CARING FOR PEOPLE WITH LEARNING DISABILITIES

Learning disability is the term currently used in England to describe people with impaired social functioning due to intellectual deficits. It replaced the term mental handicap. The international term remains mental retardation, which is synonymous with learning disability. It is a medically heterogeneous group.

The borderline between learning disability and normality is not sharp. As in most conditions such as dementia, alcoholism or even diabetes, the way in which people are diagnosed as having learning disability depends as much on social events leading them to contact with medical staff as on their intellectual deficits. This is most noticeable at the borderline where few people reach psychiatric services and those who do usually do so because of behavioural or emotional difficulties.

Intelligence tests usually test only processing and conceptual intelligence, and do not assess temperament, social intelligence, or sensory functioning. Raw scales are transposed into standard scores with mean of 100 and standard deviation of 15. Almost two thirds of people will have an IQ between 85 and 115. An adult with an IQ of 100 might be expected to have the educational age of 15 – 16.

• diagnosis of Learning Disability requires:
  reduced level of intellectual functioning (IQ less than about 70)
  resulting in impaired adaptive behaviour [and therefore a need for support]
  occurring before age 18

• there is a further subdivision on IQ:
  mild (IQ 50 - 70) theoretical prevalence 2.5%
  moderate (IQ 35 - 49) theoretical prevalence 0.2%
  severe (IQ 20 - 35) theoretical prevalence 0.05%
  profound (IQ less than 20) theoretical prevalence

Adults with mild learning disability can be expected to acquire independence in most self care and domestic activities as well as being able to earn money in unskilled work. The main difficulties can be expected in reading, writing and monetary skills, in emotional and social immaturity, and in ability to readily adapt to social expectations, or external stressors. Less than 20% of them are likely to be known to specialist services.

Adults with moderate learning disability usually have more obvious impairments, and rarely achieve more than simple literary or monetary skills. Most need a level of supervision and close support to participate in domestic activities. Most are likely to be known to specialist services.

Adults with severe learning disability frequently have additional disabilities such as epilepsy, or physical or sensory disabilities. Most need supervision with their self-care.

Adults with profound learning disability usually need close supervision and care for their entire life. Many are able to feed themselves with a spoon. Most can understand and make simple statements and requests. Most have multiple disabilities.
Aetiology
In discussing the aetiology of learning disability, one needs to take the medical model used when discussing disease, one stage further: the medical model has a primary cause, usually a physical cause (eg a cerebral haemorrhage) producing secondary effects, or signs and symptoms, (eg memory loss or hemiplegia). The third stage that should always be considered is that of the social disability that the signs and symptoms cause. This is little discussed in most branches of medicine, but is gaining increasing emphasis especially in the rehabilitation of people with chronic illness. Because of the multiple stages in producing "disability" it is usually a gross simplification to speak of one cause for the aetiology of a person's learning disability. Similarly it is very difficult to predict accurately the degree of learning disability that will be produced by a specific organic cause.

Discussion of the aetiology of learning disability is further complicated by the fact that several "causes" such as autism, cerebral palsy, or hypothyroidism are in fact not primary causes, but secondary effects of known or unknown primary causes.

Chromosomal and genetic primary causes
The two most common and important genetic causes of learning disability are those found in Down syndrome and Fragile X. Down syndrome is the most common autosomal chromosome abnormality causing intellectual impairment. Other autosomal chromosome abnormalities are much rarer and usually lead to death in infancy.

Learning disability is more common in males than females by about 15% in the community. This is partially due to recessive genes on the X chromosome being unopposed in males, who by definition do not have a second X chromosome. Fragile X is the most common gross abnormality of the sex chromosomes that leads to intellectual impairment. Though an abnormal number of sex chromosomes is more common, such as Turner syndrome (X0), Klinefelter syndrome (XXY), and the "super male" (XYY), it is unusual for these syndromes to produce marked learning disabilities.

Down Syndrome
Down syndrome is the most common cause of severe learning disability known, being diagnosed in one third of people with an IQ less than 50. It was first well described by Dr John Langdon Down in 1865, who labelled it mongolism, a label now discouraged. The majority of people with Down syndrome (95%) have an additional chromosome 21 (trisomy 21). This additional chromosome is normally of maternal origin (probably more than 90%) and the risk of occurrence rises 50-fold with increasing parental age. About 3-4% of people with Down syndrome have additional material from chromosome 21 due to an unbalanced translocation of chromosomes which attaches part of a third chromosome 21 to another chromosome. About half of these are fresh mutations, but the remainder occur because a parent has a balanced translocation. Such a parent has a high risk of further children with Down's syndrome. A further 1-2% of people have Down syndrome due to mosaicism, with only part of their cells having trisomy 21. From studies of people with only part of an additional chromosome 21 (as a translocation) it is possible to say that it is the distal segment of the long arm of chromosome 21 whose triplication is vital for Down syndrome.

