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Editor
Indumathy Santhanam

Foreword
T Dorairajan

Illustrated Textbook of
PEDIATRICS

Highlights

- Exclusive chapter on Pediatric Resuscitation and Emergency Medicine
- Presented with more than 650 clinical photographs
- Includes color atlas on how to perform neurological examination of an infant
- Attractive history panel with every chapter









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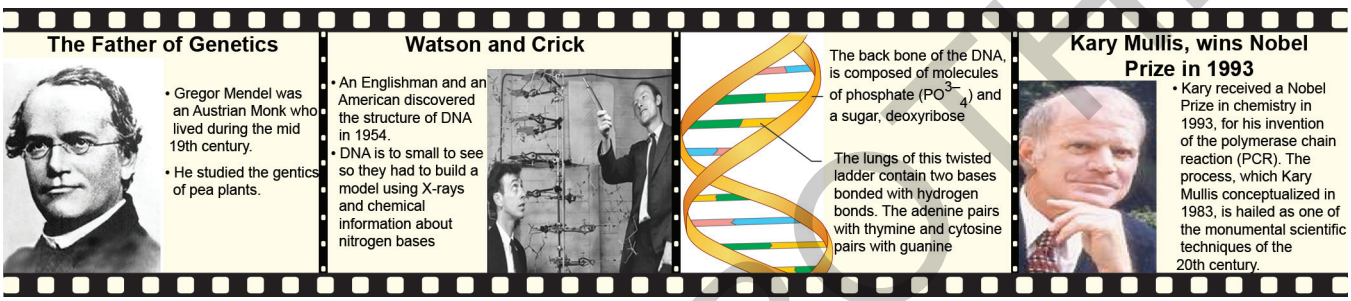
CHAPTER

3

Genetics

Srilakshmi Rajagopalan



OBJECTIVES

1. To understand heredity and the role chromosomes play in causing structural and functional abnormalities
2. Gain or loss of chromosomes can result in disease

DEFINITION

Genetics is the study of heredity that is grounded on the combined understanding of the structure of the genome, the DNA sequence, and its phenotypic expression in an organism (Fig. 3.1). The foundation for the science of genetics was first laid by an Austrian monk Gregor Mendel in the latter half of the 19th century.

The term "Mendelian" is given to inherited diseases resulting from defects of individual genes.

MENDEL'S LAWS OF INHERITANCE

Two identical genes are referred to as homozygous whereas each gene with a different characteristic is referred to as heterozygous. The genes responsible for these characteristics are called "alleles".

Mendel derived three laws area as follows:

1. *Unit inheritance*: It states that when two homozygotes with different alleles are crossed, all off-springs in the first generation are identical and heterozygous. The characteristics will manifest in later generations.
2. *Segregation*: Each individual possesses two genes for a particular characteristic. However, only one of these genes can be transmitted at any given time. Very rarely, both the allelic genes may fail to separate due to chromosome nondisjunction during the first meiotic division.
3. *Independent assortment*: Allele pairs separate independently during the formation of gametes. This results in transmission of traits to offspring independent of one another.

The human genome project was completed successfully in 2001.

It is currently estimated that the human genome contains 30,000–35,000 genes, although the function of many of these genes remain unknown.

Clinical application of these advances is available to families through specialized genetic centers, that

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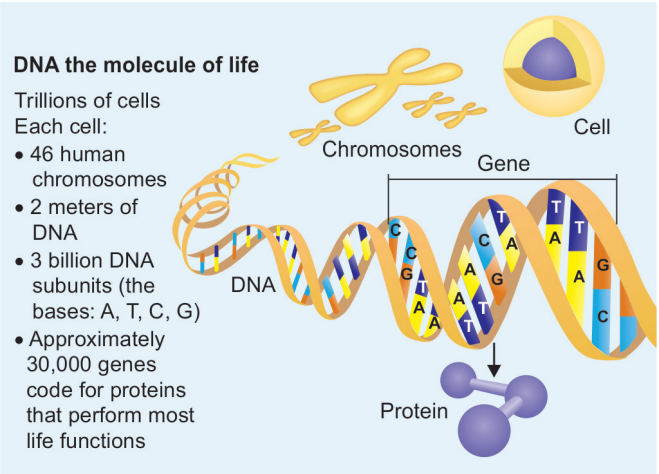

Fig. 3.1: Shows the structure of the DNA.

offer diagnosis, investigation, counseling, and antenatal diagnosis for an ever widening range of disorders. Genetic databases, such as "Online Mendelian Inheritance in Man" (OMIM) and London dysmorphology database have lead to an explosion in knowledge about the basis of genetic diseases.

