Nursing Care of Children and Young People with Chronic Illness

Edited by

Fay Valentine
RGN, RSCN, MA, PGDip. HSSM, ENB998
Director of Children and Young People’s Directorate, School of Nursing and Midwifery Studies, Cardiff University, UK

Lesley Lowes
RGN, RSCN, PGCHPE, DPSN, M.Sc., Ph.D.
Research Fellow/Practitioner, Nursing, Health and Social Care Research Centre, School of Nursing and Midwifery Studies, Cardiff University, UK

Foreword by
Simon Weston, OBE
Nursing Care of Children and Young People with Chronic Illness
This book is dedicated to all children and young people with chronic illness and their families
Nursing Care of Children and Young People with Chronic Illness

Edited by

Fay Valentine
RGN, RSCN, MA, PGDip. HSSM, ENB998
Director of Children and Young People’s Directorate, School of Nursing and Midwifery Studies, Cardiff University, UK

Lesley Lowes
RGN, RSCN, PGCHPE, DPSN, M.Sc., Ph.D.
Research Fellow/Practitioner, Nursing, Health and Social Care Research Centre, School of Nursing and Midwifery Studies, Cardiff University, UK

Foreword by
Simon Weston, OBE
Contents

Foreword xi
Contributors xii
Introduction xv

Chapter 1 The Definition and Aetiology of Chronic Illness 1
Fay Valentine and Suzanne Hazell

Introduction 1
Aim of the chapter 1
Intended learning outcomes 2
Genetic knowledge 2
The need for genetic knowledge 3
The ethical, legal and social implications in the screening, testing and recording of genetic information 3
The determinants of genetic disease 5
Chromosomal abnormalities 5
Chromosomal nomenclature 7
Single gene (Mendelian) disorders 8
Autosomal recessive inheritance 9
Autosomal dominant inheritance 10
X-linked recessive inheritance 11
X-linked dominant inheritance 12
Inherited variations 12
Antenatal period 15
The neonatal period 18
Post-neonatal period 19
Adolescent period 23
Conclusion 24
Chapter 2  Context of Care and Service Delivery  
*Fay Valentine and Peter Mcnee*

Introduction  
Aim of the chapter  
Intended learning outcomes  
Context of change  
Political influences  
Economic influences  
Social influences  
New models of service delivery  
Modernising workforce  
Increasing patient expectations and engagement  
Locally based commissioning  
Staffing implications  
Staff education and competence  
Meeting parental needs  
Innovative practices – new roles  
Telemedicine  
Key worker role  
Mental health issues  
Multi-agency working  
Multidisciplinary team (MDT) working  
Nursing implications, challenges and opportunities in MDT working  
Conclusion  
Useful websites  
References

Chapter 3  Impact upon the Child and Family  
*Lesley Lowes*

Introduction  
Aim of the chapter  
Intended learning outcomes  
Theories of grief, loss and change  
Initial impact  
Type 1 diabetes  
Impact on parents  
Impact on the child/young person with chronic illness  
Continuing care  
Impact on siblings  
Coping, adaptation and change  
Theories of stress and coping
## Chapter 4  A Holistic Approach to Meeting Physical, Social and Psychological Needs
*Beverly Hodges and Julia Tod*

<table>
<thead>
<tr>
<th>Section</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Introduction</td>
<td>84</td>
</tr>
<tr>
<td>Aim of the chapter</td>
<td>84</td>
</tr>
<tr>
<td>Intended learning outcomes</td>
<td>85</td>
</tr>
<tr>
<td>Prevalence of eczema</td>
<td>85</td>
</tr>
<tr>
<td>Nursing considerations</td>
<td>86</td>
</tr>
<tr>
<td>Presentation of eczema</td>
<td>87</td>
</tr>
<tr>
<td>Treatments of eczema with emollients</td>
<td>87</td>
</tr>
<tr>
<td>Effects of the itch scratch cycle</td>
<td>89</td>
</tr>
<tr>
<td>Family stress and coping</td>
<td>90</td>
</tr>
<tr>
<td>Assessment and immediate intervention</td>
<td>92</td>
</tr>
<tr>
<td>Current care management plan</td>
<td>93</td>
</tr>
<tr>
<td>Pain assessment</td>
<td>94</td>
</tr>
<tr>
<td>Play and distraction</td>
<td>96</td>
</tr>
<tr>
<td>Bullying</td>
<td>96</td>
</tr>
<tr>
<td>Spirituality</td>
<td>99</td>
</tr>
<tr>
<td>Complementary and alternative medicine</td>
<td>101</td>
</tr>
<tr>
<td>Health promotion</td>
<td>102</td>
</tr>
<tr>
<td>Conclusion</td>
<td>103</td>
</tr>
<tr>
<td>Acknowledgement</td>
<td>104</td>
</tr>
<tr>
<td>Useful websites</td>
<td>104</td>
</tr>
<tr>
<td>Recommended reading</td>
<td>104</td>
</tr>
<tr>
<td>References</td>
<td>104</td>
</tr>
</tbody>
</table>

