LEARNING DISABILITIES

1. Introduction

In people of working age, learning disability is the commonest disability in the UK.[1]

1.1 Definition

Learning disability consists of three main components:

1. Impaired intellectual function.
2. Onset at birth or in early childhood.
3. Impaired coping and social skills.

Learning disability's intellectual effects have a particular impact on language and numeracy skills. Physical disability, epilepsy, mental illness, incontinence and immobility are important associated problems.

The terminology used to describe learning disability changes with fashion, political correctness, and attempts to avoid stigmatising terms. In the UK, the term "learning disability" is used, but the expression "mental retardation" is preferred by the W.H.O. All the terms mean "arrested or incomplete development of the mind".[2]

Intelligence is a broad concept, including the ability to reason, comprehend and make judgements. Psychometric testing leads to an IQ (intelligence quotient). Intelligence is distributed in the population along a normal distribution curve.

An IQ of 100 is the centre of the distribution curve. IQs of 70 and over are considered normal.

IQ scores are used to define categories of learning disability:

1.2 Classification of Learning Disability

Classification	IQ	EQUIVALENT MENTAL AGE[3]	Proportion[4]
Mild	50 – 69	8 – 12 years	85%
Moderate	35 – 49	3 – 8 years	10%
Severe	20 – 35	1 – 3 years	3.5%
Profound	< 20	< 1 year	1.5%

1.3 Functional Effects of Learning Disability

There is considerable overlap of functional ability between the categories of learning disability.

1.3.1 Mild Learning Disability

This is not usually associated with abnormalities in appearance or behaviour. Language, sensory and motor abnormalities are mild or absent. Because it is mild, the problem is not usually apparent until school age. Adults may have difficulty coping with stress, and may need support with complex functioning such as parenting and handling their finances. However, the majority are able to live independently in the community and manage some form of employment.[5]

1.3.2 Moderate Learning Disability

People with moderate learning disability are rarely able to live independently, but they may learn to wash, dress and feed themselves. This group has limited but useful language skills. However, receptive skills tend to be better than expressive skills, leading to a high incidence of frustration and challenging behaviour. Help is needed with road sense and finances. Moderate learning disability is often associated with epilepsy, neurological, and other physical disabilities.

1.3.3 Severe and Profound Learning Disability

This group of claimants have very limited verbal and self-care skills. Severe physical handicaps are very common. Epilepsy affects 33%, incontinence 10% and inability to walk 15%. Behavioural disturbance such as purposeless, self-harming or inappropriate sexual behaviour becomes more common with increasing severity of learning disability. It occurs in up to 40% of children and 20% of adults in these categories.[5,6]

2. Prevalence

In England, it is estimated that there are 1.2 million people (2% of the population) with mild or moderate learning disability, and about 120,000 adults with severe or profound learning disability.[3,7,8,9] About 600,000 require input from specialist services.[9]

- There are more males with learning disability than females, in a ratio of 1.5:1.[3] This is probably due to the greater prevalence of sex-linked inherited learning disability in males.
- Learning disability is more common in developing countries because of a higher incidence of birth injury and anoxia, and early childhood infections.[2]
- Mild learning disability is more common in lower socio-economic groups.[10]
- The incidence of severe learning disability is falling due to improvements in prevention.[3]
- In the UK, the amount spent on services for learning disability is about £3 billion per annum.[11]
- In the UK, about 7,000 people with learning disability are in **supported** employment.[12]

3. Aetiology

There is an obvious cause for mild learning disability in about half of cases.[9] In the remainder, a combination of social, educational and emotional deprivation are the main contributory factors.[5]

As the severity of learning disability increases, there is a rising chance of finding a specific cause. In severe learning disability, 80% have evidence of organic brain damage.[3,9]

Birth injury accounts for 10% of those diagnosed with learning disability.[4] It is estimated that up to 5% of learning disability is due to physical and emotional child abuse.[9]

3.1 Genetic Factors

- Chromosomal abnormalities.
  Down's and Fragile X Syndromes are the commonest chromosomal causes of learning disability.[9]
- Metabolic disorders: Phenylketonuria and Tay-Sachs Disease (recessive) are examples.
- Tuberous Sclerosis (autosomal dominant).

