LEARNING OUTCOMES

By the end of this chapter you should:

● Understand the causes of learning disability, attention deficit/hyperactivity disorder (ADHD), cerebral palsy and communication disorders
● Understand the principles of management of learning disability, ADHD, cerebral palsy and the different types of language impairment and communication disorder
● Understand the concept of the autistic spectrum and what it includes.

You should also take the opportunity to ensure that:

● You can make an initial assessment of a child with learning difficulties, ADHD, delayed walking and cerebral palsy, and delayed language development
● You are aware of the different ways of assessing language impairment and communication disorder
● You are aware of appropriate management.

Introduction

The prevalence of physical and multiple disabilities in children is estimated to be approximately 10–20 per 1000. Chapter 18 describes the concepts and causes of disability, and emphasizes that its management requires a multidisciplinary approach, often focused on a Child Development Centre. This chapter concentrates on the causes, investigation and management of common specific neurodisabilities of childhood. Although each disability is considered separately, multiple disabilities in the same child are common.

Learning disability

Problem-orientated topic: the slow learning child

Thomas is a 3-year-old boy who is referred because of suspected developmental delay. His vision and hearing are normal. There is no family history of learning disability, fits or serious illness. His parents are not
related. Thomas was born at term weighing 3.5 kg, following a normal pregnancy. He had no neonatal problems. There is no relevant past medical history and no evidence of fits. He smiled at 8 weeks, sat by 8 months and walked at 15 months. He has had no feeding difficulties. His early social interaction was normal. On examination he is not dysmorphic. His head circumference is on the 70% centile. He has no neurocutaneous lesions; his gait, fundi and deep tendon reflexes are normal, and his plantar reflexes are down-going. Arm and leg tone is normal and symmetrical. He shows good eye contact, early turn-taking and uses his index finger to point for a drink or food. The only word he uses is ‘Mum’, with some other early babble.

Q1. What is the definition of learning disability?
Q2. How do you make the diagnosis of learning disability?
Q3. What are the pros and cons of investigating a child with a learning disability?
Q4. What investigations would you perform, if any?
Q5. How would you manage this child and family?

Q1. What is the definition of learning disability?

A child or young person has a learning disability if he or she has ‘a greater difficulty in learning than the majority of children of the same age’ (Box 29.1). The International Statistical Classification of Diseases–10th revision (ICD-10) defines it as ‘a condition of arrested or incomplete development of the mind which is especially characterized by impairment of skills manifested during the developmental period contributing to the overall level of intelligence, i.e. cognitive, motor and social abilities’. This is not a helpful definition for parents.

For research and study it is sometimes necessary to have a definition that can be measured. Average intelligence quotient (IQ) is 100, with a standard deviation (SD) of 15. Learning disability may then be defined as > 2 SD below the mean or the ICD-10 definitions:
- Mild IQ 50–69
- Moderate IQ 35–49
- Severe IQ 20–34
- Profound IQ < 20.

In practice, people in the UK often use the definitions shown in Table 29.1.

Q2. How do you make the diagnosis of learning disability? (Box 29.2)

Mild/moderate learning disability
While children with SLD often have associated problems such as cerebral palsy, those with MLD often have no other problems. Many will be the tail-end of the normal distribution; others will have learning difficulties as a result of environmental factors (lack of early opportunities or iron deficiency) and be functioning below their genetic potential; some will have an identifiable remediable cause such as vision or hearing problems; some will have an intrinsic problem such as neurofibromatosis or a chromosome abnormality.

We will use the abbreviations ‘SLD’ for severe learning disability and ‘MLD’ for mild/moderate learning disability.

Some psychometric tests
The following two tests are commonly used by paediatricians. Both require training and special equipment:
- Griffiths. This has been standardized on 0–8-year-old British children but relies on parental reporting; many items are timed. It has recently been updated.
- Bayley II. This scale (age range 0–42 months) is used to assess developmental age and has been standardized on American children.

Q3. What are the pros and cons of investigating a child with a learning disability?
Q4. What investigations would you perform, if any?
Q5. How would you manage this child and family?
All children with significant learning difficulties should have at least some paediatric assessment to contribute to identification of special educational needs.

When seeing a child referred from school with MLD enquire about:
- History
- Birth
- Progress from birth
- Family
- Other concerns
- Hearing and vision
- Behaviour
- Poor general health
- Time off school, leading to under-achievement
- Whether children are working at their best in school
- Epilepsy (absences, minor status).

Look for dysmorphism or other clues to aetiology.

**Severe learning disability**

It is important to ask or examine for:
- Genetic abnormalities:
  - Dysmorphism
  - Malformations
- Metabolic defects:
  - Failure to thrive
  - Hypotonia
  - Consanguinity
  - Recurrent unexplained illness (especially anorexia and vomiting)
  - Loss of skills
  - Coarse facies
  - Ocular abnormalities
  - Macro- or microcephaly
  - Family history of unexplained illness or death
- Brain malformation:
  - Abnormal skull
  - Focal deficit
  - Loss of skills
  - Micro- or macrocephaly
  - Seizures
  - Visual abnormality

**Syndromes**

These are more likely in children with SLD but should be considered in all children with learning difficulties. In children with SLD, about one-quarter have a chromosomal disorder; 80–90% of these have Down’s syndrome. The next most common disorder is fragile X syndrome (p. 32).

There are now over 2000 syndromes and the number continues to increase. From a practical day-to-day perspective they fall into two broad groups: the more common or easily recognized syndromes, such as Down’s, Edwards’ and Sturge-Weber; and others with a number of abnormal features not immediately recognizable as a syndrome but in whom it is possible to make a diagnosis.


**OMIM (Online Mendelian Inheritance in Man) dysmorphology database**

**Developmental regression**

If there is progressive loss of skills (Ch. 28) it is important to consider:
- Hydrocephalus
- Poorly controlled epilepsy
- Metabolic disorder/neurodegenerative disorder
- Rett’s disease
- Infection, particularly in an immunocompromised host (e.g. AIDS)
- Vascular problem, e.g. repeated minor strokes from moyamoya or sickle cell disease; malformations causing vascular ‘steal’.

True regression can be hard to ascertain because development is taking place at the same time. All children, and particularly those with a learning disability, will sometimes learn something new and then appear to forget it for a while.

**Reasons to ask for further assessment**

- To obtain an objective assessment of abilities
- To identify strengths and weaknesses that may help with management
- To assess progress
- For the court, such as in cases of neglect
- For research.

**Q3. What are the pros and cons of investigating a child with a learning disability?**

**Pros**
- Treatable cause, e.g. hypothyroidism
- Genetic counselling may be useful
- For prognosis
- The parents may be helped by knowing the cause.

**Cons**
- False positives and false negatives
- Pain and complications of investigations (especially anaesthesia)
- Financial cost.
Q4. What investigations would you perform, if any? (Box 29.3)

Investigations should be performed on the basis of clues from the history and examination. The following investigations may be indicated, particularly in children with SLD:
- Chromosome analysis
- Brain imaging
- Metabolic investigations.

Q5. How would you manage this child and family?

The neurodevelopmental paediatrician’s role is:
- Establishing whether there is a learning disability (usually done with a multidisciplinary team). In younger children it is the health services that are primarily involved in this. In older children it is primarily school-based.
- Identifying the cause. This may be from the history and examination or may include investigation.
- Referral to other professionals as appropriate. These may include:
  - Speech and language therapy (SLT), occupational therapy (OT), physiotherapy
  - Psychology
  - Other medical specialties
  - Education
  - Social services.
- Looking for and managing associated difficulties. There may be problems with hearing, vision, motor function, behaviour or epilepsy. Some are specific, e.g. hypothyroidism in Down’s syndrome.
- Counselling parents. The neurodevelopmental paediatrician may be the initial person to do this, although others may take up the role later.
- Liaison with education.
- Explanation to child and parents of the likely effects of the disabilities.
- Responding to concerns.

In metabolic disorders some pharmacological treatments may have an effect on progress. This is a very specialized area and one that is constantly changing, but it is a good reason for trying to make a specific diagnosis.

