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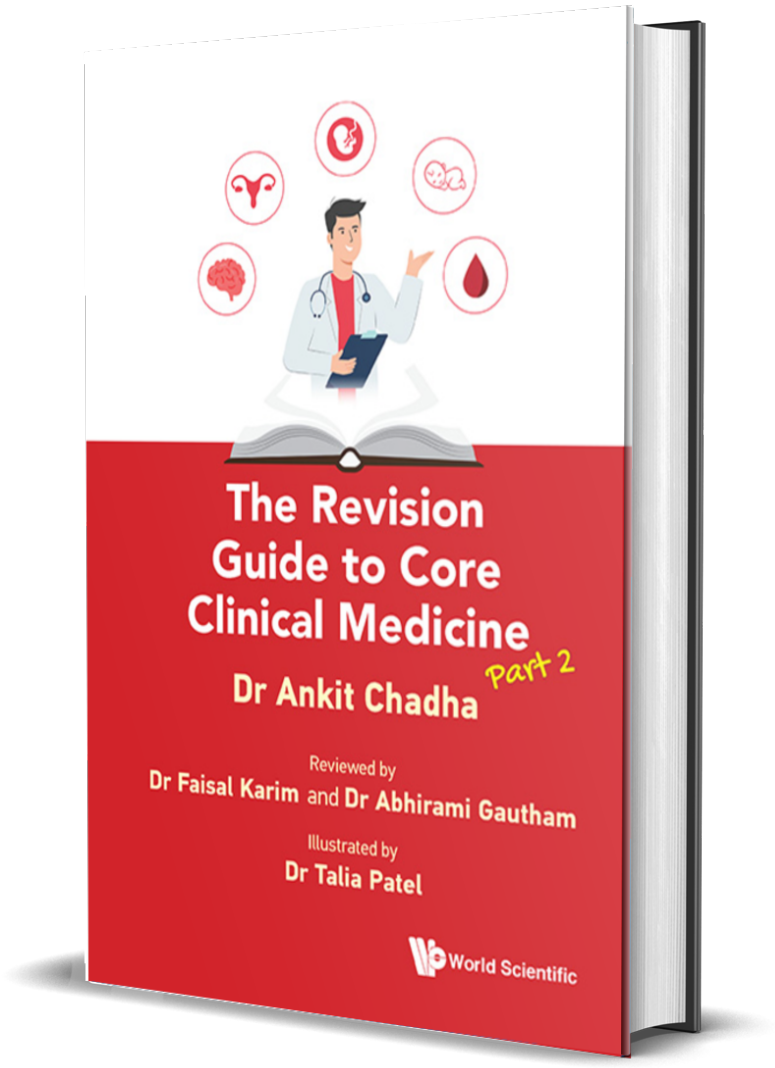

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The Revision Guide to Core Clinical Medicine

Part 2

Dr Ankit Chadha
University of Cambridge, UK

Reviewed by

Dr Faisal Karim and **Dr Abhirami Gautham**

Illustrated by

Dr Talia Patel

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Contents

Paediatrics

BACKGROUND

MEDICAL CONDITIONS

Neonatal Background

After birth, several physiological adaptations occur to allow neonates to transition from intrauterine to extrauterine life.

In utero, the foetal lungs are filled with fluid and are not involved in gas exchange.
- Pulmonary vessels are constricted, leading to high pulmonary vascular resistance.
- Most of the blood from the right side of the heart bypasses the lungs via two foetal shunts: the ductus arteriosus and the foramen ovale.

There are several changes that occur in the period just before and during labour.
- Production of lung liquid decreases.
- The infant's chest is compressed as it moves through the birth canal, helping to expel some fluid from the lungs.
- Breathing is triggered by several factors, including exposure to cooler temperatures and a surge in adrenaline.
- Catecholamines stimulate alveolar fluid reabsorption into the pulmonary circulation.
- Once the baby takes its first breath (typically within 6 seconds), remaining lung fluid is rapidly absorbed, and functional residual capacity is established.
- Regular breathing is usually achieved within 30 seconds of delivery.

After birth, there are several steps that are performed by medical professionals to optimise outcomes for the baby.

- **Cord clamping**

After the baby is born, the umbilical cord is clamped and cut.
- In term infants, delayed cord clamping (by 2–5 minutes) is recommended.
- This helps increase the neonate's circulating blood volume and reduces the risk of anaemia later in infancy.

- **Neonatal assessment – APGAR score**

The APGAR score assesses the baby's condition at 1, 5, and 10 minutes after birth.
- It may be repeated every 5 minutes if the condition remains poor.
- If the infant appears unwell, immediate drying and assessment should begin and the clock is started.

