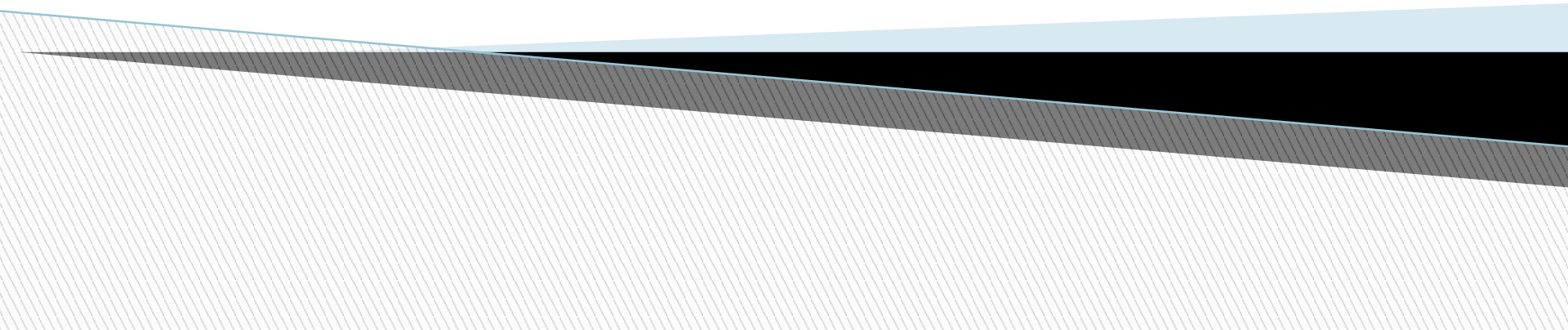
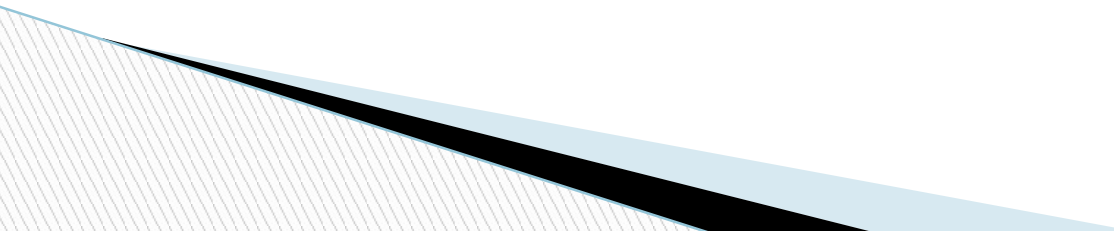


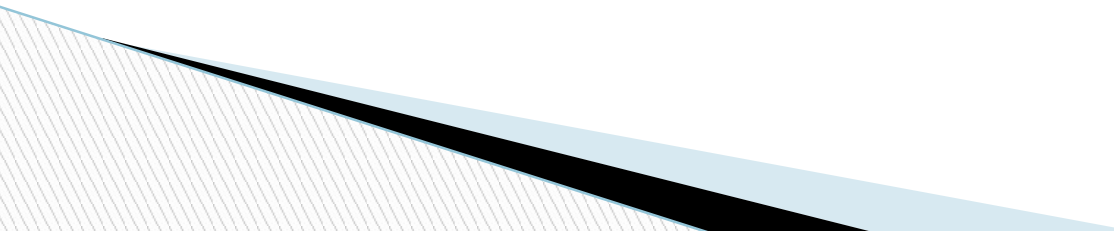
Learning disability (mental retardation)



Introduction

Terminology: Over the years, several terms have been applied to people with intellectual impairment from early life. In the nineteenth and early twentieth centuries, the word 'idiot' was used for people with severe intellectual impairment, and 'imbecile' for those with moderate impairment. The special study and care of such people was known as the field of *mental deficiency*. When these words came to carry stigma, they were replaced by the terms *mental subnormality* and *mental retardation*.