Down’s syndrome (Tables 29.2 and 29.3)

Incidence is approximately 1 in 1000 live births. The risk of having a child with Down’s syndrome increases with maternal age, so that for a mother in her twenties the risk is less than 1 in 1000, but greater than 1 in 100 in mothers over 40. However, most babies are born to mothers in their twenties and thirties.

Almost everyone can recognize a child or adult with Down’s syndrome. One of the problems with a well-recognized syndrome is that people can have preconceived ideas about what a child with Down’s syndrome is like. Children with Down’s syndrome can be as different from each other as any other group of children in the population. Some children are able to follow a mainstream curriculum and achieve GCSE passes. Others may never develop language. Some have very limited exercise tolerance, while others achieve sporting excellence.

Genetic types (see also Ch. 9)

Most are caused by non-disjunction in meiosis, resulting in an additional chromosome 21 (47 XY with additional chromosome 21). In 20–25% the extra chromosome is paternal. When Down’s syndrome is caused by trisomy 21, the recurrence risk is about double that of a woman of the same age without a previous history.

Three to four percent result from translocation of material from chromosome 21 on to another chromosome. A parent may often have a balanced translocation (one of their chromosome 21s is attached to another chromosome), but this causes no problem because they have a normal total amount of chromosome material. However, this tagged-on chromosome may be present in a gamete in addition to a normal chromosome 21, giving rise to extra chromosome material in the offspring. There is a greatly increased risk of a couple having a second affected child.

Mosaicism accounts for 2–6% and such individuals are usually affected to a lesser degree.

Diagnosis

If the diagnosis is not made on antenatal screening, it is usually made early in the neonatal period by recognition of the typical features of Down’s syndrome. It may be a midwife, a paediatrician or the parents who first recognize that there is a problem with the baby. There is strong evidence from parents to suggest that disclosure should be made as soon as the diagnosis is suspected, preferably with both parents present. The diagnosis is confirmed by chromosome analysis.

Management

Down’s syndrome has possible effects on all body systems. Management of children therefore needs
A multidisciplinary approach. Of the many other potential problems in a child with Down’s syndrome, there are two that deserve particular mention:

- **Congenital heart disease.** This occurs in 40–50% of babies with Down’s syndrome. All newborn babies should be evaluated, with observation (of feeding etc.), physical examination, electrocardiogram (ECG), chest X-ray (CXR) and echocardiogram.

  - **Atrioventricular (AV) canal defects** (endocardial cushion defects) occur very specifically in children with Down’s syndrome (p. 215). Any newborn found to have an AV canal defect should have chromosomal analysis.
  - **Patent ductus arteriosus (PDA), ventricular septal defect (VSD) and atrial septal defect (ASD)** are also more common in children with Down’s syndrome.
  - Damage to pulmonary vasculature with irreversible pulmonary hypertension can occur much earlier in children with Down’s syndrome than expected from the size of the shunt alone. PDA, ASD, VSD and AV canal defects should be considered for early surgical intervention.

- **Atlanto-axial instability.** Routine cervical spine X-ray used to be recommended but review of X-rays of the same child taken minutes apart could give rise to completely different advice. Spinal cord damage in Down’s syndrome is rare and usually

### Table 29.2 Down’s syndrome

<table>
<thead>
<tr>
<th>Feature</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Facial features</td>
<td>ternary folds, flat nasal bridge, small nose, protrusion of tongue</td>
</tr>
<tr>
<td>Brachycephaly</td>
<td></td>
</tr>
<tr>
<td>Wide hands, short fingers</td>
<td>Simian crease may be present in normal individuals</td>
</tr>
<tr>
<td>Distal tri-radius, clinodactyly</td>
<td></td>
</tr>
<tr>
<td>Single palmar crease</td>
<td></td>
</tr>
<tr>
<td>Wide gap between first and second toes</td>
<td></td>
</tr>
<tr>
<td>Brushfield spots</td>
<td>Spots on iris</td>
</tr>
<tr>
<td>Fine soft hair</td>
<td>Can have alopecia</td>
</tr>
<tr>
<td>Dry, hyperkeratotic skin</td>
<td>Helped by simple emulsifying cream and appropriate bath oil</td>
</tr>
<tr>
<td>Other skin problems</td>
<td>Such as vitiligo, papular erythema, mottled skin (cutis marmorata)</td>
</tr>
<tr>
<td>Hypotonia</td>
<td>Prominent in the neonatal period. Influences motor development. May result in joint dislocation</td>
</tr>
<tr>
<td>Orthopaedic problems</td>
<td>Atlanto-axial instability (see below)</td>
</tr>
<tr>
<td>Cardiac problems</td>
<td>Hip dysplasia/dislocation</td>
</tr>
<tr>
<td>Bowel problems</td>
<td>Dislocation/displacement of other joints</td>
</tr>
<tr>
<td>Infections</td>
<td></td>
</tr>
<tr>
<td>Hypothyroidism</td>
<td>Higher incidence of autoimmune hypothyroidism; important to screen for this</td>
</tr>
<tr>
<td>Leukaemia</td>
<td>About a 10–20-fold increase. Children may cope badly with intensive treatment</td>
</tr>
<tr>
<td>Low fertility</td>
<td>Females are fertile</td>
</tr>
<tr>
<td>Behaviour difficulties</td>
<td>Males often have undescended testes and hypogonadism</td>
</tr>
<tr>
<td>Poor growth and weight problems</td>
<td>Frequently poor feeders in infancy. Later a tendency to become overweight and attention to diet and activity levels is needed</td>
</tr>
<tr>
<td>Presenile dementia</td>
<td>Important for long-term support, as carers may be elderly with an affected young adult</td>
</tr>
</tbody>
</table>

### Table 29.3 Average milestones for children with Down’s syndrome

<table>
<thead>
<tr>
<th>Milestone</th>
<th>Mean age</th>
<th>Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sitting</td>
<td>13 months</td>
<td>6–30 months</td>
</tr>
<tr>
<td>Standing</td>
<td>22 months</td>
<td>9–48 months</td>
</tr>
<tr>
<td>Walking</td>
<td>30 months</td>
<td>12–60 months</td>
</tr>
<tr>
<td>Single words</td>
<td>34 months</td>
<td>12–72 months</td>
</tr>
</tbody>
</table>

40% of children with Down’s syndrome are able to learn to read.
Neurodevelopmental disability

insidious rather than acute. It is important not to frighten parents and cause children to be wrapped in cotton wool and prevented from joining in appropriate activities; however, parents do need relevant information to enable them to recognize symptoms that should be reported. The three most common symptoms are:
- Deterioration/change in gait or manipulation skills
- Neck pain/stiffness
- Difficulties with sphincter control.

Children with any of these should be investigated urgently.

Advice should be given to other regular carers of children with Down’s syndrome and included in medical advice for assessment of special educational needs.

http://www.dsmig.org.uk

Guidelines on surveillance for people with Down’s syndrome

Fragile X syndrome

This is probably the second most common known syndromic cause of global learning disability (about 1 in 1360 males and 1 in 2000 females; a further 1 in 1000 females are asymptomatic carriers). The degree varies from mild to severe in boys, and mild to moderate in girls. The defect in fragile X is now known to be an expansion in a specific DNA triplet repeat (CGG) on the X chromosome.

Physical features include:
- Long face and slightly increased head circumference
- Macroglossia
- Large protuberant ears
- Flattened nasal bridge
- Abnormal dermatoglyphics
- Macro-orchidism
- Infantile hypotonia
- Connective tissue dysplasia (joint laxity and soft velvety skin)
- Aortic dilatation and mitral valve prolapse
- Recurrent otitis media
- Failure to thrive in infancy
- Tonic–clonic or partial epilepsy, temporal spikes on electroencephalogram (EEG)
- MRI scan abnormalities (especially cerebellum).

Psychological features include:
- Variable intellectual impairment
- Language delay
- Social impairments, such as those seen in autism
- Attention and concentration difficulties.

Features are very variable and some are only evident in adolescent or adult life. Clinical diagnosis in a young child is difficult.

Families need careful genetic advice.

Other sex chromosome abnormalities in which learning disability may occur are discussed in Chapter 9.

Neurocutaneous syndromes

These may also cause learning disability (pp. 8–10).