The APGAR score assesses the baby according to 5 criteria, each scored from 0–2.

- The five criteria are appearance (skin colour), pulse, grimace (reflexes), activity (muscle tone), and respiration.
- Scores are interpreted as follows, good condition is 7–10, moderately depressed is 4–6, severely depressed is 0–3.
- A low or decreasing APGAR score signals the need for urgent intervention and may require more intensive support.

Score	0	1	2
Heart rate	Absent	< 100 bpm	> 100 bpm
Respiratory effort	Absent	Gasping or irregular	Regular, strong cry
Activity	Flaccid	Some flexion of limbs	Well flexed, active
Reflex irritability	None	Grimace	Cry, cough
Appearance	Pale/blue	Body pink, extremities blue	Pink

- **Vitamin K injection**

This is given shortly after birth to prevent vitamin K deficiency bleeding (VKDB).

- This is a condition that can cause serious bleeding in newborns due to low levels of vitamin K-dependent clotting factors.

- **Newborn hearing screening**

All babies in the UK are offered an otoacoustic emission (OAE) test shortly after birth.

- If the result is abnormal, the baby is referred to a paediatric audiologist for further assessment (e.g. auditory brainstem response test).

- **Pulse oximetry screening**

This is performed in some hospitals across the UK within the first 24 hours of life.

- It measures oxygen saturation which can help detect critical, duct-dependent congenital heart disease.
- If oxygen saturations are abnormally low or there is a significant difference between pre- and post-ductal measurements, further assessment is needed including medical review and echocardiography.

- **Newborn and infant physical examination (NIPE)**

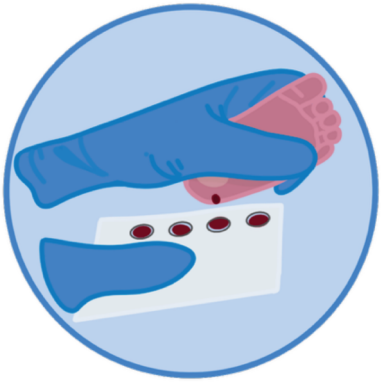
This is a screening examination which is performed on babies within 72 hours of birth, and then once again between 6 to 8 weeks.

- The purpose of the examination of the newborn is to screen for congenital abnormalities that will benefit from early intervention.
- It also helps to make referrals for further tests or treatment as appropriate as well as providing reassurance to the parents if the examination is normal.
- The main organs and tissues tests are the eyes, heart, hips and testes.

- **Newborn blood spot screening (Heel prick test)**

This is performed between days 5–8 of life.

- A small blood sample is taken from the baby's heel and screened for 9 conditions.
- You can divide the 9 conditions into groups to help remember them.



Blood disorder

- Sickle cell disease (SCD)

Congenital conditions

- Cystic fibrosis (CF)
- Congenital hypothyroidism

6 metabolic disorders

- Medium-chain Acyl-CoA dehydrogenase deficiency (MCADD)
- Maple syrup urine disease (MSUD)
- Phenylketonuria (PKU)
- Homocystinuria (HCU)
- Isovaleric acidaemia (IVA)
- Glutaric aciduria type 1 (GA1)

Vaccinations

Maternal antibodies begin to transfer to the foetus during the last trimester.
- However, at birth, this transfer stops, making infants susceptible to infections.
- Breast milk, particularly colostrum (the first milk), is rich in antibodies (mainly IgA).
- These IgA antibodies are transferred to the baby's digestive system.
- These antibodies help protect the baby's gut and fight infections.
- While maternal antibodies provide crucial early protection, a baby's own immune system gradually develops, becoming fully functional around 2–3 months of age.

In the UK, infants have a set vaccination schedule to help prevent various infections.

Age	Diseases protected against
8 weeks	- 6 in 1 (diphtheria, tetanus, pertussis, polio, Haemophilus influenzae type B (Hib), Hepatitis B) - Meningococcal group B (Men B) - Rotavirus
12 weeks	- 6 in 1 (diphtheria, tetanus, pertussis, polio, Hib, Hep B) - Pneumococcal (13 serotypes) - Rotavirus
16 weeks	- 6 in 1 (diphtheria, tetanus, pertussis, polio, Hib, Hep B) - Meningococcal group B (Men B)
1 year old (12 months)	- HiB and Men C - Pneumococcal - Measles, mumps, rubella (MMR) - MenB
3 years 4 months*	- 4 in 1 booster (diphtheria, tetanus, pertussis, polio) - MMR (2nd dose)
12–13 years old	- Human papillomavirus (HPV) types
14 years old	- 3 in 1 booster (diphtheria, tetanus, polio) - Meningococcal groups A, C, W and Y

*For children born after 1st July 2024, at 18 months, they get a 4th dose of the 6-in-1 vaccine and 2nd dose of the MMR vaccine (rather than at 3 years 4 months).
**From January 2026, a chickenpox vaccine is being offered on the NHS. The vaccine is a MMRV jab, protecting against measles, mumps, rubella, and chickenpox.
- It is given in two doses as part of the routine immunisations at 12 and 18 months.

