*., 2013). The degree of disability for a child will depend upon which chromosomes are affected and the extent of genetic material lost or gained. There will, unfortunately though, always be some degree of disability in an unbalanced translocation.

Deletions and duplications

Partial chromosome abnormalities involve a deletion (missing) or duplication (extra) segment of a chromosome. A classic deletion syndrome is Cri du chat, where there is a deletion of the short arm of chromosome 5. Contiguous gene syndrome has been used to identify smaller sections of chromosome abnormalities, such as microdeletions and microduplications (Prows, 2019). The end result is an altered, normal gene dosage, which leads to a specific and complex phenotype that, in some cases, is recognised as a generic syndrome (Strachan *et al*., 2014; Pereira and Marion, 2018). Some major contiguous gene syndromes include DiGeorge syndrome

and Prader-Willi syndrome, both occurring as a result of microdeletions. DiGeorge syndrome involves chromosome 22 and children with this syndrome tend to have cardiac defects, learning difficulties, feeding and speech problems due to a cleft palate or weakness of the palate. Other medical problems can be kidney abnormalities, poor immune systems and neurological and endocrine abnormalities. Prader-Willi syndrome involves a microdeletion on chromosome 15. Classic features of this syndrome include floppy muscles and, initially, poor feeding and weight gain. However, by 3 years of age, children with Prader-Willi syndrome develop large appetites and suffer from obesity. There is also associated pubertal delay along with learning and behavioural challenges.

Chromosomal nomenclature

At a certain stage during cell division, chromosomes form into visible structures and can be detected by photography, producing a picture known as an ideogram.

This picture represents the complete diploid number of chromosomes in a cell called the karyotype.

An official chromosomal nomenclature exists (McGowan-Jordan *et al*., 2016) and designates the chromosomal complement in the following manner:

- The total number of chromosomes (e.g. 45, 46 or 47).
- A comma.
- The sex chromosome complement (XX in normal females; XY in normal males).
- The specific abnormality, if any.

A + or − sign indicates the addition or absence of autosomes in a complement. This is followed by the specific chromosome responsible.

Examples of official nomenclature include:

46, XY	Normal male karyotype
46, XX	Normal female karyotype
45, X	Monosomy X
47, XXX	Polysomy X
47, XXY	Polysomy X
47, XY+21	Trisomy 21 Down syndrome
46, XX, Sp-	Cri du chat syndrome (caused by a deletion on the short arm of chromosome S)

Single gene (Mendelian) disorders

Single gene disorders occur as a result of a mutation or defect, usually involving only a single genetic locus, rather than a partial or total chromosomal abnormality. These disorders normally follow a simple, definite inheritance pattern. However, the transmission of mutant genes within families is dependent upon whether the gene is dominant

or recessive in nature and also whether the mutant gene is located on an autosome or sex chromosome. This leads to the possibility of five transmission patterns:

- autosomal dominant
- autosomal recessive
- X-linked dominant
- X-linked recessive
- Y-linked.

If the homologous chromosomes contain both dominant genes, then the genotype is homozygous dominant and if both are recessive genes, homozygous recessive. If both dominant and recessive genes are present, then the genotype is heterozygous for that trait. This is illustrated in Punnet squares 1 and 2. Mendelian patterns of inheritance are illustrated in Punnet squares 3–12.

Punnet squares 1 and 2.

Example of homozygous and heterozygous gamete

Father has brown eyes with homozygous dominant gamete (BB), and mother has blue eyes with homozygous recessive gamete (bb).

	FATHER		
		B	**B**
MOTHER	**b**	**Bb**	**Bb**
	b	**Bb**	**Bb**

All offspring will have brown eyes and be heterozygous (Bb) for that trait.

If that child, when an adult, has a child with a partner who has blue eyes with homozygous recessive gamete (bb)

	Child now Adult		
		B	**b**
PARTNER	**b**	**Bb**	**bb**
	b	**Bb**	**bb**

then there will be a 50% chance of the offspring having brown eyes and being heterozygous (Bb) for that trait and a 50% chance their child will have blue eyes and be homozygous recessive.

Autosomal recessive inheritance

A large proportion of genetic diseases appear to be inherited in a recessive manner. Consequently, for the gene mutation to be expressed, the offspring must be homozygous recessive for that trait. The heterozygous offspring will be carriers for that gene mutation, with the ability to transfer it to their own children. Examples of autosomal recessive disorders include cystic fibrosis, thalassaemia, sickle cell anaemia and phenylketonuria.

Test your knowledge

With autosomal recessive cystic fibrosis, if one parent has cystic fibrosis and has a child with an adult who is heterozygous for the affected mutant cystic fibrosis gene, what is the percentage chance that their offspring will:

- Be carriers of the cystic fibrosis disease?
- Have cystic fibrosis disease or that their children will be normal?

Punnet squares 3–5.

If the father is heterozygous for the mutant gene cystic fibrosis (Cc) and the mother is homozygous normal (cc)

	FATHER		
		C	c
MOTHER	c	cC	cc
	c	cC	cc

then there will be a 50% chance of the offspring having heterozygous (Cc) and being carriers for cystic fibrosis.

If the father is heterozygous for the mutant gene cystic fibrosis (Cc) and the mother is heterozygous for the mutant gene cystic fibrosis (Cc)

	FATHER		
		C	c
MOTHER	C	CC	Cc
	c	Cc	cc

then there will be a 50% chance of the offspring having heterozygous (Cc) and being carriers for cystic fibrosis, a 25% chance their child will be homozygous normal (cc) and a 25% chance that they will have cystic fibrosis and be homozygous (CC).

	FATHER		
		C	C
MOTHER	c	cC	cC
	c	cC	cC

If a parent who has cystic fibrosis, and is therefore homozygous for the mutant affected gene, has a child with an unaffected homozygous adult, their offspring will all be carriers.

For all Punnet square examples, C is the defective mutant cystic fibrosis gene.