- **The development of ideas about learning disability**
 - Early in the twentieth century, Binet's tests of intelligence provided quantitative criteria for ascertaining the condition. These tests also made it possible to identify lesser degrees of the condition that might not be obvious otherwise. Unfortunately, it was widely assumed at the time that people with such lesser degrees of intellectual Impairment were socially Incompetent and required institutional care.
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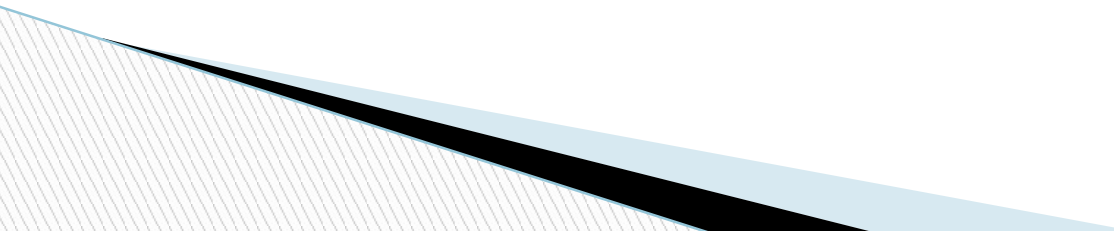
Epidemiology of learning disability

- Studies have produced variable results, but a current estimate for the UK is 9-14 in 1000 children, and 3-8 in 1000 adults. In recent years, the incidence of severe learning disability has fallen substantially, because of the recognition of preventable prenatal and perinatal causes of learning disabilities. However, the prevalence has not fallen, and in fact is expected to rise by over 10% by 2020. This is due to First, people with learning disability, particularly those with Down's syndrome, are living longer.
- Secondly, improvements in maternal and neonatal care are resulting in a growing number of children with learning disabilities, who have survived significant events such as extreme prematurity.

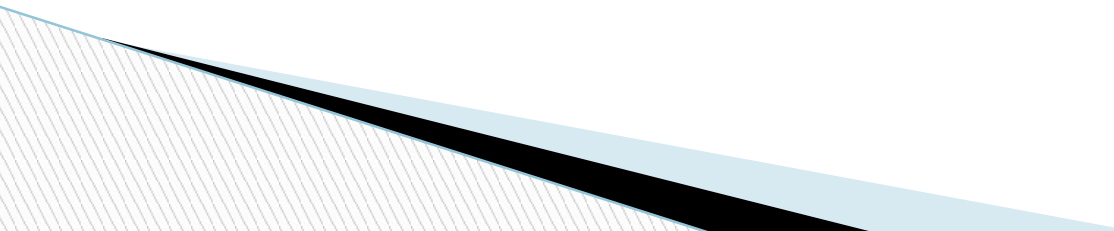
Clinical features of learning disability

- Learning disability is usually divided into three or four subtypes, which are defined by IQ which is normally distributed in most populations with a normal range of 90-110 with 20 points as a standard deviation value. People with IQ value of 71-90 are considered with borderline intelligence.
- **Mild learning disability (IQ 50-70):** People with mild learning disability account for about 85% of those with learning disability. Usually their appearance is unremarkable and any sensory or motor deficits are slight. Most people in this group develop more or less normal language abilities and social behavior during the pre- school years, and their learning disability may never be formally identified.

- ▣ **Moderate learning disability (IQ 35-49):** People in this group account for about 10% of those with learning disability. Many have better receptive than expressive language skills, which is a potent cause of frustration and behaviour problems, speech is usually relatively simple, and is often better understood by people who know the patient well. Activities of daily living such as dressing, feeding, and attention to hygiene can be acquired over time, but other activities of daily living such as the use of money and road sense generally require support.

- **severe learning disability (IQ 20-34):** people with severe learning disability account for about 3-4% of the learning disabled. In the preschool years their development is usually greatly slowed. Eventually many of them can be helped to look after themselves under close supervision, and to communicate in a simple way -for example, by using objects of reference.
 - **Profound learning disability (IQ below 20):** People in this group account for 1-2% of those with learning disability. Development across a range of domains tends to be around the level expected of a 12-month-old infant.
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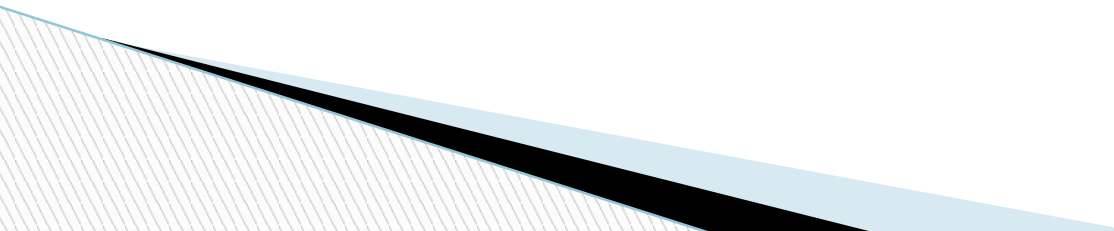
Physical disorders among people with learning disability