Rett’s syndrome

This presents as a neurodegenerative disorder but is probably a neurodevelopmental disorder. It is sometimes classified with the pervasive development disorders such as autism. Genetic advances in Rett’s have been rapid in recent years. Milder cases and cases in boys are being described. It constitutes about 10% of SLD in girls.

Specific learning difficulties

The Education Code of Practice defines children as having specific learning difficulties when they have ‘significantly’ more difficulty in a specific area than most children of the same age that is not due to general learning disabilities.

ICD and DSM (Diagnostic and Statistical Manual of Mental Disorders) definitions depend on the child having a normal IQ and no other problems, but there is no reason why a child with generalized learning difficulties cannot have specific difficulties in one particular area, over and above their general level of difficulty.

Specific reading disorder

The term ‘dyslexia’ is used, but very loosely, for a wide variety of difficulties at school.

Writing disorder

There is a lot of overlap between reading and writing problems (dysgraphia).

Mathematics disorder (dyscalculia)

Incidence is probably similar to dyslexia and dysgraphia but there are interesting differences:
- Dyscalculia is seen equally in boys and girls (though recent work suggests that dyslexia may also be equally represented).
• It is seen more in fragile X carriers, Turner’s syndrome, phenylketonuria (PKU) and ADHD.
• It is the most common learning difficulty in epilepsy.

**Developmental coordination disorder (DCD)**

Currently, this is the term used most commonly for children with motor coordination problems. A number of conditions can present with clumsiness other than DCD and these need to be excluded because management is different. Evidence of deterioration should be sought at presentation and at reviews.

**Differential diagnosis**

- Muscular dystrophies
- Cerebral palsies
- Brain tumours
- Brain injury
- Hydrocephalus
- Ataxias, such as Friedreich’s ataxia (p. 14)
- Metabolic disorders
- Polyneuropathies
- Seizure disorder (p. 5)
- Vestibular disease
- Tremors and other involuntary movements.

History, examination and, if appropriate, investigation should exclude these diagnoses.

**Presentation**

This is variable and depends on the age of the child, though most patients do not present until school age. There is a mixture of gross and fine motor problems.

Gross motor problems include:
- Awkward gait, ungainly running
- Falling a lot
- Bumping into things
- Poor balance
- Poor balancing on one leg, inability to hop
- Slow (or failure of) learning to ride a bike
- Difficulty learning to swim
- Poor at catching, throwing, batting a ball.

Fine motor problems include:
- Difficulty dressing (clothes on the wrong way round or in the wrong order, difficulty with buttons and zips)
- Feeding messy; difficulty using a knife and fork
- Poor at building with bricks, jigsaws, drawing
- Poor pencil control for writing
- Difficulty using scissors and rulers.

Secondary problems include:
- Behaviour problems
- Poor self-esteem
- School failure.

When a child first presents at the clinic, a general paediatric and neurological assessment is needed, particularly to exclude other causes. There are no specific signs on neurological examination. Assessments specifically for DCD are best carried out by an occupational therapist, but the paediatrician should perform some initial screening.

**Management**

The mainstay of treatment is occupational therapy and physiotherapy. Management can be considered under the following headings:

- Explanation to child, parent and teacher
- Specific advice to parents and teachers to help in specific areas such as handwriting and dressing
- Improving self-esteem
- Specific therapy.

**Problem-orientated topic:**

**disruptive behaviour (ADHD)**

Connor is 7 years old. His mother, a single parent to Connor and his 4-year-old sister, has always struggled with his behaviour. Now things are going very badly at school. Connor has barely started to acquire literacy skills, although he seems a bright child. His disruptive behaviour in class is now such a problem that he is frequently sent home. His class teacher has advised his mother to seek a medical appointment. He was initially slow to acquire language but other milestones were normal. Last week he set fire to his bedroom carpet.

Q1. What is attention deficit/hyperactivity disorder?

Q2. How would you assess Connor for this condition?

Q3. How should you manage Connor and his family?

Q4. What is Connor’s prognosis?

**Q1. What is attention deficit/hyperactivity disorder?**

This is not a new disorder explained by environmental pollutants or ‘made up’ to explain away naughty
Neurodevelopmental disability

Frederick Still described children in 1902 with a ‘defect in moral control’, which almost certainly was ADHD.

Individuals with ADHD have:
- Inattention
- Hyperactivity
- Impulsivity: excessive in the context of age, sex and cognitive ability.

For a diagnosis to be made these symptoms should be:
- Present in more than one situation
- Present before the age of 6/7 years
- Impairing the child’s educational or social functioning.

**Inattention**
There is poor regulation of attention and this is manifest particularly in difficult, imposed tasks that are not immediately rewarding. A child may attend to a video game or watch a TV programme with sustained attention but be unable to concentrate in school.

**Hyperactivity**
This is manifest differently at different ages:
- The preschool child will rush around, jumping and climbing noisily and being unable to settle in play.
- The school-age child may be fidgety, squirming and having difficulty remaining seated.
- The adolescent is restless, with foot-tapping and twiddling, and is unable to sit quietly.

**Impulsivity**
Impulsivity means not thinking before acting; it often results in getting into trouble for being cheeky or reckless. The child may have frequent accidents.

**Secondary problems**
Most children have secondary problems, including:
- Poor self-esteem
- Poor peer relationships
- Poor relationship with parents
- Sleep/wake problems
- Dietary problems (will not settle to eat).

**Epidemiology** (Table 29.4)
ADHD is almost certainly still under-diagnosed in parts of the UK, depending on where the child lives. All studies show a predominance of boys, but this may be over-estimated because boys show more obvious aggressive behaviour and girls have more inattention.

Table 29.4 Epidemiology of ADHD

<table>
<thead>
<tr>
<th>Population</th>
<th>Incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>UK (inner city)</td>
<td>1.5% of 7-year-olds</td>
</tr>
<tr>
<td>UK (general)</td>
<td>0.5–1% (hyperkinetic)</td>
</tr>
<tr>
<td></td>
<td>3–7% ADHD</td>
</tr>
</tbody>
</table>

**Pathology**
Development of the frontal lobes is relatively late and myelination is not complete until adolescence. Neuroimaging studies have been inconsistent but the frontal cortex and its connections, as well as intracerebral connections via the corpus callosum, have abnormal activity. There may be differences in brain volume and size of the cerebellum. Functional magnetic resonance imaging (MRI) shows diffuse and decreased activity when individuals with ADHD undertake tasks requiring concentration. There is abnormal handling of noradrenaline (norepinephrine) and dopamine in the brain. This theory is supported by response to treatment with drugs affecting these neurotransmitters.

**Aetiology**
- **Genetics.** Genetics is the major factor governing whether or not a child has ADHD. It has been estimated that there is between 54% and 98% heritability.
- **Environment.** This also plays a part, particularly maternal depression and social disadvantage. It is likely that there is an interplay of genetics and environment, with environmental factors maintaining or exacerbating ADHD rather than causing it.
- **Central nervous system damage.** ADHD is also more frequent following:
  - Perinatal problems and prematurity
  - Antenatal insults, such as fetal alcohol syndrome or maternal smoking
  - Head injury, especially frontal lobe damage
  - Encephalitis and meningitis
  - Hypoxic episodes, such as drowning and strangling
  - Cerebrovascular accidents
  - Chronic neurological illness, such as epilepsy, metabolic problems (e.g. PKU)
  - Medical treatments, such as cerebral irradiation, anticonvulsants
  - Certain conditions such as Williams’ syndrome, hypothyroidism, tuberous sclerosis, XYY, XXY and fragile X syndrome.
Q2. How would you assess Connor for this condition?

Diagnosis is made by assessing information from a variety of sources. This is time-consuming and more than one clinic visit is usually needed.

History
- Current concerns, with specific examples, onset of problems and situation
- Antenatal and perinatal history for possible risk factors
- Early development: babies may be hyperactive with sleep problems, feeding difficulties, colic, waking early
- Medical problems for risk factors and differential diagnosis
- Educational problems for difficulties in different environments
- Relationships with parents and peers
- Family history
- Social situation, looking for other causes of difficulties
- Possible comorbidities (see below).