Down syndrome occurs in about 100/100 000 live births. A further 15/100 000 pregnancies are probably currently medically terminated because of Down syndrome. In
the United Kingdom, most live births (60%) are diagnosed prenatally. Most infants with Down syndrome are recognised immediately after birth upon routine paediatric examination.

A diagnosis of Down syndrome can be confirmed prenatally by amniocentesis. Amniocentesis carries a risk of miscarriage, and should only be offered to high-risk pregnancies where the pregnancy will be terminated if abnormal. This risk is calculated after sampling maternal blood for alpha-fetoprotein, unconjugated oestriol and human chorionic gonadotrophin. It can also be detected by skilled ultrasound. A few suffer from heart or gut abnormalities which require rapid intervention for the person to survive infancy, but most who survive the first 5 years can expect to live until at least middle age.

People with Down syndrome are usually severely intellectually disabled, and in addition suffer from a wide range of physical abnormalities which need monitoring, including heart and gut abnormalities, eye and teeth problems, recurrent respiratory infections, obesity and sleep apnoea, dry skin, hypotonia, joint laxity and atlanto-axial instability; deafness (often due to ear wax) and epilepsy. In addition after age 30, they have an increasing risk of hypothyroidism and an alzheimer-type dementia (55% by age 50). They need regular medical reviews to ensure maximal physical and mental health.

**Fragile X**

This is caused by abnormal duplications for Argine (CGG) at (q27.3) on the long arm of the X chromosome. Probably 3% of the population have a small number of these duplications, and are therefore carriers. The abnormality increases with each generation, and the number of replications determines the severity of effect, people needing at least 150 copies to develop the full syndrome. The abnormality can now be demonstrated by DNA probes. It is estimated that up to 60/100 000 all births and 100/100 000 male births may have Fragile X syndrome. Up to 9% of people known to have a learning disability probably have Fragile X syndrome.

Many children with Fragile X syndrome are not identified until they are older. They are usually male, tend to have a distinctive long face with prominent forehead, large simple ears, and almost all males have large postpubertal testes. There is usually a family history of males with learning disability, though some females may also be affected. About a third of people with fragile X are either severely or profoundly intellectually disabled, but a significant number pass their initial milestones well, and only fall behind after the time of speech development.

Several physical problems are common in the group, with excessive joint laxity, flat feet, sight abnormalities, and mitral valve prolapse reported. Most suffer from hyperactivity and attention deficits as children, and benefit from the usual treatment for hyperactivity. Autism may be more common in this group, but they are usually keen to be sociable, though when greeting people they often avert their gaze, turning the top half of the body to do so. They are often stressed by a lot of things occurring around them and when so stressed many characteristically bite themselves, developing calluses on their wrists. They sometimes need treatment for anxiety. Speech and language development is always retarded, and the more vocal often repeat themselves to maintain the pace and flow of conversation. This can benefit from speech therapy.
Tuberous Sclerosis
The frequency of Tuberous Sclerosis, or Epiloia, is generally estimated at 10/100 000 but this is probably an underestimate. This is an autosomal dominant condition, though 60% are due to fresh mutations. It appears to be due usually to a gene on chromosome 9. Its expression is highly variable and many people are found to have it only after they have been examined closely because a relative has a more severe form. Only 40% develop a learning disability.
A child with Tuberous sclerosis appears normal at birth, but as time progresses, many develop poorly controlled seizures, often within the first year of life, at the time of vaccination, which can raise anxiety of vaccine damage. There is then increasing mental impairment. Many later develop a classical butterfly distribution of flesh coloured nodules (angio-fibromas) on the face. In addition to the facial nodules, other skin abnormalities may occur such as cafe-au-lait spots, shagreen patches, fibrous patches and nail bed fibromas. Nodules occur elsewhere, as hamartomas in the brain and rectum, as rhabdomyomas in the heart, as phakomas in the retina, and as angiomyolipomas in the kidneys. Most lumps are asymptomatic. Some with multiple brain tumours have autistic traits and hyperactivity. A few die young due to fits and a few die later due to brain tumours or kidney problems.