Developmental Milestones

Developmental milestones are a set of functional skills or age-specific tasks that most children can do at a certain age range.

- They are used by paediatricians to assess a child's growth and development across several domains. Certain diseases can lead to delays in one or more of the domains.

There are 4 main domains used to assess development.

- Gross motor skills – this refers to large movements using the arms, legs, or the whole body (e.g. crawling, walking, jumping).
- Fine motor skills – this refers to small movements using hands and fingers (e.g. grasping, drawing, using utensils).
- Speech and language – this refers to an ability to understand and use language to communicate (e.g. babbling, saying words, following instructions).
- Social – this refers to a child's interaction with others, including caregivers, other children and their ability to express feelings and develop relationships.

The main developmental milestones and key ages are summarised below.

- Failure to reach these milestones at the correct age should be taken seriously as it can be an early sign of a disease and warrants referral to a paediatrician.

6 weeks

Gross motor: Stabilises head in sitting position

Fine motor: Can track object/face

Speech: Startles at loud noise

Social: Smiles

6 months

Gross motor: Can sit up briefly/with support, and roll over from prone to supine

Fine motor: Develops palmar grasp, reaches for objects

Speech: Babbles two syllable sounds, turns to their name

Social: Shakes rattles, reaches for bottle and puts objects to mouth

9 months

Gross motor: Sits up steady, attempts crawling, stands up holding on

Fine motor: Index finger poke, learns object permanence

Speech: Responds to their name, understands "no", imitates sounds

Social: Finger feeds, apprehensive to strangers

12 months (1 year)

Gross motor: Can stand alone and starts walking (anytime 9–18 months)

Fine motor: Develops pincer grip, casts bricks (should disappear by 18 months)

Speech: Can say 3 words, shows an understanding of nouns (e.g. mummy)

Social: Waves "bye-bye", claps, expresses desires with pointing

18 months

Gross motor: Can run and jump

Fine motor: Can draw to and fro, builds tower of 4 blocks

Speech: Can say 1–6 different words, understands nouns

Social: Imitates activities

2 years

Gross motor: Runs tiptoe, walks upstairs both feet each step, throws ball

Fine motor: Draws a vertical line, builds tower of 8 blocks

Speech: Shows understanding of verbs, uses 2 words joined together

Social: Eats with fork/spoon

3 years

Gross motor: Hops on one foot, starts doing stairs one foot per step

Fine motor: Copies drawing a circle, builds a bridge with blocks

Speech: Understands adjectives and negatives

Social: Uses knife and fork, shares toys with friends, plays alone without parents

4 years

Gross motor: Can walk upstairs and downstairs in adult manner

Fine motor: Cuts paper in half, copies cross, draws man with 3 parts

Speech: Counts to 10, understands complex instructions

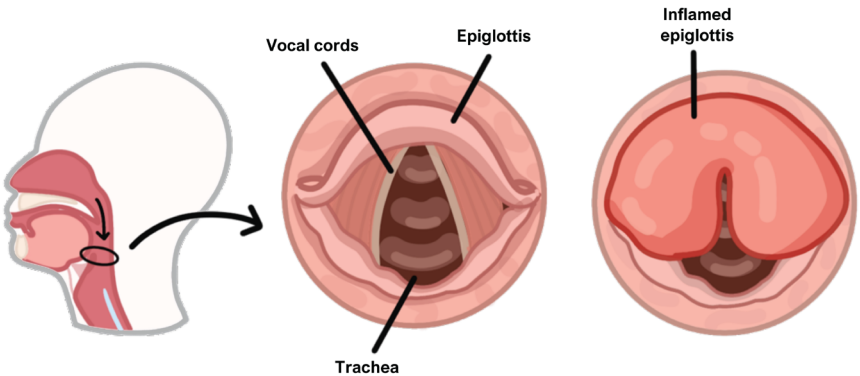
Social: Shows concern for others hurt, has a best friend

Respiratory Infections

- **Acute epiglottitis**

This refers to inflammation of the epiglottis, which typically has been caused by the bacterium Haemophilus influenzae type B.