- ▣ *Sensory and motor disabilities and incontinence* are the most important physical disorders in people with learning disability. People with severe learning disability (especially children) usually have one or often several of these problems. Only one-third are continent, ambulant, and without severe behaviour problems. Around 25% are highly dependent on other people. Among the people with mild learning disability, similar problems occur, but less frequently.
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□ *Epilepsy* is a frequent and clinically important problem in learning disability. Around 15-25% of people with learning disabilities have a history of epilepsy, compared with 5% in the general population. The prevalence increases with the severity of learning disabilities, (up to 80% of the profound group are epileptic). Epilepsy is more commonly associated with certain causes of learning disability, such as fragile X syndrome, tuberous sclerosis, Angelman syndrome, and Rett syndrome.

□

Psychiatric disorders among people with learning disability

- It is now generally agreed that people with learning disability experience psychiatric disturbances similar to those which affect the general population. However, the symptoms are sometimes modified by low intelligence, and may not be easily recognized or communicated.
 - **Schizophrenia**: affects 3-4% of people with learning disability, compared with less than 1% in the general population. The overlap between the two conditions largely reflects shared genetic factors. Clinically, delusions may be less elaborate than in patients with schizophrenia of normal intelligence, hallucinations may have a simpler content, and thought disorder is difficult to identify.
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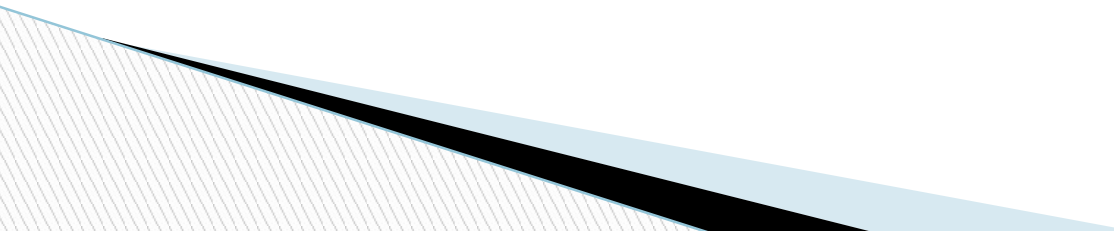
- ❑ **Mood disorder:** The rate of depressive disorders is comparable to, or slightly higher than, that of the general population . However, people with learning disability are less likely than those of normal intelligence to complain of mood changes or to express depressive ideation. Diagnosis has to be made mainly on the basis of an appearance of sadness, changes in appetite and sleep, and behavioural changes of retardation or agitation.
 - ❑ **Anxiety disorders:** Adjustment disorders are common among people with learning disability, occurring when there are changes in the routine of their lives.
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- **Autism and attention-deficit hyperactivity disorder (ADHD)**: Hyperactive behaviour and autistic-like behaviour are frequent symptoms of learning disability. In addition, the diagnoses of autism and ADHD are more common than among the general population.
- **Abnormal movements**: Stereotypes, mannerisms, and rhythmic movement disorders (including head banging and rocking) occur in about 40% of children and 20% of adults with severe learning disability. Repeated self-injurious behaviours are less common but important.