3.2 Intrauterine Factors

- Malnutrition.
- Fetal alcohol syndrome.
- Infections: Rubella, Toxoplasmosis and Cytomegalovirus infections are examples.
- Pre-eclampsia.

3.3 Perinatal Factors

- Prematurity.
- Hypoxia.
- Intracerebral bleed.
- Neonatal infections.

3.4 Postnatal Factors

- Meningitis and encephalitis.
- Head Injury (accidental or physical abuse).
- Malnutrition.
- Toxins. (e.g. Lead).
- Hypothyroidism.

3.5 Environmental Factors

- Malnutrition (uncommon in developed countries.)
- Socio-economic deprivation.
- Emotional abuse.

4. Co-morbidity

Learning disability is associated with a high prevalence of epilepsy and mental health problems.[13]

4.1 Physical Co-morbidity

The more severe the learning disability, the higher the prevalence of serious physical disabilities.[14]

Physical Impairment	Prevalence in severe learning disability	Prevalence in mild learning disability
Cerebral Palsy	20%	8%
Epilepsy	35%	15%
Severe Visual Impairment	8%	5%
Severe Hearing Impairment	9%	4.5%

For a child with learning disability, the prognosis is poorer when there are multiple problems, especially those interfering with social relationships, and those inhibiting learning and play.[13]

4.1.1 Cerebral Palsy

Cerebral Palsy causes spasticity and physical disability, but may be associated with normal intelligence.[5]

4.1.2 Epilepsy

Making a diagnosis of partial epilepsy can be difficult, as patients with learning disability may be unable to describe their symptoms. Treatment of their epilepsy is complicated because they may have difficulty in describing side effects. To address these problems, there are national specialist education, treatment and assessment centres for adults with learning disability, including: the Chalfont Centre for Epilepsy and the David Lewis Centre.

4.2 Psychiatric Co-morbidity

40% of those diagnosed with learning disability also have a mental illness.[4,9] The risks increase with the severity of the learning disability.[9] In adults with learning disability, schizophrenia, mood disorders, personality disorder and neurotic disorders are all more common.[3,9,14]

- Learning disability + personality or behavioural disorder – 25 - 30%.
- Learning disability + mood disorder – 10 -15%.
- Learning disability + obsessive-compulsive disorder – 4%.
- Learning disability + schizophrenia – 3%.
- Learning disability + dementia – 3%.

Because of difficulty with communication, those with learning disability may not have the skills to express and describe what they are experiencing, so presentations may differ from those with a normal IQ. The observation of behavioural changes such as psychomotor retardation, agitation and possible responses to hallucinations can be helpful, and information from family and carers is especially important.[9]

Co-morbid Condition	Effects of Learning Disability
Schizophrenia	Delusions are less elaborate and hallucinations are simple and repetitive.
Depression	Patients are less likely to express depressive ideas. Carers may observe sadness or alterations in behaviour or sleep pattern. The suicide rate is lower.[4]
Adjustment Disorders	Common when there are changes to routine, such as loss of carers.
Phobias	Easily overlooked because of language difficulties.
Obsessive-Compulsive Disorder	More frequent than in the general population. Over-eating and unusual dietary preferences are frequent.
Personality Disorder	Common, and can lead to greater management problems.
Dementia	Tends to occur at a younger age in those with learning disability.
Sleep Disorder	Common, and may cause significant stress in carers.
Criminal Behaviour	Mild learning disability is associated with a higher rate than in the general population. Arson and sexual offences (exhibitionism) are particularly common.

5. Autistic Spectrum Disorders

The term 'autistic spectrum disorders' is used to describe a group of developmental conditions that affect the way the brain processes information. People with autism are severely affected, while Asperger syndrome describes people at the higher functioning end of the autistic spectrum.