Examination
- Physical and neurological examination for associated problems (e.g. clumsiness), other problems (e.g. hearing, vision) and other diagnoses putting the child at risk (e.g. dysmorphism, tuberous sclerosis)
- Mental state looking for poor self-esteem, depression, anxiety
- Developmental assessment: behaviour inappropriate for developmental age.

Observation in different settings is vital:
- During the initial assessment
- In school/playground/nursery/playgroup
- At home (parental report may be adequate).

Structured questionnaires, e.g. Conner’s Scales, play an important role in the screening and diagnosis of ADHD. Psychometric testing is helpful in identifying those children whose primary problem is a learning difficulty and comorbid specific learning difficulties.

Differential diagnosis
- Physical illness
- Drugs (either prescribed or of abuse)
- Attachment difficulties
- Social issues (e.g. family break-up)
- Child abuse (especially if change in behaviour)
- Depression/anxiety
- Hearing problems
- Unrealistic expectations on the part of parents or teachers
- Poor parenting
- Bullying
- Bored bright child
- Learning difficulties
- Sleep problems
- Conduct disorder.

Investigations
These are rarely indicated but you may need to exclude other causes of hyperactivity or inattention, such as hearing loss, epilepsy, thyroid disorders, side-effects of drugs:
- Test chromosomes if the child is unusually tall (XYY) or has learning difficulties (fragile X) or dysmorphism.
- Order an EEG if there is suspicion of subclinical epilepsy or absence epilepsy (poor concentration rarely is absence epilepsy).

Comorbidities
Comorbidity appears to be the rule rather than the exception in ADHD. The common additional problems are:
- Reduced cognitive ability (IQ on average is 5–15 points lower)
- Specific learning difficulties (particularly in reading)
- Delayed language development and poor language skills
- Developmental coordination difficulties (DCD)
- Oppositional/defiant disorder and conduct disorder (ODD/CD)
- Mood disorders (anxiety, depression)
- Obsessive–compulsive disorders (OCD)
- Tourette’s syndrome
- Autistic spectrum disorders.

It is important to identify these to optimize treatment.

Q3. How should you manage Connor and his family?

Information is an important aspect of management. Making the diagnosis and making this known to all involved may, in itself, help the child, parent and teachers to cope with the ADHD:
- Oral and written information should be made available.
- The child should be informed as well as the parents.
- Teachers should be informed about the diagnosis and, if necessary, about what it means.
Support groups
These can be very helpful to parents and child.

Educational measures
Simple suggestions can be very helpful such as:
- Having the child sit near the teacher
- Removing distractions where possible
- Clear, frequent and small rewards and discipline
- Working alone or in small groups
- Addressing any learning difficulties.

Behaviour modification
Positive reinforcement is very important. Children with ADHD often have low self-esteem. Children respond best to a well-structured, predictable environment where expectations and rules are clear and consistent, and consequences are set down ahead of time and delivered immediately.

Medication
Medication is the single most effective approach in severe ADHD.

Stimulant medications (methylphenidate, dexamphetamine) affect the dopamine pathways in the brain but the exact mechanism of action is unclear. They may stimulate areas of the brain that are not functioning properly. They do not affect the underlying pathology but control some symptoms, so that behavioural management can be more effective, school work can progress and social relationships can develop better. They work best in controlling hyperactivity and impulsivity but are less effective in controlling inattention. Methylphenidate, the most frequently used medication, is usually started at a dose of 2.5–5 mg twice or three times a day, increasing by 2.5–5 mg weekly until the desired effect is achieved. A maximum of 20 mg per dose, or 45–60 mg per day, should be used. If there is no effect after 3 weeks at maximum dose, it should be stopped.

Sustained-release products are now available. These have a lower incidence of side-effects and, as they are long-acting, do not need to be given in school.

Side-effects may include somnolence, gastrointestinal effects and rarely liver problems.

Diet
Diet has long been suggested as an important cause of behaviour problems. Additives in the diet worsen hyperactivity but do not cause ADHD. It is worth checking whether the parents have noticed any foods that cause deterioration in behaviour and removing them from the diet.

Sleep
Many children and young people with ADHD have poor sleep patterns and cannot usually stop themselves waking others when they are awake. It is seldom safe to leave such a child unattended for long and families are often very sleep-deprived. This may greatly limit their capacity to cope with their constantly active offspring in the daytime! Although stimulants may cause insomnia in some children, a teatime dose may actually help a child to get off to sleep by calming a racing mind. In other children, the use of melatonin to regulate sleep patterns and quality is very useful.

4. What is Connor’s prognosis?
Some children continue to have difficulties in adult life. Various groups have reported similar findings, with approximately 30% within the normal range as adults, 50–60% continuing to have problems with concentration, impulsivity and social interaction, and 10–15% having significant psychiatric or antisocial problems (depressed, suicidal, drug and alcohol abuse, convictions for assault, armed robbery etc.).

The prognosis is best for those children with ‘pure’ ADHD and worse for those with severe symptoms, comorbidities, and poor family and educational support.

The cerebral palsies
Cerebral palsy (CP) is defined by the Oxford Register of Early Childhood Impairments as:
SIX

A permanent impairment of voluntary movement or posture presumed to be due to permanent damage to the immature brain. Children with progressive disorders and those with profound hypotonia and no other neurological signs (often associated with severe intellectual delay) are excluded. It is an umbrella term which includes a heterogeneous group of conditions and can arise at any point during brain development.

There are three main types:

- **Spastic CP**, which can be divided into diplegia, hemiplegia and quadriplegia depending on areas affected
- **Ataxic CP**
- **Dyskinetic CP**.

### Terminology

See Boxes 29.4 and 29.5.

### Classification

CP refers to a group of disorders. Classification is based upon clinical descriptions of neurological signs. It is commonplace to find mixed patterns with one predominant aspect, e.g. hemiplegia with some involvement of the good side, diplegia with asymmetry in the upper limbs etc.:  

- **Spastic diplegia**. Recent magnetic resonance studies show that the underlying lesion in most cases of spastic diplegia is periventricular leucomalacia (p. 367).
- **Spastic hemiplegia**. Spastic hemiplegia constitutes about 25% of all cases of CP. The cause is usually an infarction within the distribution of the middle cerebral artery (p. 367).
- **Total body involvement CP**. In these cases the brain pathology most commonly originates in the prenatal period and may be due to a variety of abnormalities such as primary cerebral dysgenesis (lissencephaly/pachygyria), early pregnancy infections (e.g. cytomegalovirus (CMV), toxoplasmosis), or vascular malformations and vascular accidents (e.g. hydranencephaly). **Spastic tetraplegia** with bilateral cerebral hemisphere infarction, sometimes with extensive cyst formation (multicystic encephalomalacia) and severe learning difficulties, may occur as a result of brain injury in late third trimester. Prolonged partial asphyxia in a term infant may be the cause (p. 365). Acute profound asphyxia may develop in the third trimester as a consequence of antepartum haemorrhage, cord prolapse or uterine rupture and may lead to damage in the basal ganglia and thalami which may be confirmed on MR scanning. The clinical correlate is the later development of dyskinetic cerebral palsy, often with relatively preserved cognitive function. The contribution of perinatal asphyxia to the overall prevalence of CP is debatable, but most agree an estimate of about 10% of all cases.

**Dyskinetic CP** may also arise due to bilirubin encephalopathy in the neonatal period. These cases were more common in the past, but prevention and improved management of rhesus iso-immunization have resulted in a dramatic fall in the number of cases.

**Ataxic CP** (about 5% of CP) is mainly of prenatal origin. There may be strong familial patterns, with autosomal dominant, X-linked and autosomal recessive modes of inheritance. Sporadic cases are also seen. Children show ataxia, intention tremor and dyskinesia, usually before 2 years of age. Some may achieve independent walking by 4–6 years, although in these cases handwriting...
remains problematic and, in more severe cases, learning difficulties and seizures may complicate the presentation. About 30% show normal or borderline intellectual function. A magnetic resonance study of ataxic CP showed that over 50% were unclassifiable, 23% were genetic, and only 4% (3 cases) may have had a perinatal cause.

**Epidemiology**

Prevalence of the cerebral palsies is about 1.7–3 cases/1000 live births. There may have been a trend of increase in the overall prevalence of CP in children born in the 1970s and 1980s. The main area of increase has been in the most immature babies weighing under 1 kg. The reasons for this increase are unclear but probably relate to dramatic changes in survival of very immature infants.