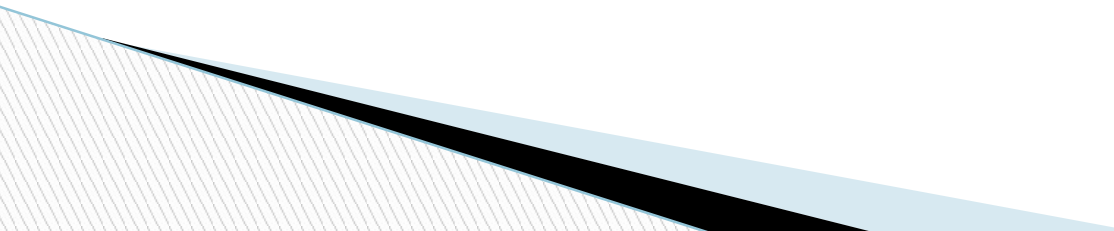
Aetiology of learning disability

- in mild MR mostly no cause can be found while in the moderate & severe to profound group usually the cause can be identified.
- *Genetic factors* are a major cause of learning disability. This is in part because intelligence is heritable (with estimates of around 30- 50). a specific chromosomal or genetic defect can be the necessary and sufficient cause of a person's learning difficulty. These include Down's syndrome and fragile X syndrome, the two commonest causes of learning disability.

□ *Environmental factors* are conveniently divided into pre-, peri- and postnatal factors, reflecting the time at which they are believed to have occurred. The relative importance of environmental factors varies according to setting. For example, they are more significant where healthcare provision or general health are poorer, and they may be affected by local factors (e.g. areas of low iodine predispose to congenital hypothyroidism).

- Overall, it is estimated that prenatal (genetic and environmental) factors cause 50-70% of learning disability, with 10-20% originating perinatally, and 5-10% originating postnatally; the proportions, and the causes within each category, depend on the population being studied.
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Prevention

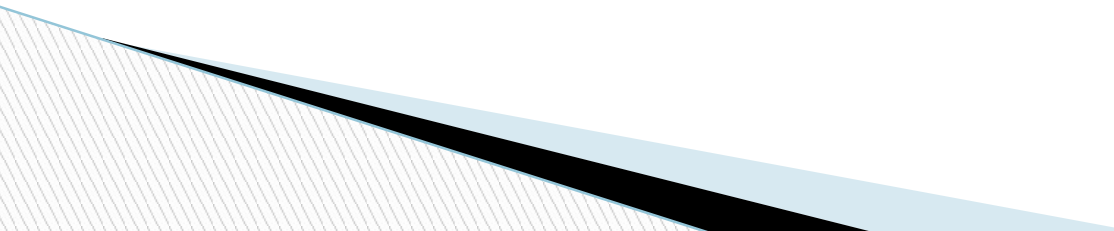
- Genetic screening and counseling
 - Prenatal care begins even before conception, with immunization against rubella for girls who lack immunity, and advice on diet, alcohol, and smoking.
 - *Rhesus incompatibility* is now largely preventable. Sensitization of a rhesus-negative mother can usually be avoided by giving anti-D antibody.
 - Postnatal prevention In the UK, all infants are routinely tested for phenylketonuria, and testing for hypothyroidism and galactosaemia is becoming increasingly common.
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Down's syndrome

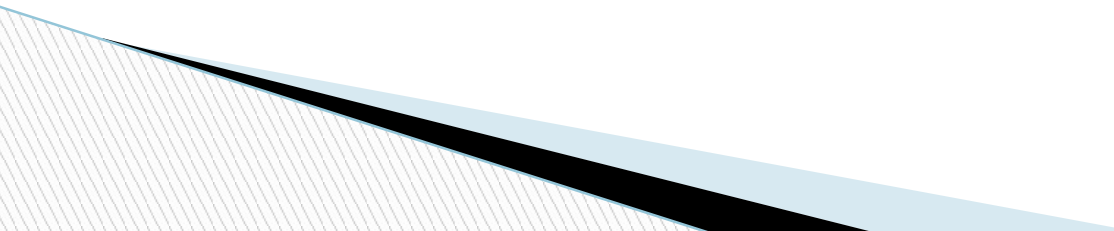
This condition is a frequent cause of learning disability, occurring in 1 in about every 650 live births. It is more frequent among older women, occurring in about 1 in 2000 live births to mothers aged 20-25 years, and 1 in 30 .live births to those aged 45 years

- In 1959, Down's syndrome was found to be associated with the chromosomal disorder of trisomy (three chromosomes instead of the usual two). About 95% of cases are due to trisomy 21. These cases result from failure of disjunction during meiosis, and are associated with increasing maternal age. The risk of recurrence in a subsequent child is about 1 in 100. The remaining 5% of cases of Down's syndrome are attributable either to translocation involving chromosome 21 or to mosaicism.
- IQ is generally between 20 and 50, but in 15% of individuals it is above 50.

