John M. Hutson Spencer W. Beasley

# The Surgical Examination of Children

Second Edition



# The Surgical Examination of Children

### John M. Hutson • Spencer W. Beasley

# The Surgical Examination of Children

**Second Edition** 



Authors
John M. Hutson, AO, BS, M.D.
(Monash), M.D., DSc (Melb)
FRACS, FAAP (Hon)
Department of Urology
Royal Children's Hospital
Melbourne
Australia

Spencer W. Beasley, MB. ChB, M.S. (Melb), FRACS
Paediatric Surgery Department
Otago University
Christchurch Hospital
New Zealand

ISBN 978-3-642-29813-4 ISBN 978-3-642-29814-1 (eBook) DOI 10.1007/978-3-642-29814-1 Springer Heidelberg New York Dordrecht London

Library of Congress Control Number: 2012945914

#### © Springer-Verlag Berlin Heidelberg 2013

This work is subject to copyright. All rights are reserved by the Publisher, whether the whole or part of the material is concerned, specifically the rights of translation, reprinting, reuse of illustrations, recitation, broadcasting, reproduction on microfilms or in any other physical way, and transmission or information storage and retrieval, electronic adaptation, computer software, or by similar or dissimilar methodology now known or hereafter developed. Exempted from this legal reservation are brief excerpts in connection with reviews or scholarly analysis or material supplied specifically for the purpose of being entered and executed on a computer system, for exclusive use by the purchaser of the work. Duplication of this publication or parts thereof is permitted only under the provisions of the Copyright Law of the Publisher's location, in its current version, and permission for use must always be obtained from Springer. Permissions for use may be obtained through RightsLink at the Copyright Clearance Center. Violations are liable to prosecution under the respective Copyright Law.

The use of general descriptive names, registered names, trademarks, service marks, etc. in this publication does not imply, even in the absence of a specific statement, that such names are exempt from the relevant protective laws and regulations and therefore free for general use. While the advice and information in this book are believed to be true and accurate at the date of publication, neither the authors nor the editors nor the publisher can accept any legal responsibility for any errors or omissions that may be made. The publisher makes no warranty, express or

Printed on acid-free paper

Springer is part of Springer Science+Business Media (www.springer.com)

implied, with respect to the material contained herein.

To study the phenomena of disease without books is to sail an uncharted sea, while to study books without patients is not to go to sea at all.

Sir William Osler: Books and Men, Address dedicating a new building for the Boston Medical Library, 1901

#### **Preface**

This book aims to teach medical students and other medical and clinical personnel how to perform a clinical examination in the infant or child who has a potential surgical condition. Most textbooks of paediatric surgery focus on the pathological classification and overall management of disease, rather than on the practical details of how a diagnosis is made. However, no matter how much knowledge trainees and surgeons have about diseases and their treatment, this is useless if the diagnosis is wrong: the foundations must be solid if we wish to construct a sophisticated plan of management. In clinical practice, experienced physicians and surgeons usually use a problem-oriented approach to clinical diagnosis, yet this is rarely taught to students. This book attempts to redress this imbalance by providing a clinical approach to the patient which the student can learn with a minimum of factual information. Therefore, it should remain useful to the practitioner throughout his or her medical career. This book includes a detailed coverage of the common presentations of common diseases and does not attempt to cover all aspects of the presentations of uncommon diseases unless their recognition is important for the well-being or survival of the child. With the increasing sophistication and specialization of children's hospitals, the exposure of medical students to rare diseases has increased in recent years. This is a counterproductive trend since it means they obtain little experience in the range of problems that they will meet in practice subsequently. For this reason, the commonest problems are discussed in the early chapters of the book, and the last few chapters deal with uncommon or postgraduate topics.

Clinical examination is a very subjective process: one should not shy away from this in a quest for objectivity, but rather one should use deliberately the subjective nature of the mind to advantage.

Melbourne, Australia Christchurch, New Zealand John M. Hutson Spencer W. Beasley

#### **Acknowledgements for the First Edition**

A large number of people have made valuable contributions to this book. We thank Anne Esposito from the Royal Children's Hospital Education Resource Centre for her devotion and effort with the artwork, which is central to the success of this book. Elizabeth Vorrath, Judith Hayes and Vicki Holt are thanked for their untiring help with the manuscript, despite their normal, heavy secretarial duties. We are grateful to our colleagues for support and encouragement and for their criticisms of various chapters: Alex Auldist (Chaps. 8, 16 and 17), Dr. Agnes Bankier (Chap. 2), Dr. Kester Brown (Chaps. 12 and 20), Bob Dickens (Chap. 13), Justin Kelly (Chaps. 7, 14 and 15), Max Kent (Chaps. 5, 9 and 21), Geoffery Klug (Chaps. 10, 12 and 18), Dr. Peter McDougall (Chap. 2), Nate Myers (Chaps. 1 and 3), Prof. Peter Phelan (Chaps. 1, 3, 6 and 8), Keith Stokes (Chap. 12) and Hock Tan (Chaps. 4, 6 and 11).

Dr. Richard Barling of Heinemann Medical Books is thanked for his encouragement, enthusiasm and help in completing the project.

### **Acknowledgements for the Second Edition**

Sincere thanks to Shirley D'Cruz for scanning the original text and with great patience turning it into a useable electronic document, thereby enabling this book to be updated and edited for a new edition. We also thank Lloyd Ellis for transforming the negatives of the original diagrams into digital format.