Another important aspect of epidemiology is survival. Most children with CP now survive to adult life, even when disease is severe. This is having an impact on services for adults as well as children.

### Problem-oriented topic:

**delay in walking**

Matthew is 18 months old and his mother is concerned that he is not yet walking. He is a bright, sociable child, who has several single words. He was born at 27 weeks’ gestation and had a difficult neonatal course.

**Q1.** What questions would you ask to elucidate a cause?

**Q2.** What are the possible diagnoses?

**Q3.** What are the principles of management?

**Q1.** What questions would you ask to elucidate a cause?

An underlying cause may not be apparent, but the following should be considered in history-taking:

- **Prenatal:**
  - Genetic
  - Infection (e.g. CMV, rubella, chorioamnionitis)
  - Toxins (e.g. drugs)
  - Trauma
  - Nutritional (‘placental insufficiency’)

- **Perinatal:**
  - Prematurity (intraventricular haemorrhage/ periventricular haemorrhage/periventricular leucomalacia)

**Q2.** What are the possible diagnoses?

One likely cause for this history is CP and this is confirmed by abnormal physical signs. A familial delay should also be considered, as well as rarer causes such as Duchenne muscular dystrophy (p. 17) in boys.

It is important to remember that everything that looks like CP may not be. Many infants with complex congenital abnormalities (dysmorphic syndromes) will display central motor impairment. These children will require similar services.

**Q3.** What are the principles of management?

This depends upon the stage of the disorder (Fig. 29.1).

Different approaches to treatment have, from time to time, attracted considerable interest and enthusiasm, as well as opposition. Only recently have attempts been made to study the relative merits of each in objective ways. No single approach will suit all children with a particular form of CP. In most centres in the UK staff follow an eclectic approach, deriving therapeutic ideas from a variety of ‘methods’. No study has convincingly shown benefits of one approach over another.

Management involves regular assessment of the child (with parent/carer involvement) and close multidisciplinary working.
Key professionals
- The physiotherapist is responsible for development of motor skills, and assessment for lower limb orthoses and specialized supportive equipment, such as standers and mobility aids. In the early stages physiotherapy is aimed at interrupting the circle of malachievement caused by abnormal muscle tone. The child’s carers are shown methods of handling and carrying out everyday tasks that help this.
- The speech and language therapist plays these key roles in CP:
  - Most importantly, helping with feeding early in life
  - Helping early communication development
  - Help with speech, which may be severely impaired
  - Management of dribbling
  - Provision of communication aids.
- The occupational therapist will assess the need for equipment to facilitate aspects of daily living, e.g. bathing, toileting, static seating, feeding etc., and fine motor skill function, perceptual skills and the use of upper limb orthoses. Adaptations may also be required in the home.

Specialized equipment
- Orthoses. The purpose of an orthosis is to restore the normal distribution of forces acting through the limb, thereby normalizing musculoskeletal relationships and establishing a normal pattern of motion and/or prevention of progressive deformity. Hence children with a persistently equinus foot may wear an ankle orthosis. Other orthoses facilitate hand function. Some children experience upper limb spasticity at night; a night resting splint will hold the hand in a neutral position in children, thus optimizing functional use during the day.
- Special seating, standing and lying frames. These are used to try to maintain good posture and to give the child optimal positioning and support for feeding and play.
- Supportive bracing. This may be needed in some quadriplegic patients to prevent progression of spinal deformity.

Specific drug treatment
Drugs are now being used more widely in CP:
- Botulinum toxin A (BT A). This works by chemically denervating the muscle, allowing it to relax, which may enable improved gait or easier care, for example. Relaxation of the muscle may also enable it to grow better by allowing stretching and thereby reducing contractures. The duration of effect is usually 10–14 weeks, and measurable effects may persist for up to 26 weeks.
- Baclofen. This analogue of gamma-aminobutyric acid (GABA) impedes excitatory neurotransmission at a spinal level. Oral baclofen is rapidly absorbed, but is protein-bound and has poor penetration into CSF because of poor lipid solubility. The half-life is 3–4 hours, requiring regular dosing (3 times daily). Response to oral baclofen is unpredictable; a number of children will show a satisfactory response, with reduction in muscle tone, but others will develop unacceptable side-effects, including somnolence, confusion, difficulties with oral control, ataxia and increased frequency of micturition. Recently, baclofen by continuous infusion has been given by an intrathecal catheter and pump delivery system to achieve higher and continuous CSF baclofen levels. Baclofen is perhaps most useful when there is generalized increase in tone, which would require multiple injections of botulinum toxin, e.g. in the child with severe spastic tetraplegia.

Surgery
The orthopaedic surgeon has a major role to play in management of CP. Orthopaedic surgery may be indicated to improve function, to prevent deterioration, to relieve pain and to facilitate care.

There are two surgical aspects to the management of CP:
- Selective posterior rhizotomy. Two groups of patients are most suitable: children who are of good intelligence, well motivated and sufficiently strong to achieve walking after spasticity is reduced, and severely affected, non-ambulant patients in whom painful spasm can be reduced.
- Single event multilevel surgery with associated gait analysis. When fixed contracture of muscles occurs, surgical release has been required to correct the deformity. The traditional approach has been to undertake soft tissue surgery in a ‘phased’ manner, dealing with one area at a time. Recently it has become clear that this approach of repeated operations, often on a yearly basis, frequently does not improve long-term function. As a result, techniques of thorough pre- and post-operative assessment have been developed, in particular gait analysis. The latter has led to a better understanding of normal gait in children and hence the abnormal gait of the child with CP. Detailed surgical planning is based upon objective rather than subjective information. Gait analysis also allows proper objective review after surgery.
**Associated problems**

Difficulty may arise from motor problems:

- **Feeding difficulties.** Feeding may be a considerable problem, leading to inadequate quantity and quality of intake. Children with spastic quadriplegia or athetoid CP may have such severe feeding difficulties that they fail to thrive. Recurrent aspiration during feeding may lead to serious chest complications, and children with severe CP commonly suffer from significant gastro-oesophageal reflux (up to 70% having oesophagitis). It is important to address positioning and consistency of food, and to consider the need for gastrostomy feeding. A multidisciplinary approach is essential for significant feeding problems and many places will have a ‘feeding clinic’.

- **Drooling.** This is associated with speech and feeding problems and can be a significant cosmetic handicap, as well as being very messy and affecting the skin around the mouth and neck. It is usually due to a problem with swallowing saliva rather than excessive production. Techniques used to help it are:
  - Prompting and rewards for swallowing
  - Positioning and exercises to improve oro-motor function and sensory awareness, now sometimes aided by intra-oral training appliances
  - Medication with anticholinergics to reduce secretions
  - Surgery to direct the ducts further towards the back of the mouth
  - Occasionally, removal of salivary glands
  - Intraglandular botulinum toxin injections.

- **Dislocated hips.** These are an important complication in CP and routine screening by X-ray is needed. Good postural management will help to prevent dislocation.

- **Bowel and bladder problems.** Incontinence may result from learning difficulties, but may be a problem of not being able to get to the toilet in time or undress quickly enough. Constipation is common, particularly in the immobile child and those with restricted diets. It may also be associated with abnormal gut sensitivity and motility. It is important to try to prevent problems by explaining to the parents and child about normal bowel function and giving dietary advice. If constipation occurs, the earlier it is treated, the better.

- **Osteopenia.** The increased risk of bone fractures in children with motor disabilities is linked to reduced bone density. Measures such as weight-bearing, particularly ambulation, good nutrition (especially calcium, vitamin D and magnesium) and sunlight will help.

Other associated problems include:

- **Vision problems.** These are common (50%), particularly myopia, cortical visual impairment and squint.

- **Hearing problems.** These occur in 20–30%, particularly sensorineural deafness. It is also important to look for conductive problems.

- **Learning disabilities.** These are found in all types of CP. Generalized learning difficulties tend to be related to severity of physical problems; however, not all children with severe motor problems have learning difficulties and children with relatively mild motor problems may have significant learning difficulties.