GENETIC DISORDERS

Genetic disorders are classified as:

- Single mutant genes disorder (Mendelian inheritance):
  - Recessive
  - Dominant
  - X-linked
- Mitochondrial
- Chromosomal disorder
- Multifactorial diseases

Genetic disorders are common in 2% of liveborn babies with significant congenital malformations. Affected individuals are a burden to the family and society, since many are associated with severe and permanent disability.

Autosomal Recessive Inheritance

- The disease characteristically manifests in siblings. Parents and other relative are normal.
- The risk of being affected is as high as 1 in 4 or 25%. Consanguinity is more likely in rare recessive disorders. In the more common conditions, parents are usually unrelated. Both sexes are affected.

A classical pedigree is shown in Figure 3.2.

Examples of autosomal recessive (AR) diseases are as follows:

- Congenital adrenal hyperplasia

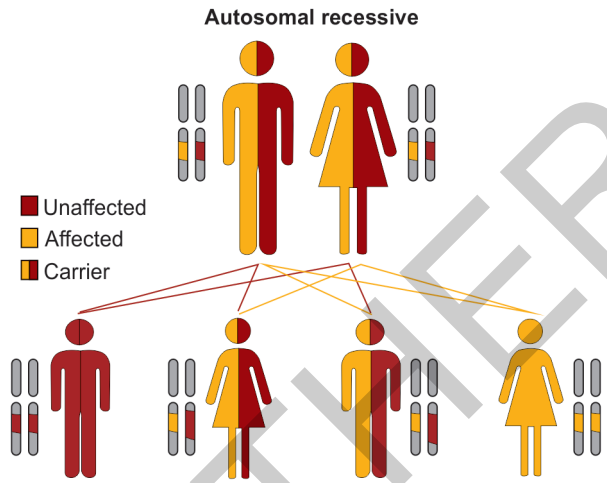

Fig. 3.2: Autosomal recessive inheritance showing how offspring is affected if both parents are carriers.

- Cystic fibrosis (CF)
- Friedreich's ataxia
- Galactosemia
- Glycogen storage diseases
- Hurler's syndrome
- Oculocutaneous albinism
- Phenylketonuria (PKU)
- Sickle cell disease
- Tay Sachs disease
- Thalassemia
- Werdnig Hoffmann disease [spinal muscular atrophy (SMA) type I].

If an individual carries a few abnormal genes, the possibility of marrying someone carrying the same abnormal gene is considerably increased when marrying a relative, particularly a first cousin.

This is due to the fact that cousins share one-eighth of the genes.

Autosomal Dominant Inheritance (Fig. 3.3)

Affects structural proteins causing disorders of connective tissue.

Example: Marfan syndrome

- Every child born of the affected parent has 50–50 chance of inheriting the affected gene.
- Severity may vary in the affected offspring.
- Homozygotes for the dominant gene die prenatally.

Inheritance of an AD gene from both parents is lethal!

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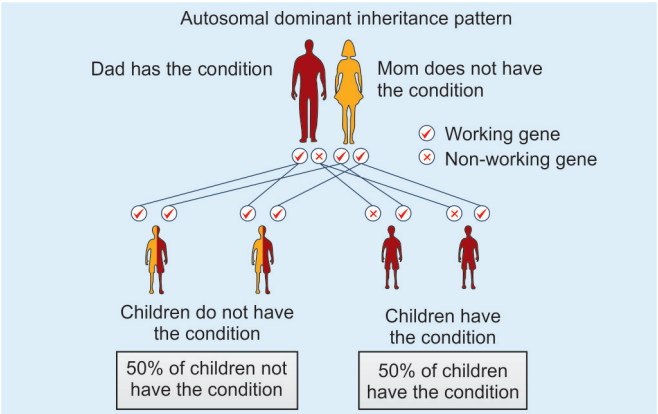

Fig. 3.3: Each generation is affected. Both sexes are equally affected.

- An affected person always has affected parents with the exception of new mutation.
- Both sexes are affected, although there may be considerable variation in the clinical severity of the disease.

Examples of autosomal dominant (AD) diseases are:

- Achondroplasia
- Ehlers-Danlos syndrome
- Familial hypercholesterolemia
- Huntington's disease
- Marfan's syndrome
- Myotonic dystrophy
- Neurofibromatosis
  - Noonan's syndrome
  - Osteogenesis imperfecta
  - Otosclerosis
  - Polyposis coli
  - Tuberous sclerosis.