- It should be recognised and treated quickly as it can lead to airway obstruction.
- It usually presents in children, but Haemophilus influenzae type B vaccination has meant that it is rare in children and now increasingly seen more in adults.
- Care should be taken when assessing a patient's throat due to risk of airway obstruction.



Symptoms

- Rapid onset of high fever and malaise
- Drooling of saliva
- Muffled voice due to very sore throat
- Inspiratory stridor, a high-pitched sound due to turbulent air flow in the upper airway
- Tripod position – this is a compensatory position where a child leans forward, extends the neck, supports themselves with hands or knees. This position helps maximise airway patency and ease breathing in severe upper airway obstruction

Key tests

- Clinical diagnosis after visualising throat (should only be done by experienced physician or ENT specialist)

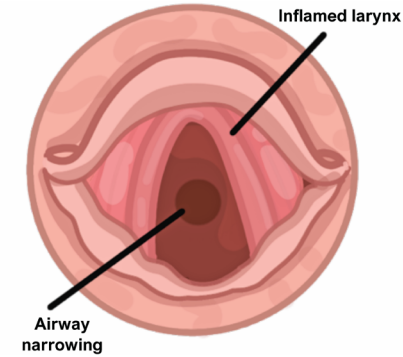
Management

- This is a medical emergency as there is a high risk of upper airway obstruction
- Anaesthetics involvement to secure airway if signs of compromise
- Management includes IV fluids, antibiotics and oxygen

- **Croup (laryngotracheobronchitis)**

This is an infection of the upper airway seen in infants and toddlers less than 3 years old.

- It causes inflammation and swelling in the larynx, trachea, and bronchi.
- It is common particularly in the autumn months.
- It is usually due to a virus, with parainfluenza virus the most common cause.



Symptoms

- Inspiratory stridor and hoarseness of voice
- Barking cough (worse during the night)
- High fever, with coryzal symptoms
- If severe can cause respiratory distress e.g. chest wall recession (Hoover's sign)

Grading

- The following criteria are used to grade the severity of croup

	Mild	Moderate	Severe
Cough	Occasional	Frequent	Frequent
Stridor	None at rest	Easily audible at rest	Prominent at rest
Recession	None/mild	Suprasternal and sternal retraction at rest	Marked sternal retraction
Behaviour	Happy, eats, drinks and plays	Little distress or agitation Interested in surrounding	Significant distress and agitation Lethargy

Key tests

- It is diagnosed clinically

Management

- As presentation can be similar to epiglottis, it is necessary to rule this out
- Steroids (oral dexamethasone) are the mainstay of management
- If severe, < 6 months of age or known upper airway abnormality, then admit to hospital, may need to administer oxygen and nebulised adrenaline

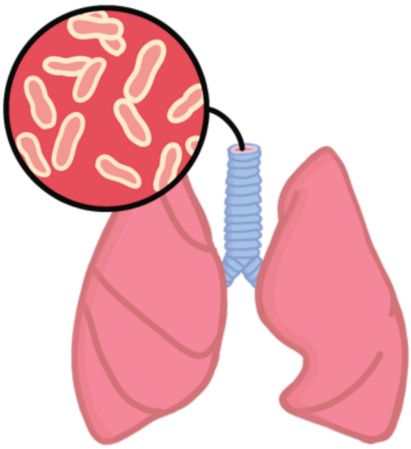
- **Whooping cough**

A condition which is caused by the Gram-negative bacterium Bordetella pertussis.

- It is a notifiable disease, which means that it is required to report confirmed cases to the government authorities.
- Although pregnant mothers are immunised during pregnancy and infants are vaccinated during childhood, these vaccines do not provide lifelong protection.
- Symptoms can last up to 10–14 weeks and are more severe in infants.

Symptoms

- Few days of coryzal symptoms initially
- Sudden coughing attacks with a distinctive inspiratory whoop, caused by forced inspiration against a closed glottis
- Coughing episodes which are followed by vomiting, more frequent at night and following meals
- Complications include rib fractures and pneumothorax
- Subconjunctival haemorrhages
- Apnoea, which can lead to cyanosis, syncope and seizures if severe



Key tests

- Nasopharyngeal aspirate or swab – this is sent for culture and PCR
- Serological testing for antibodies – a serum anti-pertussis toxin antibody concentration of > 100 IU/mL is suggestive of infection
- Blood tests show elevated white cell count and lymphocytes