Phenylketonuria [PKU]
This is the most common single gene recessive defect that is well documented, with an incidence of 12/100 000. Three different forms of defect have been identified, all resulting in the build up of the amino acid phenylalanine in the body, which at high levels poisons the nervous system. Half of cases are due to classical phenylketonuria where there is a defect in the enzyme that breaks down phenylalanine - phenylalanine hydroxylase. Typically untreated sufferers start as 'normal' infants with blue eyes and blond hair but after a few months become irritable, hypertonic, develop seizures and become increasingly retarded. A special phenylalanine-limited diet from birth prevents this from occurring. Such a diet can be suspended in adolescence, but doing so often results in an irritable person. In non-classical phenylketonuria, the enzyme defects cause a wider disruption of metabolism, and treatment is less successful. It is cost effective to test for PKU at birth and it is universally tested for in the United Kingdom but an appreciable minority still do not start the special diet within 3 weeks of birth. There is said to be an increased incidence of Autism.

Other Congenital Causes
Hypothyroidism
Congenital Hypothyroidism has variable incidence of about 25/100 000. Severe hypothyroidism, or cretinism as it used to be called, has long been recognised as a cause of learning disability, and one of the earliest colonies for ‘idiots’ was opened in 1839 in Switzerland for cretins at the Abendberg.
Congenital hypothyroidism is not clearly of genetic origin and probably has many causes, like cerebral palsy. It is cost effective to biochemically test for hypothyroidism at birth, and most areas now do so. Treatment with thyroxine is then simple and adequate treatment starting before 3 months of age will prevent intellectual disability. However many children with hypothyroidism have other abnormalities, especially deafness, which can impair intellectual development.
Cerebral Palsy
Cerebral palsy is a disorder of posture or movement, but it commonly causes learning difficulties and people with cerebral palsy often have intellectual impairment. As well as having congenital causes it can also be caused by postnatal events which cause its prevalence to increase with age and by age five it occurs in about 250/100 000 children. About 30% of the infants with cerebral palsy who were born at full term have severe learning disabilities. Though only 20% of low birth weight babies with cerebral palsy have severe learning disabilities, cerebral palsy itself is much more common in these babies, being 10 times more frequent in infants weighing under 2500g at birth, compared with those born heavier. Probably less than 10% are caused by perinatal factors. Birth asphyxia is rarely the cause of either cerebral palsy or learning disability. Much more common causes are vascular and haemorrhagic lesions, many of which occur prenatally, but both of which are very difficult to prevent.

They usually need a lot of input to help with limb spasticity and abnormal growth; and are prone to gut motility problems.

Congenital Infections
These infections by cytomegalovirus, rubella, and toxoplasma gondii are most dangerous to a foetus when occurring before 16 weeks but they currently are not a common cause of learning disability with all congenital infections probably causing severe learning disability in less than 3/100 000 births. All these people would have cerebral palsy in addition.

Neural Tube Abnormalities
People with spina bifida can have a wide range of orthopaedic, urinary, bowel, endocrine and visual problems as well as seizures and intellectual disability. Up to 10% will have learning disability and almost all of these will be multiply handicapped.

Foetal alcohol effects
Estimates of the frequency of foetal alcohol syndrome vary immensely, but it probably occurs in more than 100/100 000 births. However people with foetal alcohol syndrome are not necessarily learning disabled. The effects are variable, but often include microcephaly, a smooth philtrum with a thin smooth upper lip, heart abnormalities, hyperactivity and poor fine coordination. Measured intelligence is not usually profoundly impaired and the average IQ is in the mid 60's. Many do well in mainstream education.

Postnatal causes
Very few people with learning disability can attribute their disability directly to a single postnatal event. The most common postnatal direct causes of severe learning disability are abuse, injury, infection and toxins, but the incidence of these appears to vary widely and the data on them is of little use. Other postnatal causative factors in learning disability include epilepsy, superimposed psychiatric illness, and autism, which will be discussed below.

Children with mild learning disability usually have at least one parent with a low IQ and are nine times more likely to come from a lower socio-economic family. It is impossible to dissect the reasons for this and allocate relative risk values to congenital causes,
postnatal events, or socio-economic influences. Low socio-economic class is associated with low birth weight, higher perinatal morbidity, poor housing, big families and poor education, all of which are associated also with mild learning disability.

Epilepsy
Epilepsy is a symptom whose effects can worsen learning disabilities and which is more common in learning disability. In the United Kingdom about 0.5% of the population will have epilepsy at any one time, though probably 6% of people have a seizure at some time in their life. Epilepsy occurs in 30% of people with an IQ below 50, and in almost all profoundly and multiply handicapped people. Temporal (and probably frontal) lobe seizures are common and should be suspected of causing any behavioural disorder. Seizures provoke disability both directly and indirectly. For example seizures interfere with concentration, learning and memory; severe seizures can cause further damage; people with seizures often become anxious about the social embarrassment of having a seizure, lose self-confidence and avoid social activities.