#### **Contents**

1	General Principles	1
	What Knowledge Is Needed to Make a Diagnosis	
	or Solve a Clinical Problem?	1
	Why Does a Problem-Oriented Approach Work?	2
	Does the Problem-Orientated Approach Work for Fields	
	of Medicine Other Than Paediatric Surgery?	2
	If Anticipation of the Diagnosis Comes with Experience,	
	How Can the Student Hope to Learn the Technique	
	While Still Inexperienced?	3
	Specific Principles of Clinical Examination	3
	Further Reading	7
_		
2	Is the Baby Normal?	9
	Embryology	9
	Surgical Assessment of the Neonate	12
	Further Reading.	18
3	Abdominal Pain: Is It Appendicitis?	19
	General Considerations	20
	The History	24
	Preparation for Examination	26
	Is There Peritonitis?	27
	Non-specific Signs of Peritonitis	31
	Appendicitis	31
	A Mass in the Right Iliac Fossa	35
	Other Pathologies in Suspected Appendicitis	36
	Further Reading	39
4	Inguinoscrotal Lesions	41
	Embryology	41
	The Undescended Testis	42
	The Acutely Painful Scrotum	49
	The Painless, Swollen Scrotum	53
	The Groin Lump	55
	Further Reading.	61
5	Abnormalities of the Penis	63
	The Normal Uncircumcised Penis	63
	Smegma Deposits	63

	The Normal Circumcised Penis	64
	Examination of the Non-adherent Foreskin	64
	Examination of the Adherent Foreskin	65
	Phimosis	65
	The Red, Swollen Penis	66
	Meatal Ulcer	68
	Meatal Stenosis	69
	Hypospadias	69
	Further Reading	73
6	The Umbilicus	75
O		76
	Clinical Diagnosis	82
	Further Reading	82
7	Non-Bile-Stained Vomiting in Infancy	83
	Clinical Features.	83
8	Colic	91
Ü	Intussusception	91
	The Clinical Presentation	93
	Further Reading	99
		,,
9	The Neck	101
	Torticollis (Wry-Neck)	101
	The Side of the Neck	107
	Midline Swellings of the Neck	116
10	The Head and Face	123
	The Head	123
	Facial Lumps and Anomalies	132
	Further Reading.	138
11	The Mouth	139
11	The Mouth	
	The Tongue  Lesions in the Floor of the Mouth	139 141
		141
	Lesions of the Gums	
	Lesions of the Polete	144
	Lesions of the Palate	145
	Lesions of the Palate  Lesions of the Oropharynx	145 147
	Lesions of the Palate	145
12	Lesions of the Palate  Lesions of the Oropharynx	145 147
12	Lesions of the Palate Lesions of the Oropharynx Further Reading  Trauma Initial Assessment of the Severely Injured Child	145 147 149
12	Lesions of the Palate Lesions of the Oropharynx Further Reading  Trauma Initial Assessment of the Severely Injured Child Abdominal Trauma	145 147 149 151 151 153
12	Lesions of the Palate Lesions of the Oropharynx Further Reading  Trauma Initial Assessment of the Severely Injured Child Abdominal Trauma Thoracic Injuries	145 147 149 151
12	Lesions of the Palate Lesions of the Oropharynx Further Reading.  Trauma Initial Assessment of the Severely Injured Child Abdominal Trauma Thoracic Injuries Head Injuries	145 147 149 151 151 153
12	Lesions of the Palate Lesions of the Oropharynx Further Reading  Trauma Initial Assessment of the Severely Injured Child Abdominal Trauma Thoracic Injuries	145 147 149 151 151 153 156
12	Lesions of the Palate Lesions of the Oropharynx Further Reading.  Trauma Initial Assessment of the Severely Injured Child Abdominal Trauma Thoracic Injuries Head Injuries	145 147 149 151 151 153 156 159
12	Lesions of the Palate Lesions of the Oropharynx Further Reading.  Trauma Initial Assessment of the Severely Injured Child Abdominal Trauma Thoracic Injuries Head Injuries Perineal Injuries Fractures Burns	145 147 149 151 151 153 156 159 169
12	Lesions of the Palate Lesions of the Oropharynx Further Reading.  Trauma Initial Assessment of the Severely Injured Child Abdominal Trauma Thoracic Injuries Head Injuries Perineal Injuries Fractures	145 147 149 151 151 153 156 159 169 172

13	The Limbs	185
	The Limbs at Birth	185
	The Legs in Early Childhood	187
	The Acute Limb	191
	Osteomyelitis	191
	Septic Arthritis	194
	Limp	195
	Miscellaneous Problems	201
	Further Reading.	202
	C	200
14	Urinary Tract Symptoms.	203
	The Vomiting Neonate	203
	Dysuria and Frequency	203
	Loin Pain	204
	Collecting a Specimen of Urine	204
	What Is the Significance of Proven Infection?	208
	Hypertension	209
	Wetting	209
	Haematuria	212
	Further Reading	214
15	Abdominal Masses	215
10	Questions to Be Answered	215
	Normal Masses	216
	Common Abnormal Abdominal Masses	216
	Tumour Presenting as an Abdominal Mass	217
	Uncommon Abnormal Masses	221
	Neonatal Abdominal Masses	224
	Pelvic Lesions	227
	Further Reading	230
16	Gastrointestinal Bleeding	231
	The Appearance of the Stool	231
	The Appearance of the Vomitus	231
	Minor Bleeding in the Healthy Neonate	231
	Bleeding in the Sick Neonate	232
	Small Amounts of Rectal Bleedingin a Well Child	233
	Bleeding with an Acute Abdomen	235
	Major Gastrointestinal Haemorrhage	236
	Chronic Diarrhoea and Bleeding	236
17	Jaundice	237
-,	Assessment of a Patient with Persistent Neonatal Jaundice	237
	Portal Hypertension	240
	Cholelithiasis	242
18	Spina Bifida	245
	Embryology	245
	Neonatal Assessment of Spina Bifida	245
	Spina Bifida Occulta	253
	Further Reading	255