Management

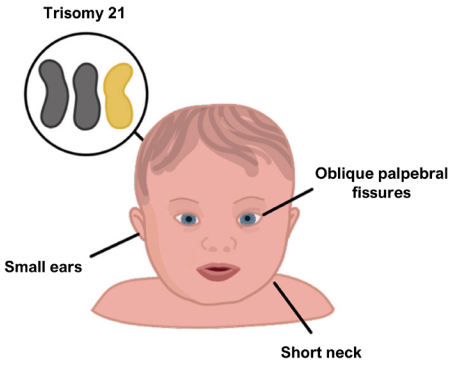
- 1st line treatment is an oral macrolide (azithromycin or clarithromycin) if onset of cough is within previous 21 days
- If patient is less than 6 months old, will usually require hospital admission
- Prophylactic antibiotics should be given to household contacts
- School exclusion for 48 hours after starting antibiotics

Trisomy Conditions

- **Down's syndrome**

This condition is caused by the presence of an extra copy of chromosome 21.
- This results in a number of physical and developmental complications.
- It is caused by non-disjunction during meiosis which produces a sex cell (sperm or egg) with 2 copies of chromosome 21 rather than 1.
- When fertilisation occurs, this results in 3 overall copies of chromosome 21.

However, in some cases it can also be due to a Robertsonian translocation.
- A translocation is a type of structural chromosomal abnormality where part of one chromosome breaks off and attaches to another chromosome.
- A Robertsonian translocation is a specific type of translocation involving acrocentric chromosomes (chromosomes with the centromere near one end).
- These chromosomes (13, 14, 15, 21, and 22) have a short arm and a long arm.



In a Robertsonian translocation, the long arms of two acrocentric chromosomes fuse, and the short arms are usually lost.
- If the long arm of chromosome 21 fuses with the long arm of another acrocentric chromosome (like 14 or 15), and the individual inherits this fused chromosome along with a normal chromosome 21, they will have three copies of the long arm of chromosome 21 (trisomy 21), resulting in Down's syndrome.

The likelihood of Down's syndrome increases significantly with maternal age.

Maternal age (years)	20	30	40	45
Risk	1 in 1500	1 in 800	1 in 100	1 in 50

Symptoms

- Intellectual – learning disability, features of autism, delayed developmental milestones, early-onset Alzheimer's disease
- Facial – flat facial profile, small ears, large protruding tongue, oblique palpebral fissures, Brushfield spots in the iris
- Respiratory – recurrent respiratory tract infections
- Cardiac – endocardial cushion defect (most common), ventricular septal defect, secundum atrial septal defect, patent ductus arteriosus, tetralogy of Fallot
- Gastrointestinal – duodenal atresia, Hirschsprung's disease
- Limbs – single palmar crease, wide sandal gap between first and second toes, hypotonia, short stature
- Endocrine – hypothyroidism
- Reproductive – subfertility in both males and females

Key tests

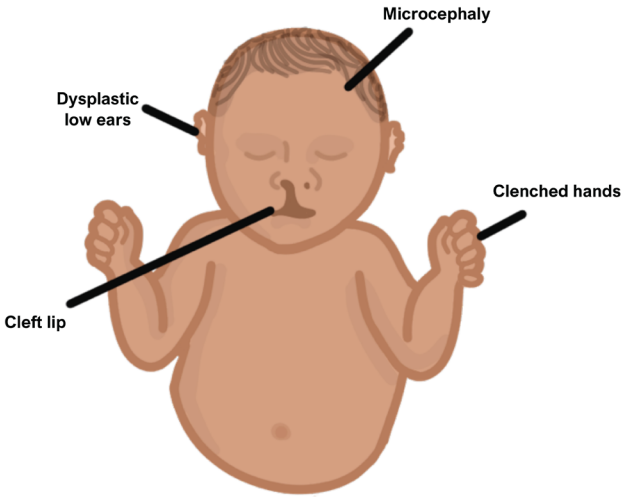
- Diagnosis is via chromosomal analysis (karyotyping)

Management

- No cure, requires lifelong MDT support and management of complications

- **Patau syndrome**

This is caused by the presence of an extra copy of chromosome 13 (trisomy 13).