Sensory Deficits
Sight and hearing deficits are very frequent associations of learning disability. Subtle deficits can easily be missed by carers and are often not looked for in the psychiatric examination. However they are a potent cause of further handicap and both cause and worsen learning disability, as well as causing frustration and behavioural disorders. All people with learning disability should have their eyesight carefully checked.

Population surveys suggest that up to one third of people with a learning disability have impaired hearing. A minority of these are recognised by the carers (a clinician becomes accustomed to the statement "he can hear if he wants to"). People who do have hearing impairment benefit from an environment which caters for this: where people use sign language as well as speaking, people look directly at each other when speaking, and loop systems are installed to improve the usefulness of hearing aids. Hearing aids are useful in the more severely deaf, but need perseverance and enthusiasm by all concerned to ensure that they do not become a further handicap.

Psychological effect of learning disability
All families in which disability occurs are different. For each family and person affected, the disability has a different meaning and impact. Having said this, the feelings engendered by having a disabled child occur at many levels and in many directions. Guilt, embarrassment and fear as well as bereavement and grief at the loss of a normal child are all common. This ocean of interacting emotions can produce a large number of reactions that significantly handicap the development of the child and result in a parent burying the family in helping others or campaigns; neglect of other relationships within the family. The clinician must also be aware that each major stage of a child's development - for example going to school, adolescence, leaving school or moving away from home - will create a further surge of feelings in a parent making it even more difficult for that stage to be successfully crossed. Through all this, the clinician must also be aware of any siblings and try to ensure that their needs are not neglected.

Few people care to imagine what a person with learning disability experiences as he or she develops socially. However he is normally aware of his social failings, throughout his life. Awareness of one's own disability is a continuous process: for example as an infant you are treated differently, you attend a different school, your younger siblings
overtake you in ability and their friends no longer play with you, you then cannot find a girlfriend, and discover that you will not be able to have the job, car, house or spouse that the media lead you to expect. In addition you are often stigmatised and ridiculed. Your relationships with other disabled people are devalued. You are likely to be abused sexually and physically.

Not surprisingly many people with disability react adversely to these experiences, further handicapping themselves, for example by insisting on behaving like an infant, avoiding new situations or social events, refusing to take responsibility for their own behaviour, using violence or tantrums to control the world around them, or becoming extremely self centred and retreating into a fantasy world.

Psychiatry of learning disability
Psychiatric assessment of behavioural change in people with learning disability must include social history and physical examination.

Assessment should aim to determine:
- current developmental level
- social skills
- any changes in skills in the last few years
- personality
- usual reactions to stress
- recent life changes and environmental stresses
- current environmental support

as well as evidence of:
- psychiatric disorders
- physical disorders (eg pain)

Behaviour disturbance is more common in the learning disabled than in a non-disabled population occurring in 60% in some surveys. Behavioural changes are now often labelled "challenging behaviour" which itself is defined as behaviour of such an intensity, frequency or duration that the physical safety of the person or others is likely to be placed in serious jeopardy, or behaviour which is likely to seriously limit or deny access to and use of ordinary community facilities. This definition includes all behaviours that limit interaction, both active and passive. However the term is now often used more restrictively as a euphemism for aggression.

Depression
Depression is common, especially grief reactions. It often leads to regression in behaviour, with previously abandoned behaviours returning. The person will often withdraw, losing interest in socialising, or joining in tasks. He can be tearful, and appear sad. He may become incontinent. In severe depression he can have the usual biological signs of depression and hallucinations may occur. He often appears irritable, as he is reluctant to be disturbed and may resist having to join in activities (including talking to others) by using violence. He can in his retreat become obsessed with bodily sensations and constantly complain of pain. He can also become very anxious. Treatment with counselling, environmental manipulation (reducing stress) and antidepressants is usually effective, though care must be taken not to worsen any seizures with the antidepressant, which is usually needed in standard doses.
Phobia
The most common phobias in people with learning disability probably are animal phobias, which occur in about 10% of residents of institutions. Most common is a dog phobia. These phobias can usually be treated by modelling, relaxation and graded exposure.

Anxiety States
These are common, but probably under diagnosed, and frequently neglected, again because of communication difficulties. Panic disorder does occur. Treatment is the same as in the general population. Relaxation exercises, modified for understanding and associated with other cues such as relaxing music, can be as effective as in a more able group.

Attention deficit disorders/Hyperactivity
These are also more common. They can seriously stress carers as well as impairing a person's ability to learn. Treatment is the same as in the general population. They usually improve with age.