- **Specific learning difficulties.** These are also seen more frequently in CP and can easily be overlooked. Assessment can be very difficult if there are severe motor problems.

- **Epilepsy.** Around 21% of children with CP develop epilepsy, which may be difficult to control.

- **Psychological problems.** These may be due to physical difficulties, or children may have problems directly related to the underlying brain disorder.

- **Educational issues.** Most children will go to mainstream school and need a minimum of help. Some adaptations may be necessary, e.g. ramps, handrails, lifts, special toilet facilities and adapted working surfaces in the classroom.

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**Communication and its disorders**

**Basic science**

When thinking about speech and language development, you must address the different skills necessary for communication. These include the following.

**Attention control**

The child must have adequate listening skills and attention.

**Symbolic understanding**

Words are symbols, so unless children can understand the concept of symbols, they will not understand speech.

**Comprehension**

Does the child understand spoken language?

**Expressive speech**

This is the area of communication most easily identified by both parents and professionals, and so tends to be what people concentrate on in the early stages.
**Phonology**
The development of sounds proceeds largely in the same order in all children, the easier sounds being acquired earlier.

**Oromotor skills**
These depend on normal orofacial development and functioning bulbar innervation.

**Grammar/syntax**
The rules of language (e.g. plurals, tenses, word order) need to be acquired.

**Semantics**
Semantics is about the meaning of words. Children can just learn words by rote and not be able to use them appropriately in context.

**Pragmatics**
Pragmatics is the way in which language is used in the social context. It includes turn-taking, keeping to the subject, selection, context, verbal jokes and negotiation. This is an area that children with developmental language difficulties, and in particular autism, find hard to learn.

**Non-verbal communication**
A lot of non-verbal communication occurs without thought, but in children with autism it has to be taught. It includes:
- Tone of voice, pauses etc.
- Facial expression, including eye contact
- Body posture
- Gesture and signing
- Physical contact.

These become more important in children with speech problems and may be the predominant form of communication in some, such as those with profound deafness.

**Problem-orientated topic:**

**delay in speaking**
Alex is 2½ years old. His parents are concerned that he is still only using a few single words. He has achieved his motor milestones within the average range and has no significant previous medical history.

Q1. How would you assess this child?
Q2. What other information would you seek?
Q3. What investigations would you perform?
Q4. How would you manage this child?

**History**
Ask in particular about:
- Pregnancy and birth history.
- Family history of language problems or learning difficulties. There is certainly a strong genetic component in language disorder and possibly in the normal range of language acquisition.
- Early input. Careg/child interaction is clearly vital.
- Bilingualism. This was thought to be a problem for children first learning language but there is no evidence for this.
- General development. Language delay is often a marker for general delay.
- Any worry about hearing? Has the hearing been checked? Is there intermittent hearing loss or high-frequency loss? Profoundly deaf children will not learn spoken language without considerable help. Some children with mild hearing losses are delayed in their language development.
- Any feeding problems in early life?
- Any problems now with chewing or dribbling?
- Any difficulties with social relationships?

**Observation**
- Is there any clumsiness?
- Observe the child’s attention span.
- Watch children in a free play situation. Do they make good eye contact with their parent and with other people in the room? Do they turn to share enjoyment of a toy with their parent? How does the parent respond? Is there imaginative play?
- What spontaneous sounds, words or phrases do they use?
- Is there good non-verbal communication?

**Examination**
- Do the ears look normal? Undertake a full ear, nose and throat examination (Ch. 5).
- Are there local problems in the mouth? Submucous cleft or problems with tongue movements?
- Assess hearing in your clinic (Ch. 5).
- Associated disorder. Any evidence of a movement disorder, e.g. CP?
- Try to engage the child in some one-to-one activities. Some children may appear to have poor auditory attention. Does this improve when you work one-to-one with the child (as in a child
with an attention disorder) or is the child unable to tolerate this one-to-one direction (raises the possibility of autistic spectrum disorder)? These factors may interplay.

**Q2. What other information would you seek?**

There are a number of aspects of language that need to be addressed separately.

**Listening and attention**

Can the child attend to language?

**Verbal comprehension**

What level of verbal comprehension has the child developed?

- From the history, get particular examples and make sure that they show verbal rather than non-verbal comprehension. Beware the child understanding from context alone. Can the child identify single objects (from objects or pictures)?
- Can the child understand prepositions (in/on/under/behind) and concepts (big/little)?
- How many key words can the child follow? For example:
  - ‘Put the big pencil in the box’ — 1 key word. There are only pencils and a box and it is automatic to put things in boxes.
  - ‘Put the cat under the chair’ — 3 key words.

**Non-verbal strategies**

What non-verbal strategies does the child use to communicate, both for comprehension and expression?

- Pointing and facial expression plus non-speech sound to indicate needs?
- Common gestures, such as pointing, arms up for wanting to be lifted up and waving bye-bye?
- More complex gestures as a means of self-expression?

**Verbal expression** (Box 29.6)

- Pre-speech babbling or only open vowel sounds?
- Some symbolic noise or word approximations: e.g. ‘hiya’ (for ‘here you are’) or ‘brrmm brrmm’ (for car play)?
- How many single words is the child using that Mother understands (not necessarily other people)?
- Are there some learned phrases that are two- or three-word combinations learned together, e.g. ‘all gone’, or has the child started flexible word joining, e.g. ‘teddy gone’ or ‘daddy car’?
- Are longer structures used, e.g. ‘my teddy gone’ or ‘my teddy’s called Joe and he’s going to bed now’?
- Does the child ask questions?

**BOX 29.6 Pitfalls in assessing verbal competence**

Beware of the echolalic child or the child using chunks of ‘late echoing’, which will give the impression of a verbally competent child. This may indicate delayed verbal comprehension or semantic–pragmatic problems.

Beware of children who initially appear to have good expressive language using appropriate social phrases but who demonstrate a lack of variety or range in their speech. These may be children with general delay or with very little language stimulation.

- Can the child tell you what he or she has been doing?
- Does he or she tend to copy what you say?

**Phonology**

This is an area that parents often get worried about, but which has the best prognosis:

- Is it dysarthria (due to abnormal neurology, e.g. CP)?
- Is it dysphonia (e.g. hoarseness due to disorder of the voice box)?
- Is it delay (the way a younger child would talk) or deviance?

**Semantics and pragmatics** (Box 29.7)

This is an area of difficulty that may only become evident as the child develops more language. It is rare for children to have semantic or pragmatic disorders with completely normal early language development:

- Does the child have difficulties following instructions, particularly complicated ones?
- Does the child understand tenses?
- Are conversational skills age-appropriate? Can the child turn-take? Is the content of speech pertinent?

**BOX 29.7 Pitfalls in parental descriptions**

Parents often underestimate their child’s expressive language and overestimate their comprehension.

**Standardized assessments of language**

Numerous standardized tools for language assessments exist and these are described on the MasterCourse website.

**Q3. What investigations would you perform?**

- Hearing should be tested in all children.
- Test chromosomes only if there are other indications.
Order an EEG if there is loss of language or some other reason to suspect epilepsy. Any child with severe language impairment or complex problems should be seen by a paediatrician.

**Q4. How would you manage this child?**

The role of the doctor in the management of language difficulties is:
- To identify and address any possible causes (e.g. hearing loss, submucous cleft)
- To identify and address associated difficulties (e.g. epilepsy).

Management of language impairments lies predominantly with the speech and language therapist (SLT), whose role is:
- To advise parents on how they can help the child
- To monitor young children or those with mild delay
- To work with nursery or school, advising how to help language development
- To work with groups of children who have similar problems, to encourage language development
- To work with individual children, usually for short intensive bursts of speech therapy
- To advise on and implement alternative communication systems.

Details of these alternative communication systems are given on the MasterCourse website.

**Language disorders**

**Definitions**

The terms ‘communication’ and ‘language’ are sometimes used interchangeably. Communication problems encompass a wider range of disorders, including difficulties in non-verbal communication and ability to use language (as in autism). We will discuss language problems other than autism here, although this is not always a simple distinction:
- **Delay** implies that language is slow to develop; it is progressing in a normal pattern but is like that of a younger child.
- **Disorder** implies that language contains elements that are not part of normal development.
- **Impairment** is the term that is currently preferred and encompasses both delay and deviance. Specific language impairment is used for children who have isolated language impairment with non-verbal learning skills at a higher level. However, studies show that children with ‘specific’ language impairment have a high incidence of other neurodevelopmental disorders.