Symptoms

- Head and neck – scalp lesions, cleft lip or palate, microcephaly
- Eyes – microphthalmia, cataracts, retinal detachment
- Limbs – polydactyly, clenched hands
- Congenital heart defects – dextrocardia
- Urogenital – abnormal external genitals, renal abnormalities

Management

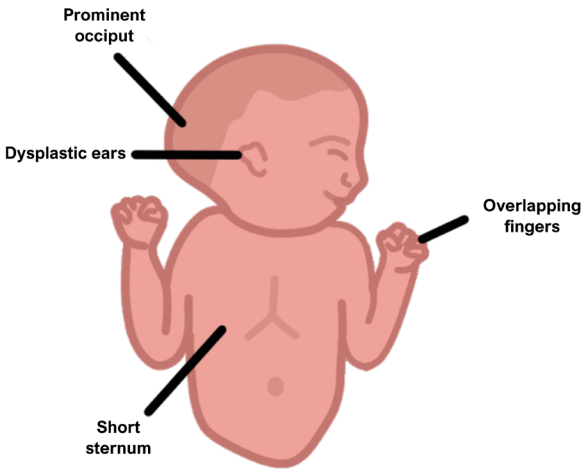
- No cure available. Most (90%) children die within the first year of life

- **Edwards' syndrome**

This condition is caused by the presence of an extra copy of chromosome 18.

Symptoms

- Head and neck – prominent occiput, low-set ears, micrognathia
- Chest – short sternum, oesophageal atresia
- Limbs – flexed and overlapping fingers, rocker-bottom feet
- Heart defects – ventricular septal defect, atrial septal defect, patent ductus arteriosus



Management

- There is no cure available, manage complications with an MDT approach
- Most pregnancies affected do not result in a live birth, and 90% of children die within the first year of life

Sex Chromosome Conditions

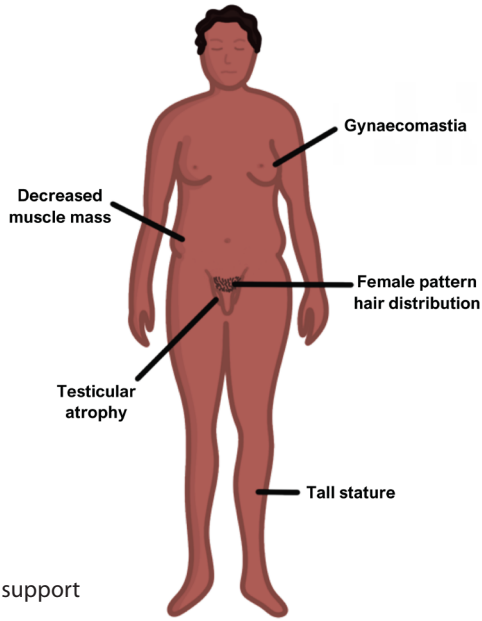
- **Klinefelter's syndrome**

This condition results from the presence of an additional sex chromosome, giving a karyotype of 47, XXY.

- Individuals are genetically male due to the presence of the Y chromosome.
- However, they may have a range of physical and reproductive abnormalities.
- It is caused by nondisjunction of chromosomes during meiosis in either parent, resulting in an extra X chromosome in the zygote.

Symptoms

- Body – tall stature
- Feminisation – gynaecomastia (increased risk of breast cancer), reduced body hair, decreased muscle mass
- Genitals – small, underdeveloped testicles, reduced libido
- Endocrine – lack of secondary sexual characteristics and infertility



Key tests

- Blood tests typically show elevated FSH and LH, with low testosterone
- Chromosomal analysis (karyotyping)

Management

- Testosterone replacement therapy to support development of male characteristics
- Infertility treatment such as ICSI or IVF may help with reproduction
- Breast tissue may be surgically removed due to increased breast cancer risk

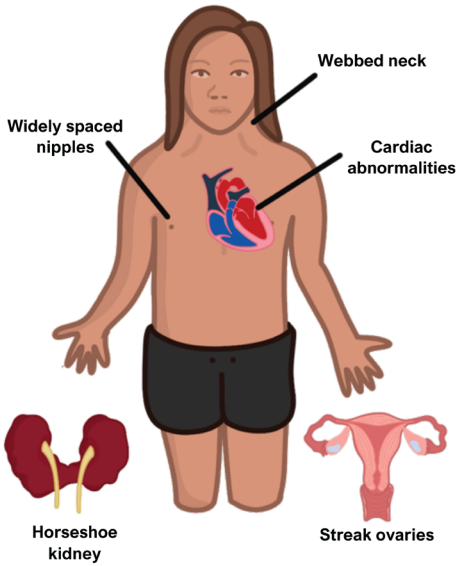
- **Turner's syndrome**

This condition results from the absence of one sex chromosome, giving the karyotype 45, XO.

- Without a Y chromosome, individuals are female and have female sexual organs.
- It occurs due to the complete loss of one X chromosome, or deletion of the short arm of one X chromosome.