5.1 Autism

Autism is a lifelong developmental disability that affects the way a person communicates and relates to people around them. The core feature of autistic spectrum disorders is difficulty in “making sense of the world.” An autistic person experiences a confusing mass of events, people, places, sounds and sights without order or meaning. Thus, a lot of time is spent trying to “work out the pattern behind everything”.[15]

The range of intellectual ability extends from severe learning disability to above average IQ. [15]

5.1.1 Epidemiology

The prevalence of autism is about 5 in 10,000.[3,9,15,16] More males than females are diagnosed with autism in a ratio of 4:1.[16] The prevalence does not vary with socio-economic class. [3,14,17]

5.1.2 Aetiology

Twin and family studies suggest a genetic component to the development of autism.[3] A family that already has one autistic child has a 3% risk of having another.[15] Autism is also strongly associated with organic causes of learning disability such as complications of pregnancy and birth. Neurochemical studies of autism have reported abnormalities in dopamine and serotonin metabolism.

5.1.3 Core Clinical Features

Autism is usually apparent by the age of 3 years. [9]

- **Abnormal Social Interaction.**
  There is failure to initiate, develop or respond to social situations, poor grasp of nonverbal social cues and avoidance of eye contact, so people with autism may appear aloof and indifferent.
- **Impaired Language and Communication Skills.**
  This includes delayed or impaired language development, difficulty maintaining conversation, lack of creativity and lack of imaginative play.
- **Restricted and Repetitive Behaviour.**
  This includes a “rigid routine,” interests and activities that have a preoccupation with dates or numbers, and a stereotyped behaviour pattern such as hand flapping, nodding or rocking.

Because they can concentrate on a single task for long periods, people with autism can become very proficient in those tasks that interest them. About 10% of children with autistic spectrum disorders have a special skill at a much higher level than the rest of their abilities - for example, music, art, numerical calculations or jigsaw puzzles.[15]

5.1.4 Complications

- 75% have learning disability.[3,9]
- 20% have fragile X syndrome and 6% have tuberous sclerosis.[16]
- 25% develop epilepsy.[17]
- Psychiatric co-morbidity is common.

Problems with communication and difficulty in adjusting to change often cause frustration, which may result in aggressive or challenging behaviour. It is best to talk to the autistic person in unambiguous terms, and maintain a routine. When challenging behaviour does occur, it can sometimes be channelled into harmless activities such as shredding paper or punching a pillow.[15]

5.1.5 Prognosis

Autism typically runs a steady lifelong course.[9] Specialised education and support aim to help a child to maximise their skills and achieve their full potential. Although some autistic adults learn to adapt partially to their disability, only 11% gain jobs on the open market, and only 15% achieve independent living.[9,17,18]

5.2 Asperger Syndrome

Asperger syndrome has the same core features as autism, but is at the high functioning end of the autistic spectrum. People with Asperger syndrome find it hard to read social signals, and as a result, they find it difficult to communicate and interact with others.[15]

People with Asperger syndrome can speak fluently, but they may not understand the reactions of the people listening to them. They may talk on and on, regardless of the listener's interest, or they may appear insensitive to the listener's feelings. Jokes, turns of phrase and metaphors can be confusing to a person with Asperger syndrome, because they tend to think in an over-literal way.

People with Asperger syndrome often develop an obsessive interest in memorising facts about a special subject, such as train timetables. They also prefer a set routine. Any unexpected happening or change in the routine can upset them.

Children with Asperger syndrome usually have normal or above average intelligence, and they attend mainstream school. Many seem clumsy: they have poor coordination and difficulties with fine motor control. Adults with Asperger syndrome can be considered eccentric, and may resemble those with a schizoid or anankastic personality disorder. (See the protocol ***Personality Disorders*** for further information.)

These problems of social interaction are the main cause of disability, and may make it difficult for a person with Asperger syndrome to cope in a working environment.[15]

The prevalence of Asperger syndrome is estimated to be about 36 per 10,000 in the UK.[15]

6. Down's Syndrome

Down's syndrome is the commonest specific cause of learning disability.

6.1 Epidemiology

The incidence of Down's syndrome is falling because of increased antenatal detection.

Overall, it occurs in 1 in 650 live births. For mothers aged 20 - 25 the incidence is 1 in 2,000 live births, increasing to 1 in 45 for a mother over 45.[4]

6.2 Aetiology

The vast majority of cases of Down's syndrome are caused by trisomy 21.