## Chapter 5  Empowering Children, Young People and their Families
*Mandy Brimble*

<table>
<thead>
<tr>
<th>Section</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Introduction</td>
<td>107</td>
</tr>
<tr>
<td>Aim of the chapter</td>
<td>107</td>
</tr>
<tr>
<td>Intended learning outcomes</td>
<td>108</td>
</tr>
<tr>
<td>Health promotion</td>
<td>108</td>
</tr>
<tr>
<td>Empowerment</td>
<td>111</td>
</tr>
<tr>
<td>Approaches to health promotion</td>
<td>112</td>
</tr>
<tr>
<td>Parental smoking</td>
<td>115</td>
</tr>
<tr>
<td>Obesity</td>
<td>116</td>
</tr>
<tr>
<td>Non-compliance</td>
<td>122</td>
</tr>
</tbody>
</table>
Chapter 6  Ethical Issues

Peter Mcnee and Maggie Furness

Introduction 131
Aim of the chapter 131
Intended learning outcomes 132
Why study ethics? 132
A definition of ethics 132
Introduction to ethical theories 133
Theory of consequentialism 133
Utilitarianism 134
Act utilitarianism 134
Rule utilitarianism 135
Theory of deontology 138
Ethical principles 140
Principle of autonomy 141
Ethical principle of beneficence 144
Ethical principle of non-maleficence 146
Ethical principle of justice 148
A nursing ethic 149
Definition of an ethical dilemma 150
Ethical decision making 151
A suggested model for ethics in practice/decision making 152
Conclusion 154
Useful websites 154
Recommended reading 155
References 155

Chapter 7  Continuing Care Needs

Melda Price and Sian Thomas

Introduction 157
Aim of the chapter 157
Intended learning outcomes 157
Complex health needs 158
Transition to home care 165
Continuing care policy 168
Framework for assessment of children in need 169
National Service Framework for Children, Young People and Maternity Services 169
Carers’ assessments 169
<table>
<thead>
<tr>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chronic renal failure</td>
</tr>
<tr>
<td>Communicating with young people</td>
</tr>
<tr>
<td>Communication issues for Thomas</td>
</tr>
<tr>
<td>Peer groups</td>
</tr>
<tr>
<td>The impact of chronic illness on growth and development</td>
</tr>
<tr>
<td>Delayed growth and puberty</td>
</tr>
<tr>
<td>Inconsistencies between physical and psychological development</td>
</tr>
<tr>
<td>Body image and self Esteem</td>
</tr>
<tr>
<td>Compliance and non-compliance</td>
</tr>
<tr>
<td>The social aspects of chronic illness</td>
</tr>
<tr>
<td>Young people, chronic illness and school</td>
</tr>
<tr>
<td>Bullying</td>
</tr>
<tr>
<td>The hospitalised adolescent</td>
</tr>
<tr>
<td>Issues for young people in hospital</td>
</tr>
<tr>
<td>Specialised adolescent units</td>
</tr>
<tr>
<td>Service delivery and adolescent health</td>
</tr>
<tr>
<td>Promoting excellence</td>
</tr>
<tr>
<td>The role of the nurse</td>
</tr>
<tr>
<td>Conclusion</td>
</tr>
<tr>
<td>Useful websites</td>
</tr>
<tr>
<td>Recommended reading</td>
</tr>
<tr>
<td>References</td>
</tr>
</tbody>
</table>