**Prevalence**

Studies on prevalence vary, depending on definition and whether children with intellectual difficulties are included. About 5–7% of children have significant language difficulties without other learning difficulties. Around 1% have severe persisting difficulties.

There is a wide variation in normal development of language. This can make it hard to decide when there is an abnormality, particularly when there is delay rather than deviance and it affects verbal expression and phonology. Many (though not all) of these children will have corrected themselves by school age or soon after.

**Children with profound and multiple learning difficulties**

The SLT will be part of a team caring for these children. He or she may be involved initially with feeding difficulties and can anticipate language problems.

Key elements for aiding communication in children with profound and multiple difficulties are:
- Using all senses to give messages about the child’s world: hearing, vision, touch, taste, smell
- Helping the child to learn to control the environment at a basic level: making simple choices, turn-taking
- Helping the child to develop relationships: limiting numbers of professionals involved directly with the child and working through other people
- Using augmentative communication systems.

**Education**

Fortunately, most language difficulties have resolved by school age or soon after. Some children will go on to have reading difficulties.

A small number of children need specialist educational input, which may be available locally at nursery or primary school, but by secondary school age the numbers are so small that the child may need to travel some distance and occasionally even board at a special school to obtain this level of specialist education.

**English as a second language**

Special problems may arise for children whose first language is not English. These include:
- **Late recognition.** People assume that there are no problems, or that any difficulties in English result from it being the child's second language.
- **Assessment.** Is there a problem in the first language or is the problem only in English? Trying to assess this means having an interpreter.
- **Treatment.** Should this be in the first language? Can this be achieved?
Classification

There are problems with classification of communication disorders; children often overlap categories or change from one to another. There is also difficulty because of the wide spectrum of normal development. A practical classification can be used to assist assessment and investigation:

- Disorders affecting speech production:
  - Neurological problems, e.g. CP
  - Structural problems, e.g. cleft lip and palate (‘tongue-tie’ rarely, if ever, causes speech problems)
  - Dysphonia, e.g. abnormalities of the vocal cords
  - Dysfluency (stammering), a frequently normal stage of development between 2 and 4 years
  - Elective (or selective) mutism

- Specific language impairments:
  - Expressive language delay
  - Articulatory dyspraxia, possibly associated with feeding difficulties
  - Difficulties in producing sounds accurately, in comprehension or in finding a word
  - Semantic–pragmatic difficulties. These children may have had normal early development of language but often have had delay or deviance that resolved. Difficulties become apparent later with inability to maintain a conversation, ‘getting the wrong end of the stick’, going off at a tangent, misunderstanding rules of conversation, or difficulty with puns and jokes. There are often problems with social interaction, which may overlap with the autistic spectrum.

- Children who stop talking:
  - Usually associated with loss of other skills, e.g. neurodegenerative disorders

- Impaired language and social interaction:
  - These are considered in the section on autistic spectrum disorders.

Autistic spectrum disorders

Problem-orientated topic: the isolated child

Robbie is 3 years of age and has been referred by his GP because he has recently started nursery school and his teacher has expressed concern that he may be autistic. Robbie is his mother’s third child and he has always acted differently to the other two. Since the age of 6 months he stopped making eye contact and is now a very solitary child, preferring his own company to that of his family. He spends hours spinning a toy top and becomes inconsolable if interrupted. His speech development is immature and he tends to echo phrases made by his brothers.

Q1. What are the three core impairments seen in autistic spectrum disorders?
Q2. How would you assess this child?
Q3. What alternative conditions do you need to consider?
Q4. What investigations should be considered?
Q5. What are the advantages/disadvantages of finding a diagnostic label for Robbie and his family?
Q6. What are your management options?
Q7. What is the prognosis for this condition?

Q1. What are the three core impairments seen in autistic spectrum disorders?

Autism is a specific type of communication disorder, which has some overlap with semantic–pragmatic disorders of language (p. 41) and also with other types of neurodevelopmental disorder. The present concept

Aetiology

Most children with language difficulties not linked with other major disabilities have no identifiable cause for their problems. Children with language disorders are known to have an increased family history of language disorders and twin studies suggest strong heritability.

Associated problems

- Epilepsy has a higher incidence in children with language disorders. This supports the idea that language disorders are due to problems with brain development.
- Left-handedness (especially in girls) is often noted but not always confirmed. The significance is unclear; it may sometimes be a pathological left-handedness.
- Clumsiness is found in 90% of children with severe language disorder. Studies and clinical observation confirm this link.
- Educational difficulties are complicated and depend on the type of language impairment.
is of an ‘autistic spectrum’, in which the three core features are:
- Impairment of social interaction
- Impairment of communication
- Rigidity of thought.

It can be useful within this spectrum to define the type (classic autism, high-functioning autism, Asperger’s syndrome) while accepting that some children do not fit any one of these labels but still show significant features within the spectrum.

**Impaired social interaction**
Eye contact may be present but is abnormal in its nature (fixed and staring, held too long). However, there may be good reasons for a child to have poor eye contact in the clinic, such as shyness, embarrassment, willfulness.

Lack of cuddliness is another popular concept, but autistic children may be willing to be cuddled and even give cuddles back, especially with their parents, though the parents may describe it as ‘on the child’s terms’ or ‘too intense’.

Key indicators of impaired social interaction are:
- Lack of sharing and directing attention
- Poor recognition of others’ affect
- Poor understanding of social situations.

**Impaired communication**
In classic autism there will be delayed development of communication, though many autistic children develop some language. About 33% of children with autism develop some early words and then lose them. It is often apparent that they had social interactional problems from the outset and/or never used their words really communicatively. Only 7–8% of autistic children have a setback in language development following completely normal development in the first couple of years with acquisition of two-word phrases. Children with Asperger’s syndrome may have normal language development, but content and use of language may be unusual.

**Rigidity/stereotyped thought and behaviour**
These include the hand flapping and turning in circles seen in classic autism and the intense narrow interests of children with Asperger’s syndrome. Some children have no imaginative play; others will have restricted imaginative play, such as going through the same routine, probably copied from a video.

Q2. How would you assess this child?
With practice it becomes relatively easy to identify most children within the spectrum in the informal clinic situation and many parents have already suspected the diagnosis. In some children it is less easy, and it is important to have reliable methods of assessment so that the diagnosis can be made or excluded with confidence and backed up with evidence.

**History**
Diagnosis depends greatly on history, so it is important to have a framework that will cover the triad of impairments. In addition a standard medical, developmental and family history may indicate a different diagnosis, associated problems or a possible cause for the autism.

There are formal scored interviews for the autistic spectrum, which have diagnostic cutoff points and may also give an indication of where in the spectrum the child lies.

**Examination**
This may identify possible causes, including:
- Dysmorphism
- Neurological abnormality, including head size
- Skin markers (should include examination under Wood’s light for depigmented patches, p. 9).

Informal observation in the clinic setting will often give clues as to diagnosis, but children may act fairly normally in this structured one-to-one situation, so it is helpful to observe them in different settings and over time.

**Assessing where on the spectrum a child lies**
The formal scored tools will help with this. Beware of being too precise early on because this may change as time goes by and as you obtain more information about the child.

**Assessing the child’s strengths and weaknesses**
Each aspect of the child’s difficulties is assessed:
- Its nature
- Its degree
- Whether it is primary or secondary
- Whether it is changing over time.

Other aspects can be assessed, such as:
- Rigidity/routines
- Motor abilities
- ‘Dangers’ of special interests
- Motivation/reliability
- Obsessional–compulsive behaviours
- Insight
- Depression/anxiety
- Temperament
- Support systems.
Children are presenting younger with possible autistic spectrum disorder and this may present difficulties around stability of the diagnostic label over time.

Q3. What alternative conditions do you need to consider?

Social impairment may be secondary to other disorders:
- Learning difficulties, leading to social immaturity
- Dyspraxia, leading to invasion of other people’s social space
- Psychopathy (p. 52)
- Shyness, leading to social awkwardness
- Conduct disorder, bullying, abuse, depression, which may all lead to abnormal social relationships
- Semantic–pragmatic disorders (p. 41), leading to abnormalities in social relationships; there is overlap between these and autism
- Secondary social impairment, which may occur as a result of ADHD (p. 33), depression, bullying or abuse
- Rett’s syndrome (p. 22), but the relationship to autism is debatable; certainly, these children develop features of severe autism but also tend to lose them as the condition progresses.

Comorbidity

Children with an autistic spectrum disorder have a greater incidence of other developmental and psychiatric disorders and these should be sought:
- Learning difficulty is the most common associated problem in classic autism (70–75%). Also, children with learning disorders may show features of autism and it is important to ascertain whether this is sufficient to give an additional diagnosis of autism.
- ADHD.
- Depression.
- Affective disorder.
- Anxiety disorder.

Q4. What investigations should be considered?

Children with autism find investigations very difficult to cope with; therefore consider the reasons for ordering them. These reasons are:
- To identify a treatable cause
- To identify genetic implications
- Parental ‘need to know’
- Research.

Investigations are most likely to be positive in children with a severe cognitive impairment.

Cyto genetics

Around 3% of children with autism have an abnormal karyotype. This figure is higher in children with dysmorphisms, severe learning difficulties and identifiable syndromes.

Imaging

Most common abnormalities are of the cerebellum but the significance of this is unknown. Cortical migration anomalies may sometimes be seen, but they are non-specific and will not help in diagnosis of autism or discovery of the cause. Features of tuberous sclerosis and neurofibromatosis may be recognized. Routine imaging is not recommended.

EEG

Between 21 and 43% of children with autism have an abnormal EEG. The longer the EEG, the more likely it is to reveal abnormalities, but these do not necessarily have implications for clinical care.

Q5. What are the advantages/disadvantages of finding a diagnostic label for Robbie and his family?

Advantages
- Changes attitudes towards the child positively (more understanding and appropriate responses)
- Mobilizes resources
- Indicates type of management for specific problems
- Gives an explanation to the child and parents
- Gives access to support networks.

Disadvantages
- Changes attitudes towards the child negatively (inappropriate lowering of expectations, assumptions about what diagnosis means)
- A label for life
- The label may need to change with passage of time.

Q6. What are your management options?

These include:
- Management of the child:
  - Medical and psychological
  - Social skills (group work)
  - Educational
- Management of the family:
  - Parent courses
  - Parent support
- Information/education about the condition:
  - For the parent
  - For the child.
Goals for management include:
- Fostering of development
- Promotion of learning
- Reduction of stereotypy
- Elimination of maladaptive behaviours
- Alleviation of family distress.

There are different approaches to management, which will be used in conjunction with each other.

**Pharmacotherapy**

In the US 30% of children with autism and 55% of children with high-functioning autism and Asperger's are on medication. The corresponding figures in the UK are only 5% and 10%.

Drugs will not generally affect the core symptoms of autism (and there is no evidence at present of long-term benefits) but they can ameliorate symptoms. It is important when considering medication that you are clear about the specific symptoms you are targeting and that ongoing behavioural and educational management is continued. Ensuring safe and consistent administration and monitoring for side-effects is essential. You also need to consider who you are treating (child, parent, doctor, teacher) and make sure you have informed consent as far as you are able. Substances suggested as a general treatment for autism are:

- **Secretin.** There is no evidence from randomized controlled trials of any benefit.
- **Vitamins, minerals, essential fatty acids and metals.** No studies have shown any effect.
- **Gluten- and casein-free diets.** These have their supporters but robust evidence is not yet available. Some children with autism have distressing bowel problems and a gluten- or casein-free diet may decrease bowel symptoms and greatly improve general wellbeing.

**Family support**

This is important from the start. Some support will come from the medical and educational teams but families should also be made aware of local and national parent groups. There are practical issues that can be addressed and which will make life easier.

**Behaviour management**

Difficult behaviours can be analysed as follows:
- Instrumental: in order to get something
- Social: in order to get attention
- Self-stimulatory.

The first two types of behaviour are most amenable to behaviour therapy and the last type to drug treatment.

**Educational management**

Many educational intervention programmes are advocated in autism; some claim to provide a ‘cure’ but most claim only improvement. Most approaches have in common a high degree of one-to-one intervention, often with highly structured activities. Some focus on parent intervention and some use professionals or trained workers.

**Multidisciplinary teams**

Many children will benefit from going through a formal multidisciplinary assessment and management. This may be at the local Child Development Centre or may be within the community.

http://www.cafamily.org.uk/NAPFront.PDF

National Initiative for Autism: Screening and Assessment

http://www.nelh.nhs.uk

National Service Framework for Children. Autism is one of the topics picked out as an exemplar condition

Q7. What is the prognosis for this condition?

The degree of independence reached by a child within the autistic spectrum depends to a major extent not only on the degree of intellectual impairment, but also on severity of the autism. Parents have a real anxiety that some children with high-functioning autism and Asperger's syndrome will find it hard to manage on their own. Undoubtedly some people with autism do very well, and there are a number of accounts written by autistic people about their experiences that give an insight into what autism is like from the inside.

**Classic autism**

Classic autism has a prevalence of about 0.5/1000. This rate has remained fairly steady over the years. The corresponding figure for Asperger's syndrome is around 0.25/1000.

Autistic spectrum disorder prevalence is between 3 and 6/1000, rising to nearly 1 in 100 in studies seeking out the whole range of disorders. It appears to be rising but this is likely to be because the diagnosis is being more readily recognized.

**Aetiology of autism**

A long list of associated conditions have been found in studies of children with autistic spectrum disorder:
- Fragile X syndrome (p. 32; either some autistic impairments or full autism)
- Tuberous sclerosis (p. 9; 43–60% of children with tuberous sclerosis will have autism or pervasive developmental disorder)
• Phenylketonuria (p. 97)
• Neurofibromatosis (p. 9)
• Down's syndrome (p. 30)
• Williams' syndrome
• Duchenne muscular dystrophy (p. 17)
• Non-specific dysmorphisms and other chromosomal abnormalities
• West’s syndrome (p. 8)
• Hydrocephalus (p. 24; 23% have autism)
• Severe sensory deficits
• Congenital infections
• Encephalitis (p. 292)
• Hypothyroidism (p. 108)
• Fetal alcohol syndrome
• Neurometabolic disorders (p. 93).

Autism is probably under-diagnosed in many of these conditions because, once a child has a label, there may be reluctance to pursue an additional label.

A variety of possible aetiological agents have been proposed, but convincing evidence for any of these is not available. There is no evidence linking measles/mumps/rubella (MMR) vaccination to autism.

High-functioning autism and Asperger's syndrome

These are not 'mild' autism! The distress caused to the child can be greater than with 'classic' autism because of the awareness the child has of the difficulties without the ability to understand them.

What worries me is that when Jonathan leaves school he'll have 6 or 7 GCSEs but he won't be able to go out and buy himself a shirt. (Parent of a boy aged 11 years)

David knows that when he walks into the tennis club he 'gets it wrong', whilst his younger brother Peter is fine. He doesn’t understand why this is and he gets very depressed. (Parent of a boy aged 11 years)

The characteristics are as follows:
• Presentation is usually after 3 years of age but, with hindsight, indicators can be identified in the first 3 years of life.
• Language may be normal, at least superficially.
• Presentation is often via the school with behavioural difficulties, school failure and ‘oddness’. The difficulties become apparent with the pressures for social conformity at school.

The terms high-functioning autism and Asperger’s syndrome are sometimes used interchangeably but there are differences.

Asperger's syndrome is characterized by:
• Highly developed special interests
• Verbal IQ being greater than performance IQ
• Frequent clumsiness
• More social interest than in high-functioning autism
• Socially approaching (but may be unusual or inappropriate)
• More insight into own thoughts and feelings
• Greater desire to fit in
• Cognitive (but not empathic) understanding of social rules.

These difficulties can give rise to:
• High levels of anxiety
• Psychiatric morbidity
• Socially manipulative behaviour, such as school exclusion
• Problems gaining independence
• Vulnerability, both socially and practically.

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