6.3 Clinical Features

Down's syndrome is associated with a typical facial appearance and short stature.

85% have moderate or severe learning disability.[9]

5% have autistic features and 25% have hyperkinetic disorder.[4,9]

Physical health problems are associated with Down's syndrome:

- Congenital heart disease – 40%, of which half require surgery.[9,19]
- Visual and hearing impairment – 50%.[19]
- Hypothyroidism – 30%.[19]
- Oesophageal and duodenal atresia.
- 20% increased risk of developing infections and leukaemia.[20]
- Atlanto-axial instability.

Cognitive decline and dementia (similar to Alzheimer's disease) occurs 30-40 years earlier than in the general population, and affects 25% of people with Down's syndrome.[20]

6.4 Prognosis

Some live independently in sheltered accommodation, and some find sheltered or standard employment.[20]

With improved medical care, survival has improved. At the beginning of the 20th century, life expectancy was less than 10 years. Now it is close to 50 years, with a quarter living beyond the age of 50.[4]

7. Fragile X Syndrome

Fragile X syndrome is the second commonest cause of moderate and severe learning disability after Down's syndrome, accounting for 20 - 30% of learning disabilities.

Fragile X syndrome is the commonest *directly inherited* cause of learning disability.

7.1 Epidemiology

Fragile X syndrome occurs in 1 in 1000 males. A milder form affects 1 in 2500 girls, who may have normal intelligence.[4,21]

7.2 Clinical Features

Fragile X syndrome is associated with a typical appearance, including an elongated face, large ears and blue eyes. Other features include flat feet, macro-orchidism and hyper-flexible joints. The physical features usually develop by puberty, with infants often appearing normal. Females only tend to exhibit large or prominent ears.

The degree of learning disability is similar to that in Down's syndrome, 80% of males having an IQ less than 70.[22,23] People with fragile X syndrome have particular problems with language skills. They also have an aversion to loud noise and strong smells. Difficulty adjusting to change, (particularly environmental change), and mood instability are also prominent features.[22]

Boys tend to have more behaviour problems and girls tend to be shy and socially withdrawn. Girls often suffer from anxiety and depression.

Fragile X syndrome is associated with autism and ADHD.[9]

80% have mitral valve disease and 20% have seizures. Recurrent ear infections and squint are more common, and there is an increased incidence of connective tissue disorders.[9,22]

7.3 Prognosis

Behavioural problems tend to improve with age.

Some people with fragile X syndrome are employed and are able to live independently. The majority need day-to-day supervision. They work in a sheltered environment, and either live at home or in supported accommodation.

8. Attention Deficit Hyperactivity Disorder – ADHD

ADHD does not have a significant effect on an individual's intelligence.[24] People with ADHD are creative and intuitive, but their full potential may not be achieved because of poor concentration. If untreated, ADHD interferes with educational and social development and predisposes to mental illness.

8.1 Epidemiology

The prevalence of ADHD varies in different countries due to different diagnostic practices. For example, in the UK it is about 1%, but as high as 5% in the US.[3] More males are diagnosed with ADHD than females in a ratio of 9:1.[3]

It occurs in all cultures and all social classes.[25]

8.2 Aetiology

The aetiology of ADHD is a mixture of genetic (prevalence 5 times higher in relatives), medical (as a result of encephalitis) and emotional (child abuse) causes.[3,4,26] Lead poisoning and food additives are also thought to play a role.[4]

8.3 Core Clinical Features

Unless it is very severe, ADHD is not usually recognised until the child has started school.[3]

- **Inattention**
  Easily distractible, forgetful, difficulty sustaining tasks such as play, learning and work.
- **Overactivity**
  Fidgety, reckless, socially disinhibited, inappropriately active, talking excessively.
- **Impulsivity**
  Interrupts and intrudes, unable to "wait their turn."

People with ADHD tend to be clumsy, accident-prone and get into trouble with parents and teachers. Others learn to avoid them, so they become socially isolated.[9]

8.4 Complications

20% of children diagnosed with ADHD have learning difficulties, including speech, language, social and relationship problems.[25]

A significant number of adults labelled as suffering from personality disorder are actually suffering from ADHD, and as such are likely to respond to medication.[26]

8.5 Treatment

Ritalin (methylphenidate) is an amphetamine-like stimulant. It has the paradoxical effects of decreasing activity level and improving attention. It helps to improve academic performance and relationships.[27]

Medication produces a short-lived improvement after each dose, but it is not a permanent cure.[24,28]

8.6 Prognosis

By the second decade, the problems of impulsivity and inattention tend to improve, even without medication. However, the learning difficulties caused by ADHD in childhood have long-term consequences.[26] About 60% of adults continue to experience problems.[25,28] There are high levels of psychiatric co-morbidity:[3,25]

Psychiatric co-morbidity in adults:

- ADHD + mood disorders – 18 - 59%
- ADHD + anxiety – 10 - 50%
- ADHD + antisocial personality disorder – 12%
- ADHD + substance abuse – 20 - 30%

Adults with ADHD are most likely to succeed in employment where it provides a stimulating, yet structured, environment.

9. Management of Learning Disability

In a specialist child development clinic, the assessment of learning disability begins with interviews with the patient, their parents and other carers. The family history, obstetric history, developmental milestones and schooling history are particularly important. A physical examination includes assessments of vision and hearing. Standardised measures of intelligence, language, motor and social skills complete the assessment.

When children with learning disabilities suffer psychiatric symptoms, drug treatments are used less often than in adult psychiatry. The emphasis is on psychological treatments and working with the whole family to solve problems. In adults with learning disabilities, the treatment of medical and psychiatric problems is similar to that in other patients, but some forms of psychological treatment may not be appropriate, depending on the patient's intellectual abilities.

9.1 Education

Children with learning disability are usually educated within mainstream schools. An educational psychologist will assess their educational needs, and in the most severe cases, (1% of children), may recommend that they attend a school specialising in the education of children with learning disabilities: Special Schooling. This assessment of special educational needs is called a "Statement of Need." The Statement is very significant because the local education authority is obliged to provide the services that it recommends.[29]

A typical UK school might include 3% of pupils with a Statement of Need and a further 17% within the less severe category: Special Educational Needs.[29] Children with mild learning disability spend most of their time as part of the main class, but receive additional individual and/or small group teaching.

Since 1992, there has been provision for college education for those with learning disabilities up to the age of 25. Courses include literacy skills, development of personal relationships and leisure activities.[12]

(The classification in this protocol uses medical definitions of mild, moderate, severe and profound learning disability. In other contexts, the terms may have different meaning, and this should be remembered when interpreting medical evidence from an educational professional.)

9.2 Family Support

The birth of a child with disabilities puts great strain on most families. The parents may grieve for the loss of their anticipated healthy child. The additional physical and financial burdens of caring for their child can lead to marital disharmony. However, the majority of families eventually adjust, with support from healthcare professionals, social workers, teachers, family, friends and self-help groups.

9.3 Creative Therapies

Activities such as art, music and drama can help a person with learning disabilities to express themselves.

9.4 Employment Opportunities

Sheltered workplaces allow those with practical skills to develop a routine and a role in society. Opportunities for supported employment can be accessed via the Disability Employment Adviser at Jobcentre Plus. Other employment agencies include: The Shaw Trust, Remploy and the Mencap Pathway Employment Scheme. The 1995 Disability Discrimination Act protects employees with learning disability.

9.5 Accommodation and Supervision

A large majority of people with learning disability are able to live independently or with their families.[5] Institutional care is only required for a minority of adults. Typically, it is now provided in small community units. Periods of respite care can provide an essential break for carers.