### Chapter 10  Transitional Care

*Siân Bill and Beverly Hodges*

<table>
<thead>
<tr>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Introduction</td>
</tr>
<tr>
<td>Aim of the chapter</td>
</tr>
<tr>
<td>Intended learning outcomes</td>
</tr>
<tr>
<td>Overview of cystic fibrosis</td>
</tr>
<tr>
<td>Management</td>
</tr>
<tr>
<td>Transitional care</td>
</tr>
<tr>
<td>Issues regarding transition for adolescents</td>
</tr>
<tr>
<td>Gaining autonomy</td>
</tr>
<tr>
<td>Managing a chronic illness</td>
</tr>
<tr>
<td>Support and decision making</td>
</tr>
<tr>
<td>The role of the nurse</td>
</tr>
<tr>
<td>Promoting the paradigm shift</td>
</tr>
<tr>
<td>The principles of successful transitional care</td>
</tr>
<tr>
<td>Conclusion</td>
</tr>
<tr>
<td>Useful websites</td>
</tr>
<tr>
<td>Recommended reading</td>
</tr>
<tr>
<td>References</td>
</tr>
</tbody>
</table>

**Index**

<table>
<thead>
<tr>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>256</td>
</tr>
</tbody>
</table>
Foreword

Simon Weston OBE

When approached to write this foreword, I felt I needed to examine my qualifications for addressing the need for a book relating to the nursing requirements of children and young people with chronic illness. I realised that as the son of a nurse, the father of a child with asthma and eczema and the survivor of 48% burns (80% body scarring) whilst only 20 years old, I am more than qualified to speak from a layperson’s perspective.

In today’s ever moving and multicultural society, this book is long overdue. The task set for the authors has been to create an all encompassing book that will deal not only with the issues of chronic disease in children and young people, but also with a whole catalogue of situations that the modern nurse will have to deal with when faced with their care.

Having examined the premise for this book, it is clear that it will alleviate the need to wade through heavily jargoned and complicated texts, spread over numerous manuals, thus using up student nurses’ valuable study time. This book provides a quality tool that will raise the awareness and effectiveness of not just student nurses but all health professionals working with children and young people with chronic illness.

The material collated spans the knowledge and experiences of several renowned and articulate professionals whose combined talents form an interlaced understanding of the total need of children and young people with chronic illness. It gives a holistic view of illness, emotions, cultures and religions, all of which may impact on the ability of the child and family to cope with a chronic condition.

I firmly believe that this book, with its compilation of informative and insightful data, will help ensure the empowerment of children and young people with chronic illness and their families through increased awareness and understanding by those involved in their care.
Contributors

Siân is a lecturer within the Children and Young People’s Directorate at the School of Nursing and Midwifery Studies, Cardiff University. Her main interests are in adolescent chronic illness, in particular cystic fibrosis, and in transitional care. Siân has worked extensively with young people in both the acute care and community setting.

Mandy Brimble RN, Dip. HE (Child), B.Sc. (Hons) Community Health Studies (Public Health Nursing/Health Visiting), PGCE
Mandy is a lecturer in the Children and Young People’s Directorate, School of Nursing and Midwifery Studies, Cardiff University. She has worked as a children’s nurse in general medicine, day surgery and at a special children’s centre. She has an interest in a wide range of medical and surgical conditions as well as a specific interest in all aspects of child protection. Her training and work as a health visitor has fostered a keen interest in public health and health promotion, particularly childhood accident prevention. Mandy has recently commenced an MSc in Education, with an aim towards gaining a deeper insight into strategic teaching, learning and educational management issues.

Maggie Furness SRN, RSCN, RNT, RCNT, WNB(6), ONC, MA (Ethics)
Maggie is a lecturer within the Children and Young People’s Directorate at the School of Nursing and Midwifery Studies, Cardiff University. She has worked in a wide range of clinical settings including orthopaedics, theatres and general paediatric areas. She has a wide range of interests, including ethical issues in children’s nursing and the history of nursing.
Suzanne Hazell RSCN, RGN, BA, MN, PGCE
Having qualified as a general nurse in 1967, Suzanne has many years’ experience of children’s nursing, mainly in the field of neonatal nursing in several UK hospitals. Subsequently, she undertook the RSCN and neonatal nursing courses and a Master’s in nursing. It was while nursing neonates that she developed an interest in genetics and the aetiology of conditions affecting children and young people. For the last 11 years, Suzanne has been lecturing in children and young people’s nursing and has continued to develop this interest.

Beverly Hodges M.Sc. in Clinical Practice (Child), PGCE, B.Sc. (Hons), Dip. Asthma, Dip. Allergy and Immunology, L/PE, RSCN, RGN
Beverly is a lecturer in the Children and Young People’s Directorate at the School of Nursing and Midwifery Studies, Cardiff University. She has worked in a variety of settings caring for children and young people. Her specialist interest areas are respiratory health, allergic disease, pain management and management of change. Beverly was a member of the external working group for the children and young people specialist services project, for the All-Wales Standards for Paediatric Respiratory Services, with the Welsh Assembly Government. She has represented the Royal College of Nursing in responding to appraisals for the National Institute of Health and Clinical Excellence.

Yvonne Knight RSCN, EN(G), B.Sc. (Hons), PGDip. (Professional Health Care Education), ENB 147, 820, 998
Yvonne is a lecturer in the Children and Young People’s Directorate, School of Nursing and Midwifery Studies, Cardiff University. Her clinical experience and research interests are in the acute and chronic nursing needs of children. She is currently studying for a Master’s in child health.

Lesley Lowes RGN, RSCN, PGCHPE, DPSN, M.Sc., Ph.D.
Lesley is a research fellow/practitioner at the Nursing, Health and Social Care Research Centre, School of Nursing and Midwifery Studies, Cardiff University. She is an experienced qualitative researcher, whose research interests include childhood chronic illness (diabetes in particular), theories of loss, grief, adaptation and change, and the involvement of service users in health and social care research. She has an extensive clinical and academic publication portfolio, particularly in the field of paediatric diabetes. In addition to her academic work, she has a 50% clinical remit as a paediatric diabetes specialist nurse with Cardiff and Vale NHS Trust.

Peter Mcnee RGN, RSCN, ENB (415), BA (Hons), PGCE, M.Sc.
Peter is a lecturer in the Children and Young People’s Directorate, School of Nursing and Midwifery Studies, Cardiff University. His clinical background is predominantly within paediatric critical care. His main interests include the acquisition of clinical skills, bereavement care, child protection and children’s participation in care decisions.
Martina Nathan RSCN, RGN, B.Sc. Professional Practice, PGCE
Martina is currently on secondment to the Children and Young People’s Directorate from the position of Paediatric Chemotherapy Nurse Trainer, Cardiff and Vale NHS Trust. She has spent nine years working in the acute paediatric oncology setting, in Ireland, Singapore and Cardiff.

Melda Price B.Sc. (Hons) RGN, RSCN, PGCHPE, Dip. DN
Melda is currently a lecturer in Children’s Nursing in the Children and Young People’s Directorate, School of Nursing and Midwifery Studies, Cardiff University. Her teaching expertise is in the care of children with chronic illness and complex conditions in the community, with an emphasis on inter-professional collaboration in care. Her research interests are community children’s nursing, paediatric oncology and paediatric palliative care.

Sian Thomas RGN, RSCN, M.Sc.
Sian is a Nurse Consultant in Community Child Health at Caerphilly Local Health Board. Prior to this post, she was the manager of the Community Children’s Nursing Service at Cardiff and Vale NHS Trust. During this time she had the opportunity to undertake a secondment to the Welsh Assembly Government as Project Manager for the Children and Young People’s Specialist Services Project.

Julia Tod RGN, RM, RSCN, ENB405/998, B.Sc. (Hons) Psychology, PGCE
Julia’s clinical nursing practice was mainly in neonatal care in North and South Wales, Glasgow and Nottingham. A joint post (Gwent NHS Trust/UWCM) as children’s nursing clinical teacher led to her current role as nurse lecturer at Cardiff University. Currently studying for a Master’s in health psychology, her research interests include health behaviours, the role of young carers and application of psychology in health care.

Fay Valentine RGN, RSCN, MA, PGDip. HSSM, ENB998
Fay is Director of the Children and Young People’s Directorate in the School of Nursing and Midwifery Studies, Cardiff University. She has a long history of working with children and young people with chronic illness. Her specialist interests are leadership, adolescence and chronic illness. She is the Welsh representative on the RCN Children and Young People’s Advisory Panel, a member of the All-Wales Senior Children’s Nursing Forum and committee member for the Welsh Nursing and Midwifery Council.

Simon Weston OBE
Following his injuries (48% burns) while a Welsh Guardsman on the Sir Galahad, which was destroyed in the Falkland Islands’ Bluff Cove, Simon has been active in a number of successful ventures, including the establishment of the Weston Spirit, a Liverpool based but UK-wide young people’s charity. He has worked tirelessly for the Royal Star and Garter Home and his charitable work earned him an OBE in the 1992 Queen’s Birthday Honours. In 2005, Simon was awarded an honorary fellowship from the School of Nursing and Midwifery at Cardiff University.
Introduction

Fay Valentine and Lesley Lowes

There are currently limited books available that analyse the context, theory and practice of nursing children and young people with chronic illness. This book provides a comprehensive and up-to-date 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 chronic illness and their families.

Although each chapter can be read independently, the book is designed to encompass a broad perception of the changing health care needs of children and young people with chronic illness and the implications for delivering nursing practices 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 scenarios and reader activities are used to apply theoretical principles and current evidence to the realities of nursing practice. In addition, readers are able to gain a greater understanding of the clinical conditions used in the case scenarios in relation to age development issues and associated care needs.

Chapter 1 revisits the aetiology of chronic illness, examining the generic basis of children and young people’s chronic 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 chronic illness, 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 chronic 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 chronic illness on the child and family, 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 chronic 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 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 chronic 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 5 provides insights into the general principles for the need to inform, educate and promote health to children and young people with chronic illness 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 6 reviews ethical, legal and professional aspects of nursing children and young people with chronic illness. 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 7 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 chronic illness 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 rural isolated family with a child with the neuromuscular disorder of Batten’s disease.

Chapter 8 recognises the importance of acute emergency care, resulting from illness or an unrelated admission, for children and young people with chronic 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 chronic illnesses. Chapter 9 provides a critical analysis of the impact of chronic 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 10 builds upon some of the themes raised in Chapter 9 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 lecturers in the Children and Young People’s Directorate at the School of Nursing and Midwifery Studies, Cardiff University, along with the only consultant children’s community nurse in Wales.
1 The Definition and Aetiology of Chronic Illness

Fay Valentine and Suzanne Hazell

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 chronic 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 nursing anatomy and physiology books.

The latter half of the chapter focuses on examining the differing onsets of chronic illness, 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 chronic illness has 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 chronic illness in children and 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
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 chronic diseases in childhood
- To determine patterns of genetic inheritance
- To investigate the origins of chronic disease
- 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?
The need for genetic knowledge

Several authors have argued that children’s nurses need genetic knowledge to maintain currency of practice (Skirton & Patch, 2002; Edgar, 2004; Skirton et al., 2005; Burke & Kirk, 2006). 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. To support this White Paper, the Department of Health commissioned the development of guidelines for use across the UK for genetic education programmes for nurses, midwives and health visitors. This resulted in the publication of a competence based education framework containing seven competency standard statements and associated learning outcomes (Kirk et al., 2003). In 2005, a series of seven papers were published by the Royal College of Nursing (RCN) outlining the framework and application of the seven competency standards to nursing, midwifery and health visiting practice (Kirk, 2005). The RCN in collaboration with the Progress Educational Trust (PET) have also produced a guide to genetics for nurses, which provides a good basic overview of genetics (PET/RCN, 2006).

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 chronic illness and their families. Sex education and genetic advice may be required for the teenager with a genetic chronic illness, 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 and the role of family counselling within this process (Edgar, 2004; Kenner & Moran, 2005). Barr and McConkey (2006) support this point when discussing the support parents require during the referral and process of genetic investigations. They highlight that parents view this process as part of a ‘longer journey’ when obtaining information, advice and support about their child’s condition or disability via services that have established protocols for effective collaboration between primary care, secondary care and regional genetic services.

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. Bradley (2005) also outlines some example scenarios in relation to the utility and limitations of genetic testing and information.

**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. 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 (Jones, 2004). 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 ‘multi-factorial 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 (Thurmon, 1999).

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 (Jones, 2004). 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 chronic illness 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 (Simpson & Elias, 2003). 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 duplications. 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 (Skirton & Patch, 2002). 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 three 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 (ISCN, 1995) 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.
The Definition and Aetiology of Chronic Illness

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

<table>
<thead>
<tr>
<th>FATHER</th>
<th>CHILD now ADULT</th>
</tr>
</thead>
<tbody>
<tr>
<td>MOTHER</td>
<td>PARENT</td>
</tr>
<tr>
<td>B</td>
<td>B</td>
</tr>
<tr>
<td>b</td>
<td>Bb</td>
</tr>
<tr>
<td>b</td>
<td>Bb</td>
</tr>
<tr>
<td></td>
<td>B</td>
</tr>
<tr>
<td>b</td>
<td>Bb</td>
</tr>
<tr>
<td>b</td>
<td>Bb</td>
</tr>
</tbody>
</table>

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

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?