Unit 2 – Causes and manifestations of learning disabilities

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Outline

This unit will:

- Outline a range of causative factors that may result in learning disabilities.
- Distinguish between pre, peri and postal natal factors.
- Identify a range of number of known conditions in learning disabilities.
- Identify some of the health challenges that people with learning disabilities may experience as a consequence of particular clinical manifestations of learning disabilities.
- Specifically outline genetic and chromosomal abnormalities and their manifestation.

Introduction

The isolation and subsequent identification of the many causes of learning disabilities has in the past, been a complex and relatively slow process. However, with recent advances in genetic technology and increasing awareness of the effects of social deprivation, new knowledge and understanding are continually being developed. Learning disabilities cannot be considered a single condition; rather it is a compendium of various degrees of impairment of intellectual and social functioning. The range of causative factors in learning disabilities is variable and diverse and the origin of many conditions is still unknown. Some of the main causes and manifestations of learning disabilities found within society will be explored in this unit. It is envisaged that if you wish to further your knowledge and understanding in this area you will use the recommended reading at the end of the unit. In this unit you will necessarily encounter a range of technical terms that you may be unfamiliar with we have left the responsibility with you to ensure that you research all new terms and understand their meaning.

Importance of diagnosis

Knowing and understanding the cause of an individual's learning disabilities is important for several reasons. Gates (2000) has emphasised that the identification of the cause of a learning disability and the provision of an early diagnosis are crucial in limiting the feelings of self-blame that may be experienced by some parents of children with learning disabilities. He has also suggested that informed parents are less likely to reject the child and more likely to adapt appropriately to the care demands posed by having a child with learning disabilities. Other reasons for the identification of the causes of learning disabilities and early diagnosis include the need to:

- Understand the possible manifestations of the identified condition over a defined period of time.
- Identify the range of therapeutic approaches that may be used to ameliorate the effects of the condition including the mobilisation of specific resources.
- Establish in some cases, the degree of risk to other family members of the condition re-occurring in siblings and offspring.

Classification of learning disabilities

Learning disabilities may be classified in a number of different ways. One way is by the origin of the cause of the learning disability. This may fall into two main categories; genetic or environmental. Genetic conditions may originate prior to conception or during the very early stages of the development of the foetus. Environmental causes on the other hand, include those external factors that may affect the development of a foetus and child either in the pre-conceptual, prenatal, perinatal or postnatal periods. The stage of development at which the damage to the child is incurred is a second method by which learning disabilities may be classified. Where the cause of the learning disability is unknown, such conditions are usually described as *idiopathic*.

The following sections will enable the reader to understand some of the clinical features associated with specific conditions or syndromes found within learning disabilities. When reading each section, the student may like to reflect upon the type of health care that maybe required in responding to the needs of affected individuals and their families.

Genetic causes of learning disabilities

Many of our physical features (phenotype) originate from our genetic make up (genotypes). The information required for the development of these characteristics exists in the form of genes that are passed from parents to offspring during the process of cell division. Genes can be found on structures called chromosomes that are present within the nucleus of every human cell and consist of the genetic material DNA (Deoxyribose Nucleic Acid). A generalised cell structure is shown in Figure 2.1 and the genetic material we are talking about here will be found located in the nucleus at the heart of the cell. This nucleus, and every other cell nucleus, contains 23 pairs of chromosomes, of which 22 are referred to as autosomes and 1 which is referred to as the sex chromosomes (XX in females and XY in males). This is shown as the normal (male) karyotype in Figure 2.2. During the process of cell division (meiosis), the chromosomes from each parent combine and then divide. During this process, changes in both the structure of the chromosomes and their respective genes can occur and this may give rise to genetic abnormalities that are the cause of some learning disabilities.

It is believed that between 30-40% of moderate to severe learning disabilities are caused by changes in the genetic makeup of an individual (Knight et al, 1999) and developments in genetic technology arising from The Human Genome Project suggest that the percentage may be higher. A relatively recent study by Knight et al (1999) has shown that a number of previously undiagnosed conditions in learning disabilities could be attributed to subtle chromosomal rearrangements.

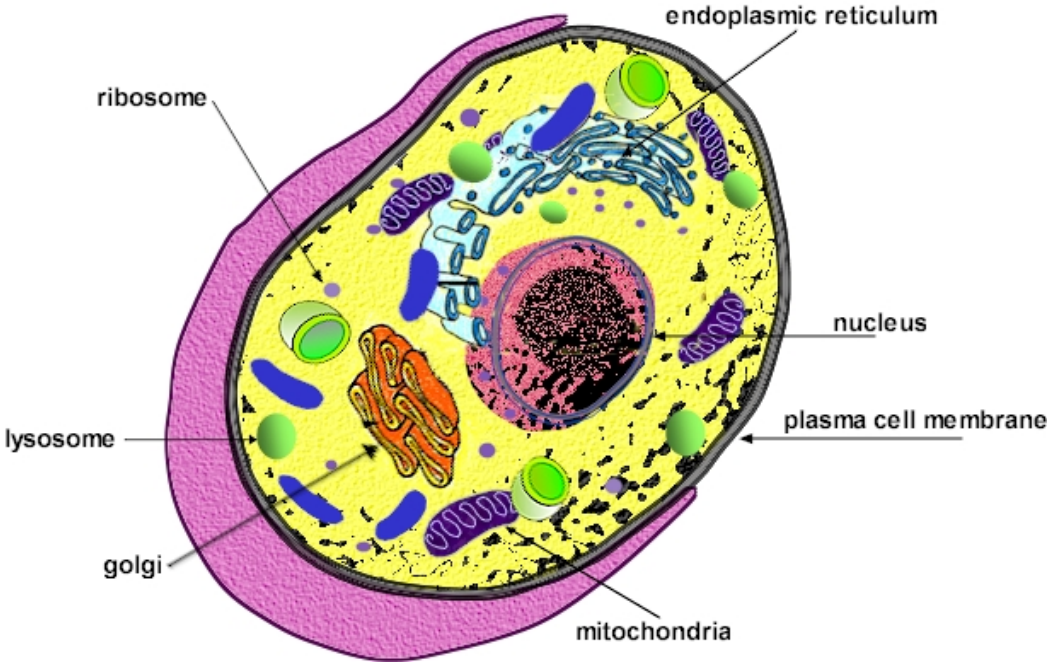

Figure 2.1 Structure of human cell

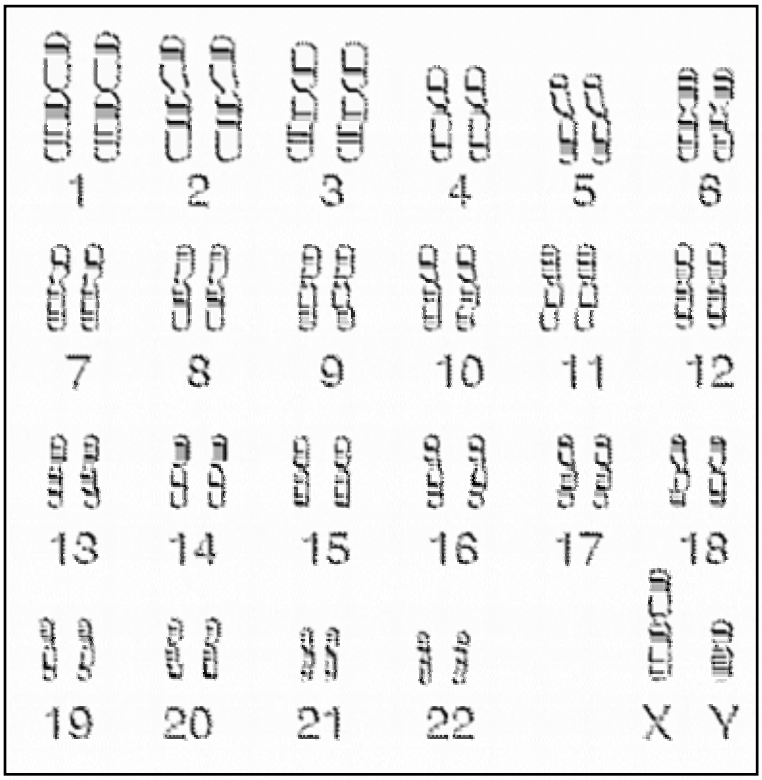

Figure 2.2 The human karyotype

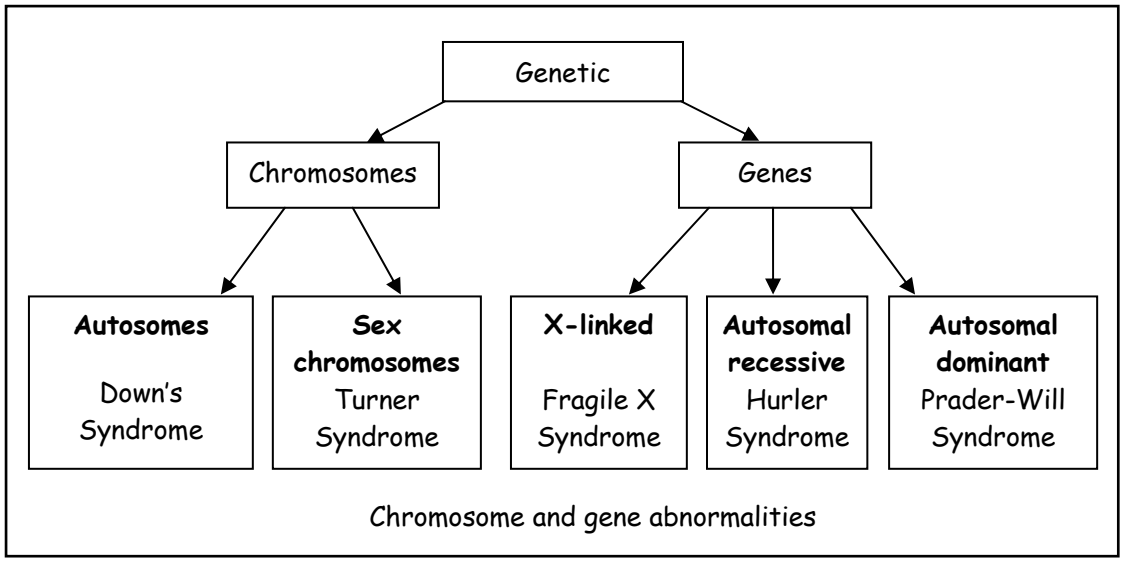

Figure 2.3 A simple classification system of the genetic causes of learning disabilities

Figure 2.3 presents a simple classification system of the genetic causes of learning disabilities. It can be seen that an example is given for each group and some further examples of each are provide in the next section of this unit.

Chromosomal abnormalities

This section will provide specific examples of conditions in learning disabilities that result from changes in either the structure or number of autosomes and sex chromosomes. Where changes in the structure of the chromosome may occur, this may include the deletion, duplication, translocation non-disjunction or inversion of genetic material.

1. Manifestations of autosomal abnormalities

Down's Syndrome (Trisomy 21) – this syndrome was first described by John Langdon Down in 1866 and results from the non-disjunction of chromosome 21 pair during cell division resulting in an individual having three rather than two chromosome 21. Incidence rate of this syndrome is between 1 in 650 and 1 in 700 (Mueller & Young, 1998) but becomes higher with an increase in maternal age. Typical characteristics of individuals include: short stature, small ears, ear and eye defects, heart defects and an increased susceptibility to infections. In rare cases, some individuals with Down's Syndrome may have a mixture of cells that contain either trisomy 21 or the normal number of chromosome 21 and this condition is known as *mosaicism*.

Cri-de-Cat – a relatively rare condition with an incidence rate of about 1 in 37,000 live births. It was first described in 1963 by Lejeune et al and given the name because affected infants are found to have high pitched cries like those of a cat. Typical characteristics include microcephaly, low set ears, wide spaced eyes and moderate-severe learning disabilities.

2. Manifestation of sex-chromosome abnormalities

Klinefelter Syndrome (XXY) – this syndrome was first described by Klinefelter and associates in 1942 and only affects males. It results from the non-disjunction of the XY chromosomes during cell division resulting in an individual having an extra X chromosome. Incidence rate of this syndrome is between 1 in 500 and 1 in 1000 births. Typical characteristics include a large forehead, ears and jaw, and following the onset of puberty hypogonadism (enlarged testicles) and gynecomastia (enlarged breasts). Psychosocial problems are also common. The degree of learning disability is said to be moderate, with a few cases of individuals with profound learning disabilities.

Turner Syndrome (XO) – this syndrome only affects females and results from the loss of one of the normal two XX chromosomes. Incidence rate is estimated to be 1 in 2,500 births. Typical characteristics include short stature, web like neck, non-functioning ovaries, and in some cases, learning disabilities although the normal range of intelligence is generally associated with this syndrome.

Essential	*Reader Activity 2.1*

Case illustration

Michael is a man with Down's syndrome who lives at home with his elderly parents. He breathes through his mouth and has a congenital heart disease. He tends to get respiratory infections fairly commonly and often he has eye infections.

Using your acquired knowledge of Down's syndrome write a short paper on the kind of support would you give to Michael and the sorts of advice you might provide for his elderly parents? Also describe any preventive measures that you would recommend in relation to Michael's common infections.

Optional	*Reader Activity 2.2*

Find out the address and contact information on a range of local organisations that offer support for parents of children with genetic conditions. This exercise is useful in raising your awareness of the types of organisations that are available to parents also it will increase your knowledge of another aspect of learning disabilities.

3. Manifestations of gene abnormalities

This section will provide specific examples of conditions in learning disabilities that result from changes in the structure of the genetic material making up a gene. These changes may include the deletion, duplication, addition, inversion and substitution of the parts of the DNA. Gene abnormalities are generally categorised by the mode of transmission of the defective gene, you may wish to refer back to the simple classification system in Figure 2.3. These forms of transmission can be described as autosomal dominant, autosomal recessive or X-linked and these are all descried below. Some conditions may result from the interaction of various genes (polygenic) although these have not been described in this unit.

Autosomal dominant conditions

In the case of autosomal dominant conditions, transmission is reliant upon only one parent carrier of the defective gene and there is a 50% chance of it occurring in the offspring. (see Figure 2.4)

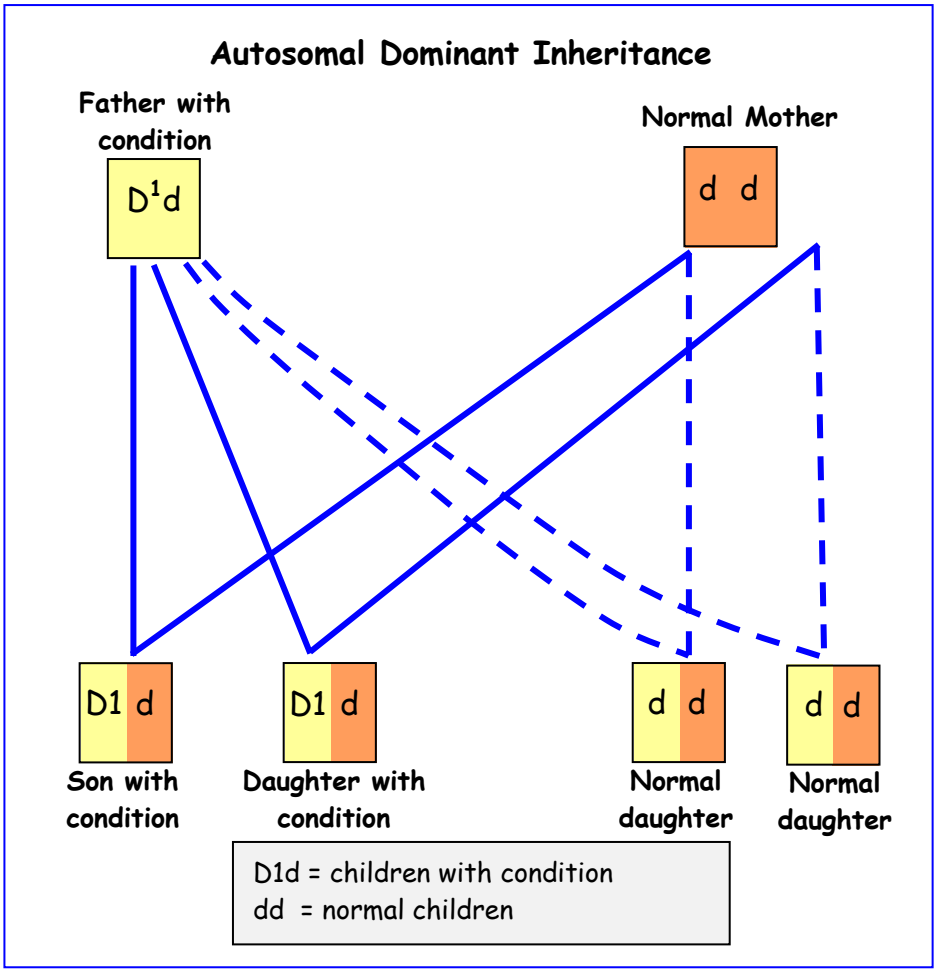

Figure 2.4 Dominant inheritance

The next section provides you with some examples of disorders that are inherited through this process.

Prader-Willi syndrome – this syndrome results from deletion of part of the genetic material on the long arm of chromosome 15 and usually originates from the father. Incidence rate is approximately 1 in 15,000 and affects males and females alike. Characteristics of this condition include small hands and feet, hypogenitalism (underdeveloped testes) and cryptorchidism (undescended testes) in males. One of the most notable characteristics, however, is hyperphagia (excessive over eating). Without proper help and support, people with this syndrome commonly experience gross obesity and the related conditions of heart disease and diabetes which may result in premature death.

Tuberous sclerosis (epiloia) – this condition was first described in 1880 and is estimated to affect between 1 in 30,000 to 40,000 births. It is characterised by growths on the brain and major organs. A butterfly shaped rash (adenoma sebaceum) will be present on the face. Epilepsy is common in people with this condition. Whilst normal intelligence may be present, 60% of affected people have some degree of learning disability.

Autosomal recessive condition

In the case of autosomal recessive conditions, transmission is reliant on both parents being carriers of the defective gene and in this case, there is a 25% chance of the condition manifesting in the offspring. (see Figure 2.5)

The next section provides you with some examples of disorders that are inherited through this process.

Phenylketonuria – this condition was described by Fölling in 1934. It is a disorder the affects protein metabolism resulting in raised blood levels of phenylalanine. If not maintained at a normal level through diet control, these levels may become toxic and result in brain damage. This condition is believed to affect approximately 1 in 12 000 live births. It is commonly diagnosed using the Guthrie's test (heel prick test) which is a blood test carried out 6 to 14 days after birth. If left untreated, typical characteristics include lack of pigmentation in the eyes, skin and hair, hyperactivity, autistic features and epilepsy. Degree of learning disability is severe in individuals where condition is not treated.

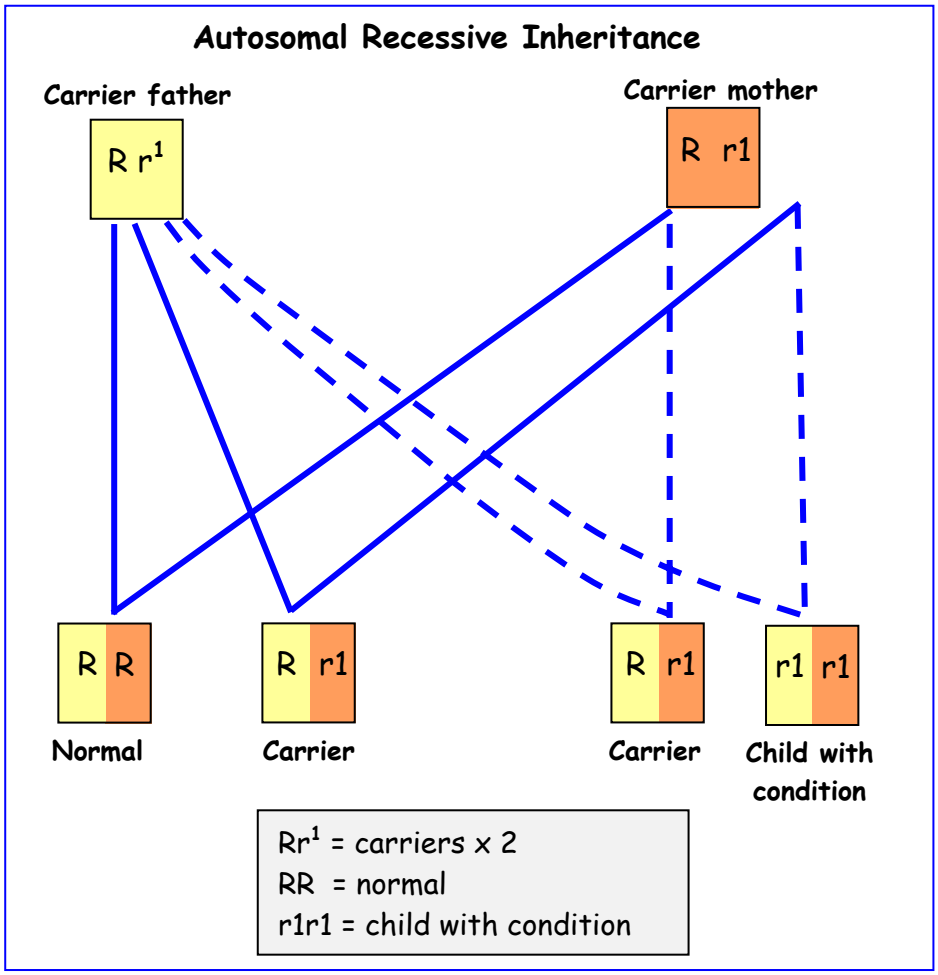

Figure 2.5 Recessive inheritance

Hurler syndrome – Hurler syndrome is one of a number of mucopolysaccharide disorders and has an estimated prevalence rate of 1 in 150,000 births. It is characterised by the abnormal storage of mucopolysaccharides in connective tissue that ensures that affected individuals are short in stature and have thick coarse facial features and a low nasal bridges. Hirsutism is a common characteristic as is the more lethal presence of heart defects. Affected individuals may also have sight and hearing impairments. Death normally occurs in adolescence.

X-linked recessive conditions

Fragile X Syndrome – a syndrome that occurs more commonly in males than females with a prevalence rate of 1 in 4000 and 1 in 8000 respectively. It is believed to be the most common cause of learning disabilities next to Down's syndrome. This condition arises from the breaking off of the bottom tip of the X chromosome making the site fragile. Common characteristics include oversized head, long face, prominent ears, large jaw, language difficulties and varying degrees of learning disability. Behavioural problems are also characteristic.

Essential	*Reader Activity 2.3*

Genetic counselling is a service offered to people who may be at risk from a disorder that may be hereditary in nature. During the consultation process the patient is informed about the characteristics of the condition and its likely progression. Advice may be given with regard to the probability of its reoccurrence in future generations providing vital information to families.

Find out about the current genetic tests that may be used to diagnose the presence of genetic conditions. To do this you may use the web or try your local library or resource centre at the University. Given that the Nursing and Midwifery Council are to introduce minimum competencies concerning genetics in the near future it seems sensible to address this issue now.

Environmental factors

Environmental factors have an important influence on the physical and intellectual development of individuals. Where the environment contains positive factors for growth such as food, warmth, love, safety and sensory stimulation, normal development should ensue. However, in some cases, certain environmental conditions may hinder the growth and development of an individual and this could result in a learning disability. Environmental factors that may exert influence on development might occur in the pre-conception, prenatal, perinatal and postnatal stages and include infections; trauma, drugs and social deprivation. (see Figure 2.6)