Symptoms

- Antenatal – early miscarriage, foetal hydrops, cystic hygroma
- Body – short stature, multiple pigmented naevi
- Head and neck – webbed neck, high-arched palate
- Chest – widely spaced nipples, broad shield-shaped chest
- Limbs – spoon-shaped nails, lymphoedema of hands and feet
- Heart – bicuspid aortic valve, coarctation of the aorta
- Ovaries – ovarian dysgenesis leading to primary amenorrhoea



Complications

- Endocrine – primary amenorrhoea, hypothyroidism
- Autoimmune – increased risk of conditions such as Crohn's disease
- Infertility due to underdeveloped ovaries

Key tests

- Karyotyping is diagnostic, this can be performed antenatally (via amniocentesis or chorionic villus sampling) or postnatally

Management

- No cure available; treatment focuses on symptom management
- Growth hormone therapy is often used to promote height development
- Oestrogen replacement for the development of secondary sexual characteristics

- **Swyer syndrome**

This is a rare condition caused by a mutation in the SRY gene on the Y chromosome.

- The SRY gene is essential for the masculinisation of the embryo. Without its function, the undifferentiated gonads fail to develop into testicles, even in an XY foetus.
- As a result, individuals with Swyer syndrome have an XY karyotype but develop as phenotypic females, with a uterus, fallopian tubes, and vagina.
- They are born with a female appearance but have non-functional streak gonads.
- During puberty, the lack of functional gonads means insufficient oestrogen is produced, leading to absent breast development and primary amenorrhoea.

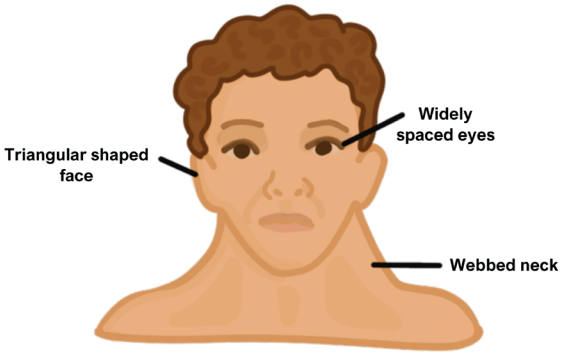
Management

- Oestrogen and progestogen hormone replacement therapy is required to induce and maintain secondary sexual characteristics and menstrual cycles

- **Noonan syndrome**

This is a genetic condition often referred to as the male version of Turner's syndrome.

- However, unlike Turner's syndrome, the mutation is not on the sex chromosomes but is autosomal dominant, usually due to a mutation in the PTPN11 gene on Chr 12.



Symptoms

- Face – low-set ears, triangular-shaped face, large head with excess nuchal skin
- Eyes – hypertelorism (widely spaced eyes)
- Skin – lymphoedema
- Body – webbed neck, short stature, widely spaced nipples
- Heart – pulmonary valve stenosis (most common cardiac defect)

Management

- Multidisciplinary approach to manage complications

Specific Gene Conditions

- **Fragile X syndrome**

This condition is caused by a trinucleotide (CGG) repeat expansion in the FMR1 gene.

- The FMR1 gene encodes the FMRP protein, which is most concentrated in the brain and testes and plays a role in transporting mRNA to the nucleus and synapses for neural development.

Inheritance

- X-linked dominant, more commonly affecting males
- Females are less affected due to random X-chromosome inactivation
- The condition shows variable expressivity, meaning that people with the same mutation may experience the condition differently, with varying degrees of severity
- The condition also shows genetic anticipation, a phenomenon where the age of onset and/or severity of symptoms of a genetic disorder become more pronounced in successive generations of a family

In unaffected individuals, the FMR1 gene has 5–44 CGG repeats

- 45–54 repeats is a grey zone
- 55–200 repeats is known as a premutation. Women with a premutation have an increased risk of having a child with the full mutation
- People with > 200 repeats have the full mutation, which results in the syndrome

Symptoms

- Intellectual – learning difficulties, autism spectrum disorder
- Facial – long face, large protruding ears, high-arched palate
- Behavioural – social anxiety, hypersensitivity, hand flapping, biting, poor eye contact
- Other features – macroorchidism, hypotonia, mitral valve prolapse

Key tests

- Diagnosis is by PCR and Southern blot to detect the CGG repeat expansion
- It can be made antenatally via chorionic villus sampling (CVS) or amniocentesis

Management

- No cure is available. Mainstay of management is multidisciplinary support including behavioural therapy, speech and language therapy, and educational interventions