Psychosis
People with learning disability can become psychotic, especially under stress, but it is often difficult to diagnose due to their problems in communicating their experiences. Antipsychotics can work but often in small doses.

Autism
Classic Autism occurs in about 20/100 000 children, but the spectrum (including Asperger syndrome) may occur in up to 1/200 children. The autistic spectrum has the common features of impaired social communication, impaired social interaction associated with rituals and routines, with impaired social imagination. There are multiple causes but genetic influences appear strong. The effect appears to be a problem with the integration of messages within the brain and a problem with pattern recognition. The behaviour of people with autism often stresses and confuses people. The more profoundly impaired have learning disability. In adult life, many improve as they are taught how to handle anxiety and stress, and how to function despite their impairments, but the underlying impairments do not disappear unless the person is intelligent enough to learn how to cope with them. This is a pervasive developmental disorder for which no effective treatment has been found other than a highly structured style of education and living, combined with treatment where necessary of hyperactivity, anxiety, mood disorders and any psychosis that occurs.

Ethics
How our society treats people with a learning disability raises many ethical dilemmas. The core dilemma is how we balance our respect for all people as humans, of inherent value and with inherent rights, whilst we also act in a way that makes it clear that we consider that abnormality should be avoided.

Foetal screening implies a decision to terminate abnormal foetuses. The dilemma is what abnormalities justify termination and why.

Similarly the neonatal period often brings the dilemma of how energetically an infant should be treated.
A different dilemma arises later when people with learning disability require treatment. As with anyone, physical treatment without consent is technically assault. Parents or guardians can only consent on a child's behalf and not for an adult. Any treatment should be discussed with the person, and his consent sought. However it must be decided beforehand if the person can give consent that is meaningful. If it is meaningful, then the person can refuse as validly as give consent, and his refusal must be respected. It is the personal responsibility of the treating clinician to decide if the person can give or withhold consent validly. What constitutes valid consent must depend in part on the nature of the treatment. Simple treatments such as having a bath, and its consequences are probably more easily understood than say, radiotherapy for cancer. People who are severely learning disabled are unlikely to be able to think ahead and appreciate fully the future consequences of their treatment. If a person cannot consent, then it is the responsibility of the treating clinician to consult with family, carers, and other doctors, to establish that there is general agreement that the treatment is necessary and in the person's best interests. In general all treatment should be appropriate and in the best interests of the person, as would be considered by a reasonable person. Experimental treatments are unethical.

**Components of a good Service**

The services need to incorporate the following:

- **Support Services**
  - Financial support for person and carers
  - Support workers and families
  - Advocacy service

- **Community Assessment and Therapy**
  - General Practitioner
  - Community Learning Disability Team
  - care coordinator

  The generic services (eg orthopaedics) should be able to provide a service to anyone whatever their learning disability.

- **Residential Services**
  - Respite care
  - Board & Lodging scheme
  - Assessment & Therapy Units (Health)
  - Homes (which are the person’s home, and not an institution)
    - with physical security
    - with 24 hour nursing staff
    - with 24 hour care staff
    - with visiting care staff

- **Day care services**
  - Resource and Activity Centre
  - Adult Education Service
  - Sheltered Employment
  - Supported open employment

A GP can expect to see a person with LD at least once a day. Problems that often present are obesity, pain, gut problems, sight and hearing problems, menstrual
difficulties, and the management of epilepsy and of ‘challenging behaviour’. Challenging behaviour can often happen as a result of pain or discomfort as much as due to stress, distress or psychiatric illness. GP’s have a strong role in discouraging discrimination and should include people with LD in health screening as any other patient. They need to make their surgeries friendly to people who cannot read or write or have other communication difficulties, and people who have mobility problems. The recent white paper ‘Valuing people’ stresses the need for people with a learning disability to be integrated into society and to access generic services. People with LD are included in all the National Service Frameworks and should be included in all screening services. Specialist Community Learning Disability Services usually have only 0.5% of the general population known to them as having LD, and so only know about 10% – 20% of those who have an IQ less than 70.

Communicating with people with a LD
All students are encouraged to talk with people with a LD, on their placement. They will find that the techniques are similar to those that are general good practice – keep instructions and questions simple; do not assume that the person can read and write; check out what they understand; use gestures and drawings to augment your communication. A person may be able to give valid consent, if time is taken to explain things. With the more profoundly handicapped, relax and work out with the carer who is accompanying how to set up an interaction. You may not be able to discuss specifics, but you should be able to obtain a constructive interaction.

Further reading: