Aetiology of learning disability

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Abstract

This contribution discusses the aetiology of learning disability. Learning disability, termed 'mental retardation' in ICD-10, is defined as significantly below average general intellectual functioning (IQ < 70), accompanied by deficits or impairments in adaptive behaviour, and with an onset in childhood. It is important to make a diagnosis of the aetiology of a learning disability if possible, for a number of reasons. These reasons can be broken down into psychological, physical, psychiatric and genetic counselling. As with most aspects of medicine, the causes of learning disability can be divided into genetic, biological and environmental. These variously occur before, during or after birth. Down syndrome is the most common specific cause of learning disabilities, followed by fragile-X syndrome. In 30–50% of people with learning disability, however, no cause is found, although the aetiology is more likely to be identified in the more severe cases. It is important to realize that in people with learning disability the cause will be multifactorial. Antenatal factors such as chromosomal and genetic factors, congenital infections, teratogens, malnutrition, radiation or unknown factors can all cause learning disability. Perinatal factors can cause a learning disability; these include asphyxia/hypoxia at birth, mechanical birth trauma, hypoglycaemia and prematurity. Postnatal factors can cause learning disability and there has been a recent emphasis on more research in this area. The obvious causes are injury due to trauma, anoxia or infection (meningitis/encephalitis). The environment is also an important factor in contributing to learning disability.

Keywords Down syndrome; fragile-X syndrome; intelligence quotient; learning disability; mental retardation

Learning disability, termed 'mental retardation' in ICD-10, is defined as significantly below average general intellectual functioning (IQ < 70), accompanied by deficits or impairments in adaptive behaviour and with an onset in childhood. The lower the IQ an individual has, the more severe the learning disability.

IQ levels follow a normal distribution curve for IQs above 70. Below 70 the curve has an asymmetric distribution, with higher than expected numbers with IQs of less than 70 and a small 'bump' at the lower end (Figure 1). The normal distribution represents the differences in IQ secondary to environmental and genetic effects right across the IQ range. The bump at the lower end is due to organic causes.

The term 'learning disability' is a descriptive diagnosis and does not infer any particular aetiology. Statistically, the prevalence of people with IQ < 70 should be 2.5% (2 SD from mean). Actually, the prevalence of people with learning disability is 1–2%. This is because of a variety of reasons, including a differential mortality rate, particularly in those with a more severe degree of learning disability. In addition, not all those with an IQ below 70 will have a learning disability, as they may have no adaptive functioning problems.

People with mild learning disability account for about 85% of all those with learning disability; those with a moderate learning disability for about 10%; those with severe learning disability 3–4%; and those with a profound learning disability about 1–2%.

What are the causes of learning disability?

As with most aspects of medicine, the causes of learning disability can be divided into genetic, biological and environmental. These variously occur before, during or after birth. The perceived importance of these factors at the different levels of learning disability is changing: whereas previously it was thought that severe learning disability was caused by genetic factors and milder learning disability by biological and environmental factors, it is now acknowledged that there are genetic causes in those with a mild learning disability such as fragile-X syndrome. A chromosomal abnormality can be detected in up to 25% of people with learning disability.

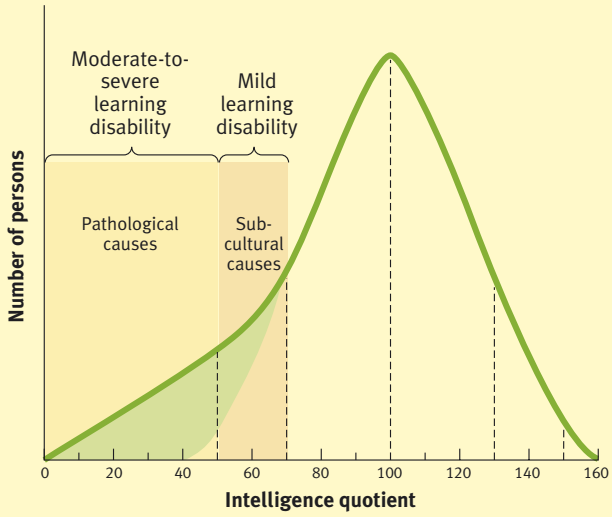

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Figure 1

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In 30–50% of people with learning disability, however, no cause is found, although aetiology is more likely to be identified in the more severe cases. It is important to realize that in people with learning disability the cause will be multifactorial; for example there may be an interaction between genetic susceptibility and various adverse psychological and social environmental factors.

Why diagnose the cause of learning disability?

Psychological

It is important to make a diagnosis of the cause of learning disability for a number of reasons. First and foremost must be the involved person's right to know their own diagnosis. As a diagnosis of learning disability is one that is often made in childhood, it is important to fully involve the parents. The diagnosis is frequently made after the first year of life and the parents have to make great changes in their hopes and expectations for their child. This can be a traumatic time for parents and finding a cause for the learning disability may help the whole family to adjust more easily to the diagnosis of learning disability. It is important to help the family at this time, particularly as it is known that any psychological trauma can impact adversely on the learning disability prognosis.

Physical

If a cause of learning disability is identified then this will allow professionals to work with the affected person and his or her family to allow appropriate investigations to be carried out and to enable access to appropriate healthcare. Many causes of learning disability are associated with particular physical health needs; for example, Down syndrome with congenital heart disease and early onset of Alzheimer's disease, and tuberose sclerosis with epilepsy.

Psychiatric

A number of causes of learning disability are associated with particular behaviours, the 'behavioural phenotype' (e.g. Prader–Willi with hyperphagia; Cornelia de Lange with self-injury). Knowledge of these associations allows for earlier targeted management plans. Other causes of learning disability are associated with particular psychiatric diagnoses (e.g. velocardiofacial syndrome with schizophrenia; fragile-X syndrome with social anxiety).

Genetic counselling

Once a diagnosis has been made the parents and, in later life, the affected person may wish to know whether there is a hereditary component to the learning disability. If so there may be implications for any future children of the parents and of the affected person. Inheritance of chromosomal and genetic disorders is a complex area and referral to a clinical geneticist will be necessary.

Causes of learning disability: before birth

Chromosomal and genetic factors, congenital infections, teratogens, malnutrition, radiation or other unknown antenatal factors can all cause learning disability (Table 1).

Aetiology of learning disability

Antenatal causes
- Genetic
- Infections
- Toxins
- Irradiation
- Maternal malnutrition

Perinatal causes
- Asphyxia/hypoxia at birth
- Mechanical birth trauma
- Hypogylcaemia
- Prematurity

Postnatal causes
- Trauma
- Anoxia or infection (e.g. meningitis, encephalitis)
- Environmental

Table 1

Genetic factors

A genetic disorder is a disease caused by abnormalities in an individual's genetic material (genome).[1] There are four different types of genetic disorders: (1) single-gene; (2) multifactorial; (3) chromosomal; and (4) mitochondrial.

Single gene disorders: there are a variety of conditions that are inherited through a dominant (i.e. tuberous sclerosis) or recessive gene (i.e. phenylketonuria).

Phenylketonuria (PKU) used to be a more common cause of severe learning disability. It has a prevalence of 1 in 12,000 births. Cases are now found with screening, thus limiting later damage.

Tuberous sclerosis (TSC) is a rare, multi-system genetic disease that causes benign tumours to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs and skin.[2] It results in a combination of symptoms including seizures, learning disability, behavioural problems, skin abnormalities and kidney disease. TSC has an estimated prevalence of 1 in 6,000 newborns. It is caused by defects, or mutations, on three genes – TSC1, TSC2 and TSC3.

Velocardiofacial syndrome (VCFS) – at least 90% of people affected have a deletion of the long arm of chromosome 22 (22q11). VCFS occurs in 1 per 4000 births and is the second most common autosomal disorder to cause learning disability. This condition is characterized by structural or functional palatal abnormalities, cardiac defects, unique facial characteristics (elongated face, almond-shaped eyes, wide nose and small ears), hypernasal speech, hypotonia, developmental delay and mild learning disabilities. Poor social interaction or behavioural difficulties are common. Psychiatric disorders (including obsessive–compulsive disorder and schizophrenia) are reported in at least 10% of those affected.

Prader–Willi syndrome – it is estimated that one in 12,000 to 15,000 people is affected. Although considered a 'rare' disorder, Prader–Willi syndrome is one of the most common conditions seen in genetics clinics and is the most common genetic cause of obesity that has been identified.[3] This is a complex genetic