40% of parents caring for a child with learning disability are over the age of 60. Projects are exploring how to support this group of people with learning disability in their transition to new living arrangements.[30]

9.6 Key Worker

The primary care team, psychiatrists with a special interest in learning disability and social workers aim to coordinate their efforts to provide for the health and social needs of adults with learning disability. Many agencies can be involved, and it has been found that the appointment of a key worker can help a person with learning disability to gain assistance when it is needed.[14]

9.7 Psychological Therapies

Suitably modified behavioural and cognitive techniques can be successfully applied to patients with learning disabilities. For example, problems such as wetting and soiling, impulsive behaviour and phobias can be treated by behavioural therapy. This approach works by offering praise and rewards for practising the desired behaviours.[9,14]

9.8 Drug Treatments

Sedative antipsychotics are occasionally used as an adjunct to behavioural strategies in managing severe behavioural disorders, although evidence of benefit is lacking.[3,31]

9.9 Self-Help Groups

People with learning disabilities and their carers can share information and gain support from others with similar experiences. These groups also aim to influence the provision of services and facilities for disabled people.

9.10 Prevention

The availability of genetic counselling and antenatal diagnosis of conditions such as Down's syndrome has led to a reduction in the incidence of some learning disabilities. Improved perinatal care reduces the risk of brain damage. The early detection of hormonal or metabolic problems such as hypothyroidism or phenylketonuria allows treatment before learning disability sets in.[5]

There is some evidence that educational intervention in children of mothers with mild learning disability may improve their educational performance, (though not IQ), and reduce the risk of conduct disorders.[5]

9.11 Outcome

Overall, people with learning disability are living longer and enjoying a better quality of life because of improvements in health and social provision.[32]

10. Main Disabling Effects

People with learning disability all have impaired performance of intellectual tasks such as learning, short-term memory, use of concepts and problem solving. Some may have problems with spatial awareness, which may cause difficulty with dressing, for example. Poor language skills cause problems with social interaction, and are strongly associated with behavioural disorders. Additional disabilities such as epilepsy, impaired vision and hearing and physical problems often compound the disabling effects of learning disability. Learning disability runs a chronic life-long course.

10.1 Assessing the Claimant

The assessment should be made using all the information available. This includes information from the claimant's file, informal observations, medical history, typical day, appropriate physical examination, and assessment of their mental state.

Some causes of learning disability are associated with particular facial appearances or physical features. When present, these may indicate the likely range of disability.

When it is available, information from family or carers accompanying the claimant will be valuable. However, sometimes carers can be over-protective, and it is essential to develop a rapport with the claimant so that their language and social skills can be observed.

10.1.1 Mild Learning Disability

Claimants who are suffering from mild learning disability will have attended mainstream schooling. They may be living in their own home, with their family, or in supported accommodation. They will be able to do most things for themselves, although they are likely to need help with managing their finances. Their typical day history will reveal little or no restriction in their activities of daily living: they will be able to travel independently on public transport, do their shopping, enjoy contact with friends and family, and develop interests and hobbies.

10.1.2 Moderate, Severe and Profound Learning Disability

Claimants within these categories of learning disability will not be able to live independently. .

Certain syndromes always cause severe learning disability. Many of these are described in the table in ***Section 12.*** Common conditions such as autism and Down's syndrome encompass a spectrum of severity, and these cases should be assessed individually.

10.2 Helpful Questions for Assessing the Disabling Effects of Learning Disability

- Who is accompanying the claimant at the assessment? Was their presence necessary? Those with mild learning disability may be able to attend an examination centre alone or cope with an assessment at home without a companion.
- Does the claimant also have a physical disability, epilepsy, or a mental illness? **The combined effects of multiple disabilities are likely to be severe.**
- What sort of education has the claimant had? Receipt of a "Statement of Need" is significant, while attendance at a "Special School" indicates a very high level of learning difficulties.
- Where is the claimant living? Do they have a home of their own, are they living with their family, in supported accommodation, or in long-term residential care?
- Is the claimant currently attending support groups or college for further education courses in life skills and independent living? What is the planned outcome: are they aiming to live independently, or to gain work in a sheltered or open environment?
- Is the claimant able to initiate and complete household tasks? Can they plan and prepare a meal? Can they go shopping independently? What is the change from £1 if a bag of sweets costs 75p?
- How did the claimant travel to the examination centre? Some claimants will be able to travel alone on familiar routes, but would not be able to cope with a journey to an unfamiliar destination.

10.3 How to Assess the Disabling Effects of ADHD

ADHD is a treatable condition, which tends to improve in adult life. Each case should be assessed individually, with special emphasis on the typical day and assessment of the mental state.