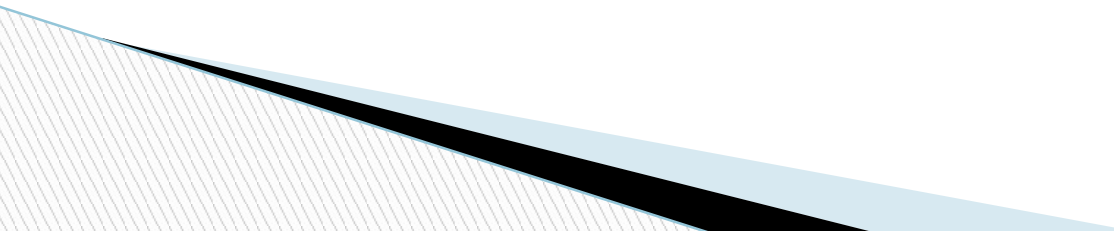
Features of Down's syndrome

- Moderate or severe learning disability
 - Placid temperament
 - Physical features
 - • Slanted eyes and epicanthic folds
 - • small mouth with furrowed tongue
 - • Flat nose
 - • Flattened occiput
 - • stubby hands, fingers, and single transverse palmar crease
 - • Hypotonia with hyperextensibility of joints
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❑ **Associated medical problems**

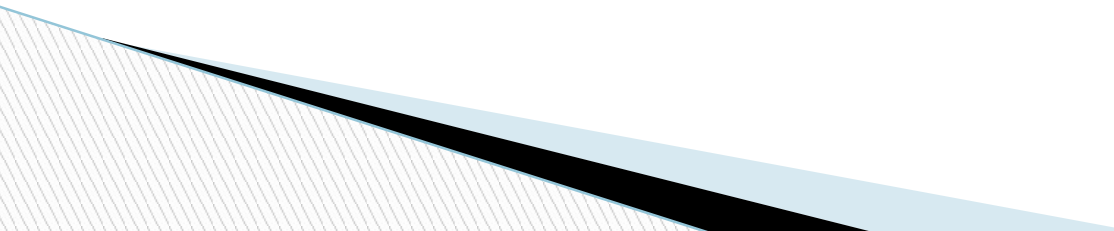
- ❑ • Cardiac anomalies, especially septal defects
 - ❑ • Gastrointestinal abnormalities
 - ❑ • Atlantoaxial instability
 - ❑ • Susceptibility to infection
 - ❑ • Impaired hearing
 - ❑ Increased risk of leukaemia, hypothyroidism, and autoimmune diseases.
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- Down's pathology is assumed to be due to the increased 'dosage' of genes, and thus increased production of the proteins that they encode. This probably also accounts for the frequent early onset of dementia in Down's syndrome, because the amyloid precursor protein (APP) gene that encodes p-amyloid and is central to Alzheimer's disease is located on chromosome 21.

Fragile X syndrome

- Fragile X syndrome is the second most common specific cause of learning disability after Down's syndrome, and is the most common inherited cause. It occurs in around 1 in 4000 males and in a milder form in about 1 in 6000 females. It accounts overall for about 10% of those with learning disability. The condition was so named because the X chromosome is 'fragile' when lymphocytes from affected individuals are cultured without sufficient folic acid

Features of fragile X syndrome

- More common in males
 - Caused FMR1 mutation
 - Learning disability
 - • Variable from mild to profound
 - • Increases late in childhood
 - • Poor attention and concentration
 - • Speech repetitive, lacking themes or content (litany speech')
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□ Behavioural features

- • Autistic features common

□ Physical features

- • Large, protruding ears
- • Long face with high-arched palate
- • Flat feet
- • Lax joints
- • Soft skin
- • Large testes (after puberty)
- • Mitral valve prolapsed.





Michael Phelps

FXS: Common Physical Features

- Elongated face & Broad forehead
- Large, prominent ears
- High arched palate
- Prominent jaw, Dental crowding
- Macro-orchidism (post-pubertal)
- Strabismus (squint)
- Murmur, Mitral valve prolapse, cardiomegaly, dilation of aorta
- Hypotonia & joint laxity
- Flat feet, Hollow chest, Scoliosis