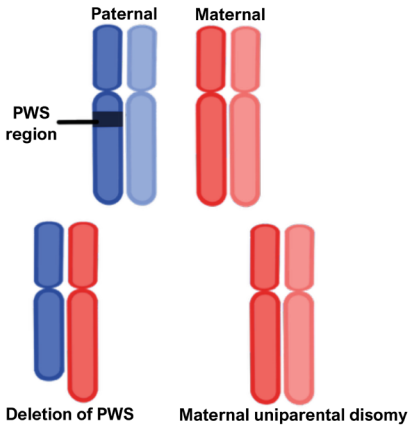
- **Prader-Willi syndrome (PWS)**

This is a genetic condition caused by the loss of function of genes in the PWS region of chromosome 15 (15q11–13).

- The condition is a prime example of genetic imprinting, where gene expression depends on parental origin of the chromosomes.
- The absence of a functioning paternal copy of the gene in the PWS region leads to Prader-Willi syndrome.
- The absence of a functioning maternal copy results in Angelman syndrome.

There are two main mechanisms by which the condition can arise:

- Deletion of the PWS region on the paternal chromosome (most common).
- Uniparental disomy – this is where the child inherits two maternal copies of chromosome 15 and no paternal copy, due to errors in meiosis.
- The maternal chromosomes retain their imprint, meaning that the PWS region on these chromosomes remains inactive.
- As a result, no PWS genes are expressed and the child develops the syndrome.



Symptoms

- CNS – developmental delay, learning difficulties, behavioural issues in adolescence
- Appetite – hyperphagia, leading to obesity and type 2 diabetes
- Short stature with hypotonia
- Distinctive facial features
- Hypogonadism and infertility

Management

- No cure; treatment focuses on managing symptoms
- Restricting access to food is essential to prevent severe obesity

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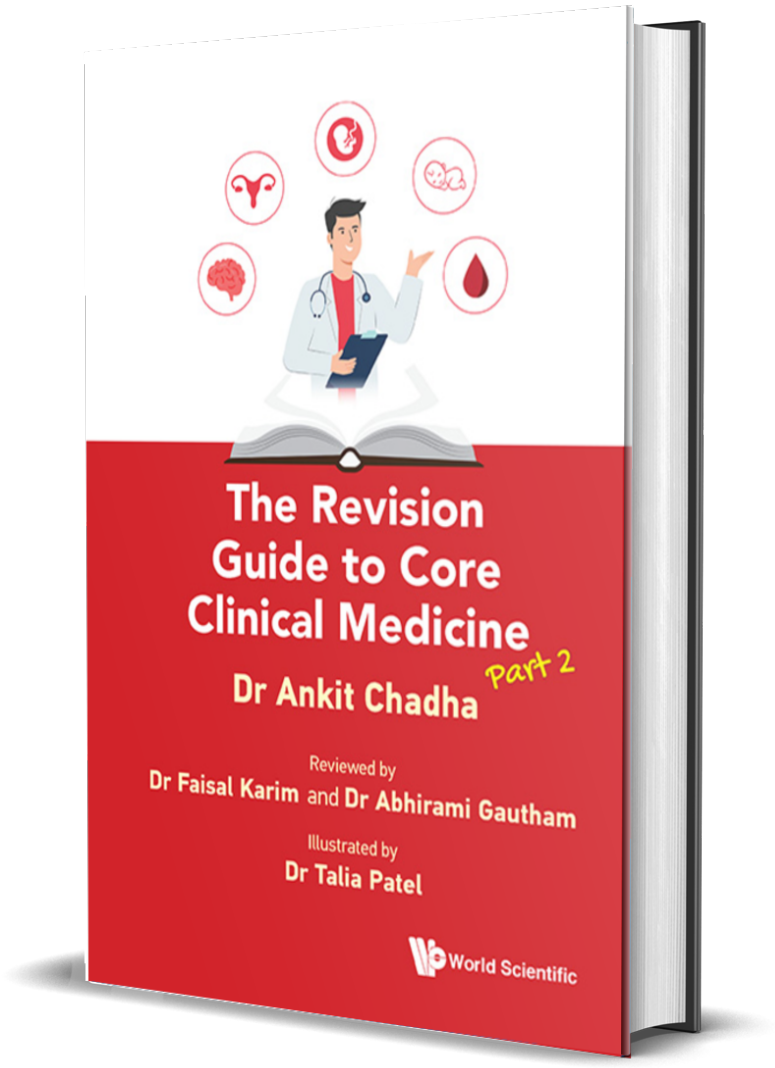

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