10.4 IB-PCA Considerations

The IB-PCA assesses the ability to work in the open jobs market, not sheltered placements.

The criterion for exemption due to severe learning disability is: "*a condition which results from the arrested or incomplete development of the brain, or severe damage to the brain and which involves severe impairment of intelligence and social functioning".*[33]

This can be interpreted as a person who is incapable of living independently. If sufficient medical evidence had been available, these claimants would have been exempted or accepted at the scrutiny stage. Note that the definition of moderate learning disability in this protocol may cause the individual to fall within the category of "severe learning disability" for exemption purposes.

Severe learning disability may include one or more of:

- An inability to learn more than the most basic skills such as feeding, dressing and using the toilet.
- The need for help with some or all bodily functions.
- A failure to be aware of dangers, thus requiring supervision.
- Severe behaviour problems that require supervision, such as self-harm or violence.

It is very rare that a claimant with ADHD fulfils the criteria for exemption on the grounds of severe learning disability.

For the purposes of the IB-PCA, dyslexia on its own does not cause significant disability.

11. Reference Table of Selected Rare Syndromes

Syndrome	Aetiology	Description[34-38]	Disabling Effects
Angelman Syndrome	Chromosome Abnormality	Characteristic happy smile, inappropriate laughter, jerky movements, and wide-based gait. There is severe learning disability without speech but with some ability to sign. Epilepsy occurs in over 80%. The condition is stable. It has an incidence of about 1 in 20,000.	Severe
Fetal Alcohol Syndrome	Toxic	There is a typical appearance and growth failure, learning disability (mean IQ 60-70), hyperkinesis and microcephaly. The severity of disability depends on the level of alcohol intake. Incidence is about 1 in 1000 births.	Moderate
Klinefelter Syndrome	Chromosome Abnormality	(XXY) Males with an extra X chromosome. Features include gynaecomastia and sparse facial hair. Mild learning disability is associated with a small proportion of cases.	Mild
Lesch-Nyan Syndrome	Disorder of uric acid metabolism	This condition affects males. Treatment with allopurinol can control the associated gout, but it cannot prevent the neurological syndrome of choreoathetosis, spasticity, learning disability (IQ 40-65), and self-mutilation.	Severe
Niemann-Pick Disease	Disorder of lipid metabolism	Occurs in A, B & C types. Until school age, development is typically normal, then there is severe motor and intellectual deterioration. There is no effective treatment. Affects 1 in 10,000 births.	Fatal
Phenylketonuria	Error of phenylalanine metabolism	An autosomal recessive disease, which occurs in 1 in 10,000 births. It is routinely screened for in the UK, and can be controlled by restricting the intake of protein.	None
Prader-Willi Syndrome	Chromosome Abnormality	Features include: short stature, small hands and feet, severe obesity and IQ 50-80. The incidence is about 1 in 20,000 births.	Moderate
Rett's Syndrome	X chromosome abnormality	This condition begins to cause severe learning disability in the first 2 years of life, and eventually results in severe global disability. "Hand wringing" movements are a typical feature. It affects 1 in 10,000 girls.	Severe
Sturge-Weber Syndrome	Sporadic	Facial port wine stain indicates haemangiomas on the ipsilateral cerebral hemisphere. These cause contralateral seizures, often with hemiparesis and hemianopia. Learning disability is common.	Moderate
Tay-Sachs Disease	Ganglioside storage disease	Progressive motor weakness from 6 months of age, and seizures, blindness, and deafness. The child dies before it is 5 years old. The incidence is 1 in 2000 in Ashkenazi Jews. Autosomal recessive.	Fatal

Tuberous Sclerosis	Chromosome Abnormality	This condition is named after tuber-like growths on the brain and other tissues. It is associated with learning disability, epilepsy, and characteristic skin lesions, including facial angiofibromas. Affects 1 in 8000 births. Normal life expectancy for all sufferers.	50% have learning disability
Turner's Syndrome	Chromosome Abnormality	(45X) These girls lack an X chromosome. They have short stature and a webbed neck. IQ is usually average, but they have impaired verbal and numerical skills and right-left disorientation.	Mild

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