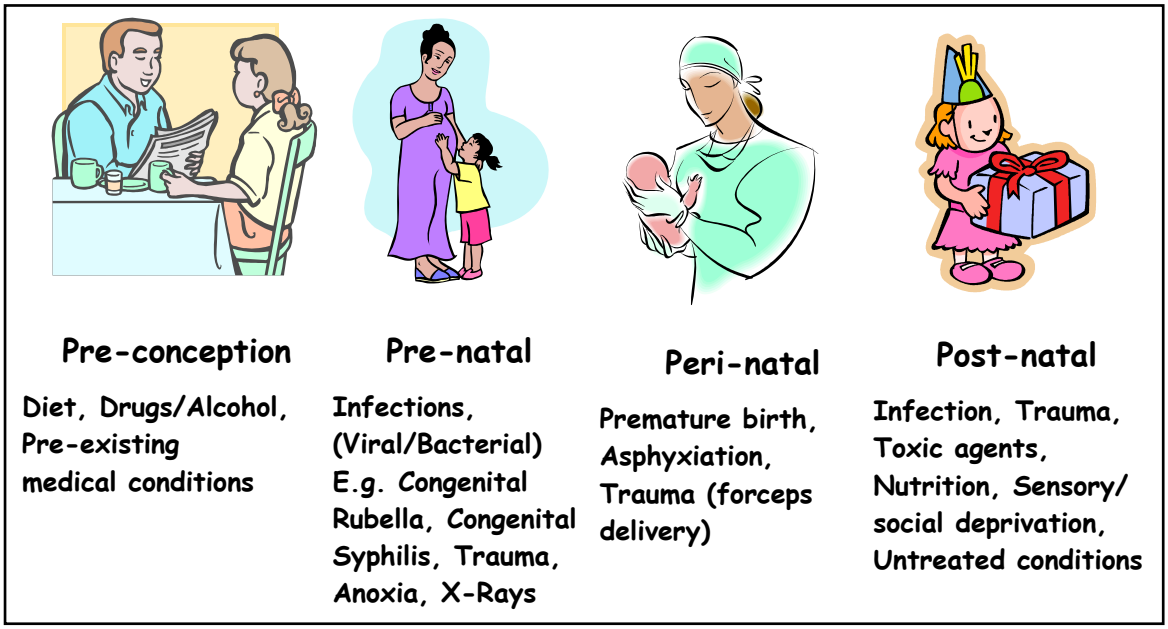

Figure 2.6 Environmental causes of learning disability

Environmental causes of learning disabilities includes trauma during the pre-natal, perinatal and postnatal phases and includes accidental and non-accidental injury. At the prenatal stage, this also includes the delivery of the child using forceps or suction. Restriction of the oxygen supply to the foetus during the prenatal and perinatal phases can result in brain damage. In the latter stage, asphyxiation may occur if the umbilical cord becomes wrapped around the baby's neck. The consumption of drugs including alcohol also accounts for the stunted growth and lack of brain development observed in some children. Toxic agents, lead poisoning, chemical pollutants and hard metals such as mercury, manganese and strontium poisoning are also known causes of brain damage. In the postnatal phase of development, poor nutrition and a lack of sensory and social stimulation can impair development and result in learning disabilities. Other causes of learning disabilities include acquired infection that can result in brain damage at prenatal, perinatal and postnatal stages of development and includes rubella (German measles, mumps and chickenpox). In the past syphilis was a common cause of learning disabilities but is now less common. Viral infections may give rise to

encephalitis or inflammation of the brain and the subsequent degree of learning disabilities can be severe. Dehydration, which will occur very rapidly, leads to brain haemorrhage, which can cause brain damage.

Congenital Rubella – this condition was first described by Gregg in 1941 and is characterized by a number of severe defects which include cataracts, deafness, congenital heart defects and learning disabilities. Damage occurs when the rubella virus passes across the placenta and attacks the developing nervous tissue in the unborn foetus. In recent years, the prevalence of congenital rubella has declined with the introduction of rigorous immunization programmes.

Conclusion

This unit has examined the varying causes and manifestations of conditions in learning disabilities. It has been demonstrated that changes in the genetic makeup of individuals can result in the manifestation of specific syndromes, whilst environmental factors can also cause brain damage in the prenatal perinatal and postnatal periods. Diagnosing the cause of learning disabilities is important for families in allowing them to adapt to their child. It is also important for health services, as it provides specific information about actual and potential needs of individuals that allows the mobilisation of appropriate resources. Caution however, must be exercised as providing a diagnostic label may reduce individuality. As it will be demonstrated in Units 4 and 5, health professionals must ensure that they always see and value the person and their individual characteristics before any diagnosis.

References

Gates, B. (2000) 'Knowing: the importance of diagnosing learning disability.' Journal of Learning Disabilities, 4(1) pp5-6.