xvi Contents

19	Abnormalities of the Chest Wall and Breast	257
	The Anterior Chest Wall	257
	The Posterior Chest Wall	259
	The Breast	264
20	Respiratory Distress in the Newborn	269
	Clinical Features	269
	Cause Obvious on External Examination	270
	Respiratory Distress Relieved by Crying	271
	The Frothy Baby in Respiratory Distress	272
	Onset of Severe or Progressive Respiratory Distress	274
	Sudden Onset of Respiratory Distress	276
21	Neonatal Bowel Obstruction	279
	The Causes of Bowel Obstruction	279
	The Presentation of Bowel Obstruction	284
	Physical Examination	288
	Further Reading	292
22	Ambiguous Genitalia: Is It a Boy or a Girl?	293
	Normal Sexual Development	293
	Abnormal Sexual Development: Ambiguous Genitalia	294
	The Clinical Presentation of DSD	297
	Investigation	302
	Absent Vagina	302
	Further Reading	304
Ind	lex	305

**General Principles** 

Clinical diagnosis in any branch of medicine requires more finesse than knowing merely how to elicit physical signs. It is often a great mystery to medical students how an experienced doctor can reach a correct diagnosis with the minimum of history and examination while they may have spent an hour or more with the same patient to no avail. They may despair of ever attaining the same level of expertise. Shrewd students, however, suspect that their senior colleague uses a direct approach to reach the diagnosis, avoiding the lengthy process of aimless and exhaustive history-taking and examination, much of which may be irrelevant to the patient's needs. It is the aim of this book to teach students of paediatric surgery how to improve their clinical acumen and to enable them to reach the correct diagnosis by the simplest route. This direct approach to clinical diagnosis is problem-oriented, where the diagnosis can be anticipated by the early elimination of alternatives. It is the same method that is used in 'decision trees', algorithms and flow charts. Although students may be aware already of this type of decision-making process, its formal description in the following pages underlines the strength of the technique and demonstrates how it can be applied in clinical diagnosis. Consider the following difference in approach to the simple mathematical problem of identifying an unknown number between 1 and 1,000 by asking questions with a 'yes' or 'no' answer. How many such questions are needed? There are two ways of solving this problem. One is to ask whether every number in sequence is the chosen one, beginning with 1 and continuing for up to 999 questions until the correct number is identified! An alternative approach is based on the realization that a 'yes/ no' question identifies specifically a number in a two-element set. Therefore, all that is needed is to divide the 1,000 choices into two and ask: 'Is the number greater than 500?' The answer will immediately eliminate half the possibilities. With this simple device, the unknown number can always be found in ten questions or fewer. The first technique is analogous to the student who asks every conceivable question and then examines every part of the body from head to toe, before thinking about the possible diagnosis. The latter approach echoes that of the experienced physician who reaches the diagnosis after limited questioning and physical examination.

# What Knowledge Is Needed to Make a Diagnosis or Solve a Clinical Problem?

First, an understanding of the scientific foundations is required; in paediatric surgery, this involves knowledge of the embryology, anatomy and physiology of the neonate and infant and of normal growth and development. Secondly, it is essential to be familiar with the common pathological processes (which vary in different times, cultures and localities) to ensure a sensible differential diagnosis.

1

2 1 General Principles

# Why Does a Problem-Oriented Approach Work?

The answer to this question is not obvious immediately, yet it is a fundamental principle of problemsolving by observation. In a clinical context, the clues to the diagnosis are often subtle and will be found only by someone who knows exactly where to look and then knows how to interpret the findings. A physical sign may be detectable if the examiner is working with a hypothesis and specifically looks to see whether the sign is present, but will be missed on 'routine' examination. It is rare that important evidence from the history or physical examination is found by accident – the observer must be consciously prepared for it (Fig. 1.1).

While it may seem improbable at first, it is a well-recognized characteristic of the brain and senses to ignore apparently irrelevant observations. Pasteur expressed the sentiment when he said: 'Fortune favours the prepared mind'. Once this limitation of the senses is appreciated, it is

evident that the secret of rapid and accurate diagnosis lies in having a prepared mind at the commencement of the interview and examination. Only then can important clues actively be sought from the history and examination.