Knight, S.J.L.; Regan, R.; Nicod, A. (1999) Subtle chromosomal rearrangements in children with unexplained mental retardation. Lancet, 345(9191) pp1676–1681.

Mueller, R.F. and Young, I.D. (1998) Emery's elements of medical genetics. 10th edn Edinburgh: Churchill Livingstone.

Further reading

Barr, O. (1999) 'Genetic counselling: a consideration of the potential and key obstacles to assisting parent adapt to a child with learning disabilities.' British Journal of Learning Disabilities 27:pp30-36.

Health Departments of the United Kingdom (1998) Advisory committee on genetic testing: report on genetic testing for late onset disorders. London: Department of Health.

Hogenboom, M. (2001) Living with genetic syndromes associated with intellectual disability. London: Jessica Kingsley.

Watson, D. (2003) Causes and manifestations of learning disabilities. In: Gates, B. (ed) Learning disabilities: toward inclusion. Edinburgh: Churchill Livingstone.

Resources

ASSERT (Angelman Syndrome Support Education and Research Trust)
P O Box 505
Sittingbourne
Kent ME10 NE
Tel: 01980 625616

The National Autistic Society
393 City Road
London EC1V 1NE
Tel: 020 7833 2299

Cornelia de Lange Foundation
Tall Trees 106 Lodge Lane
Grays
Essex RM 16 2UL
Tel: 01375 376439

Cri-du-chat Syndrome Support Group
Penny Lane
Barwell
Leicestershire LE9 8HJ
Tel: 01455 841680

Down's Syndrome Scotland
Head Office
158-160 Balgreen Road
Edinburgh EH11 3AU
Tel: 0131 3134225
Email: info@dsscotland.org.uk

SOFT UK (Edward's Syndrome)
48 Froggarts Road
Walmley
Sutton Coldfield
West Midlands B76 8TQ
Tel: 0121 351 3122

Foetal Alcohol Syndrome Trust
15 Wasdale Road
Aintree
Liverpool L9 8AS
Tel: 0151 284 2900

FORESIGHT The association for the promotion of preconceptual care
28 The Paddock, Godalming
Surrey GU7 1XD
Web: www.foresight-preconception.org-uk

Fragile X Society
53 Winchelsea Lan
Hastings
East Sussex TN35 4LG
Tel: 01424 813147

Klinefelter's Syndrome Association
56 Little Yeldham Road
Little Yeldham
Nr Halstead
Essex C09 4QT
Tel: 01787 237460

The Research Trust into Metabolic Diseases in Children
(RTMDC)
Golden Gates Lodge
Weston Road
Crewe
Cheshire CW1 1XN
Tel: 01270 250221

Society for Mucopolysaccharide Diseases
46 Woodside Road
Amersham
Bucks HP6 6AJ
Tel: 01494 434156

Neurofibromatosis Association
82 London Road
Kingston-on-Thames
Surrey KT2 6PV
Tel: 020 8547 1636

National Society for Phenylketonuria (UK) Ltd
7 Lingley Lane
Wardley
Gateshead NE10 8BR
Tel: 01845 603 9136 (helpline)

Prader-Willi Syndrome Association (UK)
2 Wheatsheaf Close
Horsell
Woking
Surrey GU21 4BP
Tel: 01483 724784

Royal National Institute for the Blind
Resource Centre, 9 Viewfield Place
Stirling FK8 1NL
Peter McConnachie
Tel: 01786451752
Email: Ekennedy@rnib.org.uk

The Scottish Society for Autistic Children
Head Office, Hilton House
Alloa Business Park
Whins Road
Alloa FK10 3SA
Scotland
Tel: 01259 720044
Fax 01259 720051
Email: ssac@autism-in-scotland.org.uk
Web: www.autism-in-scotland.org.uk/

Tay-Sachs and Allied Diseases Association
Golden Gates Lodge
Weston Road
Crewe
Cheshire CW1 1XN
Tel: 01270 250221

Tuberous Sclerosis Association of Great Britain
Little Barnsley Farm
Catshill
Bromsgrove
Worcestershire B61 0NQ
Tel: 01527 871898

The Turner Syndrome Society
C/o The Child Growth Foundation
2 Mayfield Avenue
London W4 1PW
Tel: 020 8994 7625/020 8995 025