#### Does the Problem-Orientated Approach Work for Fields of Medicine Other Than Paediatric Surgery?

The approach is applicable to all branches of medicine, but the lack of degenerative diseases in paediatrics means that multiple disease processes are less likely to account for the presenting complaint; this is an example of the principle known as Ockham's razor. William of Ockham was an English monk who provided scientific method with its fundamental principle 700 years ago when he suggested we should always favour simple explanations when trying to account for the world's mysteries. In paediatric surgery, this

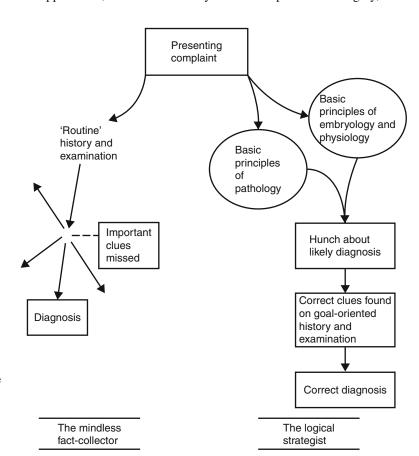


Fig. 1.1 Unless clinicians deliberately consider the relevance of the information they obtain they will become mindless fact-collectors, perform a meaningless examination and miss the clues vital to arriving at the correct diagnosis

means that a range of different physical signs are most likely to be caused by a single anomaly.

# If Anticipation of the Diagnosis Comes with Experience, How Can the Student Hope to Learn the Technique While Still Inexperienced?

This is the 'Catch-22' of medical education. It is our belief that the technique can be learnt after limited clinical experience, subject to certain conditions.

First, the significance of each piece of evidence must be considered from the beginning of the interview, so that a simple list of differential diagnoses can be formulated early. Failure to recognize the significance of information as it is provided will prejudice recognition of those clues which assist in the identification of possible diagnoses. Therefore, when first learning the method, students must ask themselves continuously what the evidence suggests and which relevant questions should be asked. Later, with experience and practice, this process occurs automatically.

Secondly, it must be understood that many problems can be solved by using basic principles and that this can compensate in part for lack of experience. The basic principles are derived from knowledge of the scientific foundations of surgery and paediatrics, many of which already will be known to the student. The following chapters are arranged in such a way that the most important principles are highlighted at the end of each section for easy reference as 'key points'. For many students, several years will have elapsed between the time they learned the basic sciences and their first encounter with paediatric surgery - yet their knowledge of the basic sciences is essential if their examination of children is to be effective. In the following chapters, it will become apparent that examination in paediatric surgery demands the fusion of the preclinical sciences such as embryology, anatomy and physiology, with the clinical science of pathology. In no other branch of medicine is a knowledge of embryology more useful, but unfortunately many students will have forgotten their embryology by the time they commence their training in paediatric surgery. While no attempt has been made to provide a comprehensive description of embryology, those aspects which are useful for clinical purposes have been highlighted.

# Specific Principles of Clinical Examination

The flow chart presented here (Fig. 1.2) provides a plan of how a goal-oriented interview and physical examination can be conducted. Important points in each step are described below.

#### Introduction

A medical interview, like any other social interaction, needs to conform to social customs. First impressions are important, and these can be used to advantage. The style adopted should be a personal one accepting that there are several minirequirements which are necessary, regardless of your personality. It is important to introduce yourself to both the parents and the child by telling them your name and position. Many parents (and even children) will expect you to shake hands, and this may be an important step in the social interaction. It represents mutual recognition that you will be friends and not antagonists during the interview, and helps both parties to relax. Many students and, unfortunately, many doctors in senior positions neglect to tell the parents their name and position. No one wants to be confronted by a nameless 'white coat' (Fig. 1.3): personal accountability is important even when still a student. It is not acceptable to remain anonymous through lack of social confidence; such action may insult the child and parents and make rapport difficult to achieve.

#### **Gain Rapport**

This is an extension of the introduction and is directed primarily at gaining the friendship and confidence of the child. If the child can be spoken to in a relaxed fashion, the parents will be reassured that you are competent and comfortable 4 1 General Principles

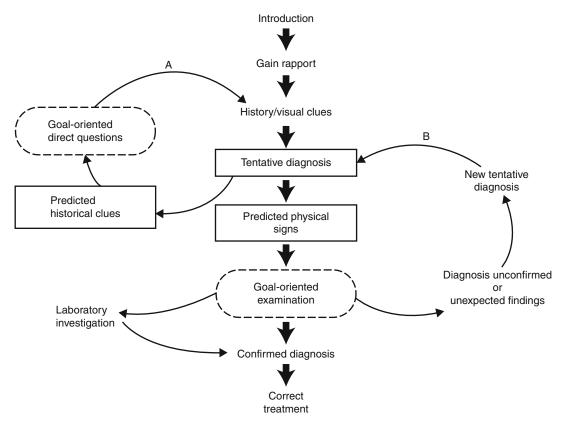


Fig. 1.2 The process of the goal-oriented examination



Fig. 1.3 Beware the nameless/faceless man

with the situation. It is important not to rush this step, because once the child is happy to cooperate, the subsequent physical examination is made much easier. Young children may have a fear of the unknown, fear of separation from parents and

fear of having an injection or 'needle', often superimposed on feelings of anxiety and malaise if unwell. Children must be given an adequate opportunity to adjust themselves to their new surroundings. Any concerns that children express should be taken seriously and discussed openly, and the explanations which are given in response to these concerns should be understandable and reassuring. This will help them to relax and cooperate. Toys are distracters for toddlers, and it is useful to be able to produce a toy with which the child might like to play. As long as their taste in toys is judged accurately, the manoeuvre will be a success and they may even be impressed! This will gain their confidence rapidly. Pleasant talk and friendly gamesmanship will usually overcome the fear expressed by the terrified 4 year old clinging to mother!

When a child is undressed and lying on a couch awaiting examination, first physical contact



Fig. 1.4 Gain the child's confidence and trust

with that child should be in a manner which will allay anxiety.

It may be appropriate to touch the child gently while still talking to the child or the parents (Fig. 1.4). The child must be informed always of what the next stage in the examination involves. Uncomfortable procedures, such as throat or rectal examination, should be performed last.

#### **History Plus Visual Clues**

Older children should be allowed to describe their own symptoms. To be able to take a good history from a child can be a challenge as well as being informative. In allowing children an opportunity to participate, an assessment of their level of intelligence and social maturity can be obtained while boosting their own self-confidence. Where children are capable of relating the history, the entire interview should be conducted from them, with reference to the parents for confirmation of particular facts, such as dates or times, as appropriate. It is surprising how often useful information is gained by patiently guiding the child

through the history. Knowledge of the intellectual limitations of the child is important when interpreting the story, in order to judge the reliability and significance of each feature described. When the child is too frightened, too ill or too young to provide a history, the information will need to be obtained from the parents. The interaction between children and their parents is often relevant to understanding any psychogenic element in the symptoms, particularly if the complaint is one of abdominal pain. In order to lessen the level of anxiety, children must always be informed of what is going to happen next, whether it be in terms of further treatment, investigation or hospital admission.

While listening to the presenting complaints, the clinician must begin to formulate the first tentative diagnosis or a short list of differential diagnoses. This is in preparation for clues which may be waiting later in the history or during the clinical examination. Visual clues should be sought during this early phase of the interview. Much can be gained from studying the appearance or behaviour of the child long before the physical examination begins.

#### **Preliminary Diagnosis**

The goal of the interview is to reach a tentative or preliminary diagnosis. If correct, this eliminates innumerable irrelevant alternatives even before the physical examination is conducted. In addition, it acts as a mental 'signpost', directing the interview along a certain path by allowing predictions to be made for a particular diagnosis about other features which might be present in the history.

#### **Predicted Clues from the History**

The predicted clues from the history are derived directly from the preliminary diagnosis and enable the child and parents to be cross-examined about specific features for which you are looking. Some general background information may be required as well, such as maternal, perinatal or

6 1 General Principles

family history, but it is important not to become too preoccupied with these areas unless they are directly relevant to the problem. It is easy to become distracted collecting unnecessary information.

Sometimes, cross-examination will reveal clues which suggest an alternative diagnosis. Should this occur, reappraisal of the evidence already obtained may allow formulation of a revised diagnosis. Further direct questions may then be required to document the predicted features of the new diagnosis. This process is represented by circle A in the flow chart (Fig. 1.2). It is reasonable to proceed around the circular path as often as is necessary to reach a firm preliminary diagnosis. Obviously, with greater experience, fewer circuits will be needed before a confident diagnosis is made.

#### **Predicted Physical Signs**

The preliminary diagnosis allows prediction of the physical signs which should be sought on examination. These signs may be positive or negative because sometimes it is essential that certain important alternatives are eliminated.

#### **Goal-Oriented Examination**

The first step in any examination is to perform a screening check of the entire body, concentrating on areas not directly related to the likely diagnosis. This collects important background information and on occasions may reveal a surprise. If it is done first, it is less likely to be forgotten. After the relatively brief general examination, the organ or system in which the predicted physical signs are to be found is examined. In this way, the prelimi-

nary diagnosis is either substantiated or refuted. There is a greater chance of elucidating subtle signs and of detecting other more obvious signs which are rarely elicited, if the examiner is looking for particular features. A good example of a rare but important sign is perineal anaesthesia; it is obvious when sought but would never be noticed in a 'routine' examination, yet it is an essential clue to the diagnosis of a neurogenic bladder.

The goal-oriented examination will usually confirm the preliminary diagnosis if the hypothesis is correct. With the diagnosis correctly ascertained, management can begin immediately. Occasionally, confirmation of the diagnosis depends on further corroboration by laboratory investigations, such as imaging or blood tests.

#### **Diagnosis in Doubt**

If the findings of goal-oriented examination do not support the preliminary diagnosis, or if unexpected signs are found, it is necessary to reconsider the diagnosis. This is shown in circle B. If an alternative diagnosis seems more probable after the initial findings, the parents and child must be questioned again for further clues in the history which might support a new diagnosis (circle A). Subsequent physical examination is then directed at new predicted physical signs derived from the new tentative diagnosis.

With greater experience, less time will be spent backtracking (circles A and B) and progression to the diagnosis will be rapid. All that is needed to master this technique is (1) some knowledge and (2) an idea of possible diagnoses. If either of these prerequisites is missing, the clinical evidence will not be recognized, that is, relevant symptoms and signs will be missed, or their significance will not be appreciated.

Further Reading 7

#### **Golden Rules**

- A 'routine examination' finds nothing because you only see what you are looking for.
  - Even obvious visible or palpable signs will be missed if you do not have a possible diagnosis in your head, from which the visible and palpable clues can be predicted.
- 2. Use the subjective nature of the mind deliberately by trying to think of a possible diagnosis (historical and visual clues), and then actively search for the evidence like Sherlock Holmes.
- 3. Because children are basically healthy (no degenerative diseases that accumulate with age), Ockham's razor applies: no matter how many signs and symptoms there are, there is usually only one disease or underlying cause.

#### **Further Reading**

- Fulginiti V.A. (1981). Pediatric Clinical Problem Solving. Baltimore: Williams and Wilkins.
- Green M. (1986). Pediatric Diagnosis. Interpretation of Symptoms and Signs in Different Age Periods, 4th edn. Philadelphia: Saunders.
- Hutson JM, O'Brien M, Woodward AA, Beasley SW (2008) Jones' Clinical Paediatric Surgery. Diagnosis and Management, 6th edn. Blackwell Publishing, Oxford
- Beasley SW, Hutson JM & Myers NA, (1993) Pediatric Diagnosis: An Illustrated Guide to Disorders of Surgical Significance. London: Chapman Hall Medical.

#### Is the Baby Normal?

Some knowledge of the aetiology and patterns of congenital malformation is helpful to answer the question, 'Is the baby normal?' This knowledge enables the clinician to perform a more specific 'screening' examination of the neonate and anticipates the physical signs of those abnormalities which may be present.

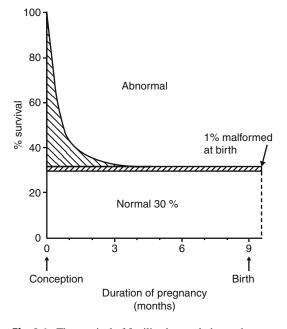
#### **Embryology**

# How Common Are Congenital Anomalies?

Nearly, two thirds of all pregnancies are affected by serious abnormalities which lead to spontaneous abortion in the first 3–4 weeks of gestation, often before the pregnancy has been confirmed. Half of these have a chromosomal abnormality. Almost 10 % of pregnancies abort in the embryonic stage, from inborn errors of metabolism or gross malformations. The surviving fetuses now represent about 30 % of the original number of fertilized ova, but a few still harbour a defect such that 3–4 % of babies born have an abnormality (Fig. 2.1).

# What Types of Abnormality Are There, and When Do They Occur?

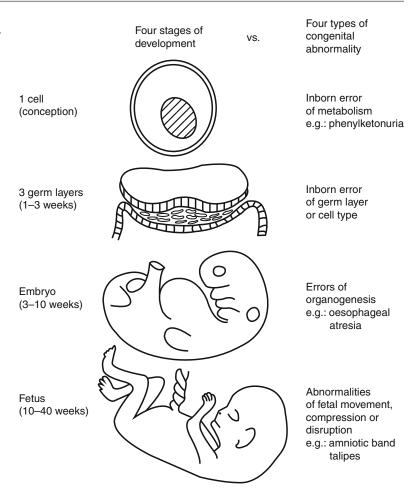
There are four main groups of congenital lesions: inborn errors affecting the fertilized ovum, abnormalities occurring at the time of the three



**Fig. 2.1** The survival of fertilized ova relative to the normal duration of pregnancy

germ layers, abnormalities of organogenesis and defects in fetal movement or compression (Fig. 2.2). Genetic or chromosomal anomalies are present from the one-cell stage, and cellular or germ layer defects occur between 1 and 3 weeks after conception. These two groups account for the enormous drop in survival during the early weeks of pregnancy. Only a small percentage of conceptuses with these defects survive to birth. Between three and 10 weeks' gestation,

**Fig. 2.2** The four stages of development and the types of congenital anomaly which occur at each stage (Adapted from Cohen 1982)



the basic shape and organs of the embryo form and this is when most surgical malformations arise. Anomalies occurring during embryogenesis may be caused by innate genetic defects or by extrinsic teratogens (e.g. rubella or other congenital infections, x-rays, drugs, chemicals or maternal dietary abnormalities).

Knowledge of the causation of many surgical malformations is poor because the embryonic stage is the most difficult phase of pregnancy to study. Despite this limitation, there are several important principles of surgical anomalies which are essential if the clinical situation at birth is to be understood:

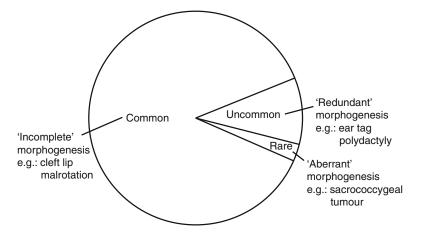
1. Anomalies are often multiple because several organs may be sensitive to the same teratogenic

- influence. If one major anomaly is present, look for others.
- 2. Multiple anomalies are related to each other in time or space. Different parts of the body may be sensitive to a particular extrinsic teratogen or mutation because they are all undergoing rapid cell division and morphogenesis at the same time. Also, several organs in the same part of the body may be affected by a 'field' anomaly of morphogenesis. These defects occur with mutations in genes controlling embryonic segmentation (e.g. Hox genes).

A good example of time-related defects is congenital rubella: the group of organs involved depends on the age of the embryo at infection. The heart, eyes and teeth will be affected at

Embryology 11

**Fig. 2.3** The different types of abnormal morphogenesis that are possible



6 weeks, while at 9 weeks, the ear will be involved, leading to deafness, but the eye is spared.

The "VACTERL" association is a further example of time-related defects, where the following abnormalities are seen in combination: V – vertebral anomaly, for example, sacral agenesis, hemivertebrae

A – anorectal anomaly

C – cardiac anomaly

T – tracheo-oesophageal fistula

E – (o)esophageal atresia

R – renal anomaly

L – limb anomalies, for example, deficiency or atresia of the radius

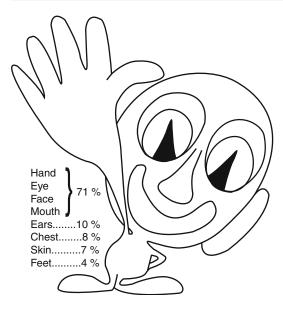
Imperforate anus often provides an example of space-related anomalies since it is commonly associated with other pelvic abnormalities, such as sacral agenesis, deficiency of sacral nerves and pelvic floor muscles and urogenital anomalies.

3. The genetic control of morphogenesis is a multi-tiered hierarchy within which only certain abnormalities are possible and only a few are common. In other words, there is not an infinite array of possible defects, but only a small number of common lesions sufficiently compatible with survival to reach birth. One never finds a baby with antlers(!), but pre-auricular skin tags do occur. The commonest form of abnormal morphogenesis is incomplete development

(Fig. 2.3). This is a normal process in which only the timing is disrupted. Less common is redundant morphogenesis, where the normal process has been partially or completely duplicated. Truly aberrant morphogenesis is rare because the abnormality will usually lead to death well before birth.

# Does It Matter If the Baby Is Dysmorphic?

Yes! Where one major anomaly is observed, the presence of others must be sought. Dysmorphism may indicate that the child has a number of malformations which may be well enough known to be called a 'syndrome'. A syndrome is a collection of features or defects which is recognized as belonging to the one entity. Individual syndromes are rare and may carry unwieldy eponymous names which make them an anathema to the average student or clinician, although obsessive/compulsive learners seem drawn to them. Their real importance is that definitive diagnosis of the syndrome will allow other features of the syndrome not yet identified to be actively sought, usually after consultation with a geneticist or database. Moreover, the prognosis will be predicted more readily than in complex cases without recognized syndromes, since outcome usually has been well defined by previously reported cases. This information is essential in the



**Fig. 2.4** The relationship between recognizable minor anomalies and parts of the body

perinatal period so that the parents can be given a realistic outline of the future. The inevitable fatality of certain syndromes may enable proper discussion about the most appropriate treatment to be offered. In many cases, accurate syndrome identification also permits the cause and risk to future pregnancies to be determined. This can be supported by specific tests in some cases where the genetic defect leading to the syndrome has been identified.

# How Do I Know If the Baby Has a Syndrome?

The principles required by the average clinician are simple, although consultation with an expert colleague is advisable for confirmation. Where a baby has several obvious malformations, the clinician should be alerted to the possibility of a syndrome being present and seek further evidence of this. The areas of the body in which minor abnormalities occur frequently are easily accessible in the neonate and should be carefully scanned (Fig. 2.4). Facial features obviously need to be assessed in the context of the appearance of

the parents. Minor anomalies are important because their presence has a strong correlation with the existence of major malformations.

The finding of a number of minor anomalies in addition to major defects adds weight to the possibility that a syndrome is present.

It is important that the clinician is not daunted by a lack of detailed knowledge of individual syndromes. A search for time-related internal defects, space-related defects and minor anomalies of the face and hands will usually yield significant information, even when performed by inexperienced examiners.

#### **Surgical Assessment of the Neonate**

This section describes those aspects of the general clinical assessment of the neonate which are commonly relevant to the infant with a surgical condition. It does not include a full description of the medical or neurological examination, but concentrates on features which surgeons may be required to assess, or upon which they may be asked to provide an opinion.

The examination of a neonate should be performed under a heat lamp or in a warm quiet environment appropriate to the infant's needs, to avoid heat loss. Lighting must be adequate and the reflected light should not obscure subtle changes in skin colour. The entire surface of the baby must be observed, but care should be taken to avoid excessive handling of the sick infant. Much information can be obtained by careful observation with minimal disturbance to the infant.

#### **General Examination**

Many abnormalities can be detected at birth by simple clinical examination provided that a systemic approach is adopted, such as outlined in this section.

First, look for obvious major anomalies which require immediate measures to avoid unnecessary morbidity, for example, a child with gastroschisis should be wrapped in plastic food wrap to prevent excessive evaporative heat loss. If there are no external abnormalities which require immediate attention, commence examination by observing the overall posture and activity of the child. Does the appearance of the infant match the presumed gestation? Next, observe the infant breathing. Is there tachypnoea, respiratory distress or cyanosis? Look for sternal retraction. Does the infant seem to be 'working hard' with breathing? Once respiratory distress is recognized, its cause should be established promptly. Details of how this is achieved are provided in Chap. 20. It should not be forgotten that distension of the abdomen exacerbates respiratory distress by elevating and splinting the diaphragm, and interfering with the mechanics of ventilation.

The colour of the skin, sclera and mucous membranes should be noted, looking for jaundice, pallor or cyanosis. Turn now to inspection of the head. Feel the anterior and posterior fontanelles and the suture lines. Assess head size (Chap. 10), measure head circumference, inspect the eyes, test the patency of each nostril and look at the hard palate for evidence of a cleft (Chap. 11). Is the tongue normal in size or excessively large? Is there excessive salivation? Study the shape of the face: are the ears in the correct position, and is the distance between the eyes normal? Are the pinnae fully developed, and is there an external auditory meatus? Is the lower jaw normal or underdeveloped?

The neck of the neonate is relatively short but should be inspected for abnormal skin lesions or deeper swellings (Chap. 9), such as a vascular anomaly (e.g. a lymphatic malformation or 'cystic hygroma').

Is the chest wall normal, and is chest expansion symmetrical? Is air entry equal on both sides? Listen to the lung fields for breath sounds. If these are abnormal, percuss the chest. The liver, which is relatively large in the neonate, is percussible in the right lower chest. Dullness of the chest (other than the liver) suggests fluid. Hyper-resonance may indicate a pneumothorax, in which case breath sounds will appear distant on auscultation. If chest movement and breath

sounds are normal, next listen to the heart. The rapid heart rate of the neonate makes auscultation difficult for the inexperienced clinician, but an attempt should be made to detect murmurs and abnormalities in rhythm. Confirm the location of the heart to exclude situs inversus.

Move to the abdomen. Here, three questions should be answered: (1) Is the abdomen distended (or scaphoid)? (2) Are there any abnormal masses palpable? (3) Is there evidence of perforation or peritonitis? Observation of the contour of the ventral abdominal wall of many normal newborn infants provides the experience necessary to judge minor degrees of abdominal distension. Gross abdominal distension is obvious even to the untrained eye and usually indicates bowel obstruction (Chap. 21). Several organs are normally palpable in the abdomen: the liver, kidneys, bladder and less commonly the spleen can be identified in most babies (Chap. 15). The prominence of the vertebral column enables the pulsation of the overlying aorta to be felt easily. Clues to the presence of peritonitis include redness and oedema of the skin of the ventral abdominal wall, tenderness to palpation and increasing distension. A localized perforation may become apparent as a fixed, tender mass.

The distal part of the umbilical cord rapidly desiccates and shrivels up to become the black rigid umbilical stump, but remains attached for about 1 week before separating from the umbilicus itself. Swelling at the umbilicus is common in the first weeks after separation, but usually disappears as the umbilical cicatrix contracts. In most babies, there are three vessels in the umbilical cord: (two arteries and a vein) the presence of only two vessels may be related to other congenital anomalies. Failure of the umbilicus to contract after involution of the fetal vessels will produce an umbilical hernia (Chap. 6).

Next examine the inguinoscrotal and perineal regions (Chaps. 4 and 5). What sex is the child (Chap. 22)? Look for groin swellings indicating inguinal herniae. In boys, identify the position of the testes to determine whether they are fully descended. Remember, inguinoscrotal

descent occurs at about 7 months' gestation such that the testes will not be in the scrotum if the infant is extremely premature. Inspect the foreskin and location of the urethral meatus. A dorsal hood of foreskin and chordee is suggestive of hypospadias.

In both sexes, look for the anus. Two questions must be answered: (1) Is it in the correct position? (2) Is it patent? If the anus is absent in the male, carefully inspect the median raphe of the perineum, scrotum and ventral surface of the penis for a fistulous opening which may be stained black with meconium. In the female, part the labia to inspect the vestibule for an opening. Where the anus is anteriorly displaced, its patency must be established (Chap. 21).

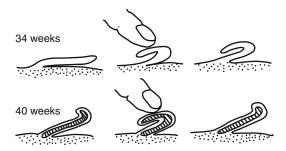
Examine the limbs for congenital dysplasia of the hip, club foot and other orthopaedic anomalies (Chap. 13). Count the fingers and toes. Inspect the skin for lesions: certain hamartomata are not present at birth but become apparent in the first weeks of life as slightly elevated red or purple lesions (e.g. strawberry naevi), whereas others (e.g. port wine stains) are present at birth and do not change. Finally, turn the infant over to examine the back. Run your fingers down the spinous processes and confirm the presence of the sacral segments and coccyx. Look for midline swellings and sinus openings. While the infant is held prone in your hand, test for muscle tone and posture.

#### **Assessment of Gestational Age**

The gestational age is an important factor in determining the infant's ability to adapt to the extrauterine environment. The best estimate of age comes from the mother's dates (when known) or from antenatal ultrasonography. Clinical assessment of the stage of development provides an adequate but less accurate estimate of gestation, but is important in confirming the mother's dates. Knowledge of the birth weight alone is inadequate as it is unable to distinguish infants with intrauterine growth retardation from those with prematurity. It is important to distinguish these two groups as they have different neonatal problems and requirements. Clinical assessment of gestational age is only approximate, but if the

Table 2.1 Assessment of gestation

Ears	Cartilage
	Form (incurving)
Breast	Breast tissue
	Nipple and areola
Sole creases	
Skin	Thickness
	Fat deposition
	Desquamation
Genitalia	Testes
	Scrotum
	Labia majora and clitoris
Vernix	
Hair	
Skull	
Posture	Resting posture
	Recoil of extremities
	Muscle tone (horizontal suspension)



**Fig. 2.5** Assessment of ear cartilage development. At 34 weeks, the pinna will remain folded, while at 40 weeks, the cartilage provides elastic recoil

guidelines described below are followed, it should be possible to determine the gestation to within about 3 weeks (Table 2.1), an error which is entirely acceptable from the therapeutic point of view.

#### **Ears**

Two features of the ear make it useful in the assessment of gestational age: (1) the amount of cartilage present and (2) the form of the pinna.

Before 34 weeks, there is no cartilage in the pinna and it feels thin and soft and will stay folded if deflected forwards. After 34 weeks, increasing cartilage is laid down, making the ear more elastic so that by term, it stands away from the head and, if folded forwards and released, springs back to its original position promptly (Fig. 2.5). Incurving of