X-linked Recessive Inheritance (Fig. 3.4)

The abnormal trait is transferred from the carrier female to her male offspring.

- Sons have a 50–50 chance of inheriting the gene and therefore, developing the disease, whereas daughters have a 50–50 chance of becoming a carriers like their mother.
- An affected male cannot transmit the disease to his sons. However, all his daughters can become carriers. This is a rare possibility, since it implies that the affected male survives into adulthood and is capable of reproduction.
- Daughters of affected males can pass the disease to their sons.

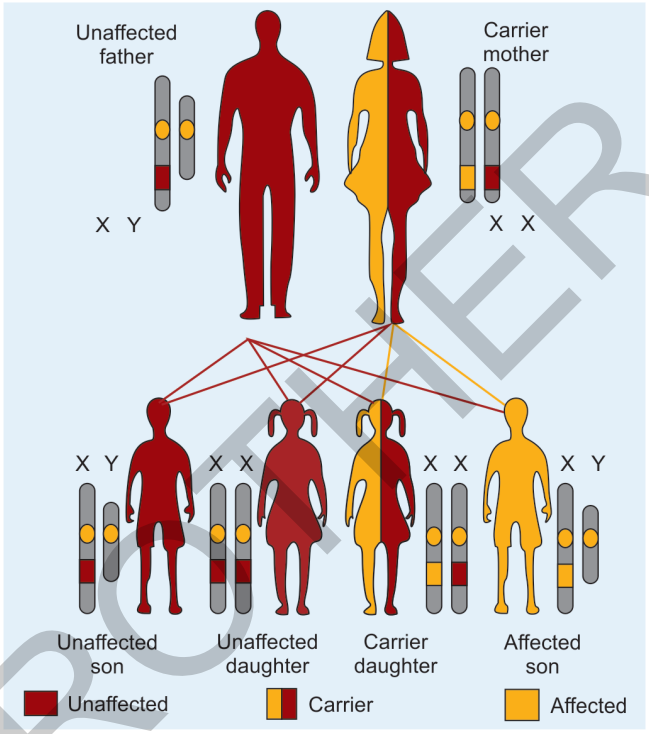

Fig. 3.4: X-linked recessive inheritance.

Females may occasionally demonstrate clinical evidence of the disease in some X–linked diseases. This is explained by the following phenomenon:

Lyon's hypothesis describes inactivation of the X chromosome, just after formation of zygote.

Examples of X-linked recessive disorders:

- Hemophilia A and B
- Duchenne's and Becker's muscular dystrophies
- Diabetes insipidus
- Fragile X syndrome
- Glucose-6-phosphate dehydrogenase (G6PD) deficiency
- Hunter's syndrome (mucopolysaccharidosis type II)
- Color blindness (red–green)
- Hypophosphatemic rickets.

X-linked Dominant Inheritance

Diseases are common in females, but more severe in males. Both sons and daughters have 50–50 chance of inheriting the gene from an affected female and, therefore, developing the disease.

- An affected male can transmit the disease to all his daughters but not his sons.

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Examples of X–linked dominant inheritance:

- Variant of vitamin D-resistant rickets
- Rett's syndrome
- Incontinentia pigmenti

Mitochondrial Inheritance

The transmission of the mitochondrial genome occurs from mother to child. Mitochondria contain their own set of genes that are responsible for metabolic pathways.

Mitochondrial genes are exclusive of the genes in the cell's nucleus.

Alterations in the mitochondrial DNA cause several well-recognized disorders and are often associated with neuromuscular features. Mutations of mitochondrial DNA have peculiar pattern of inheritance.

Although the disease affects both boys or girls, it is exclusively inherited from the mom! The affected male does not pass on his mitochondria to his children. Hence, mitochondrial disorders have a matrilineal inheritance.

Some mitochondrial conditions, however, are associated with incorrectly functioning mitochondria wherein the mutation occurs not only in the mitochondrial DNA, but also in genes within the cell nucleus that control functions in the mitochondria. It is therefore possible that mitochondrial conditions may not demonstrate matrilineal inheritance but Mendelian inheritance. Examples of mitochondrial diseases are:

- Leigh disease
- Lethal infantile cardiomyopathy
- Lactic acidosis
- Mitochondrial encephalomyopathy lactic acidosis and stroke-like episodes (MELAS).

Blood Groups

The ABO gene is autosomally inherited.

The gene responsible for ABO groups is located on chromosome 9.

- A and B blood group genes are dominant over the O blood group genes. On the contrary, both genes responsible for A and B groups are codominant (Fig. 3.5).

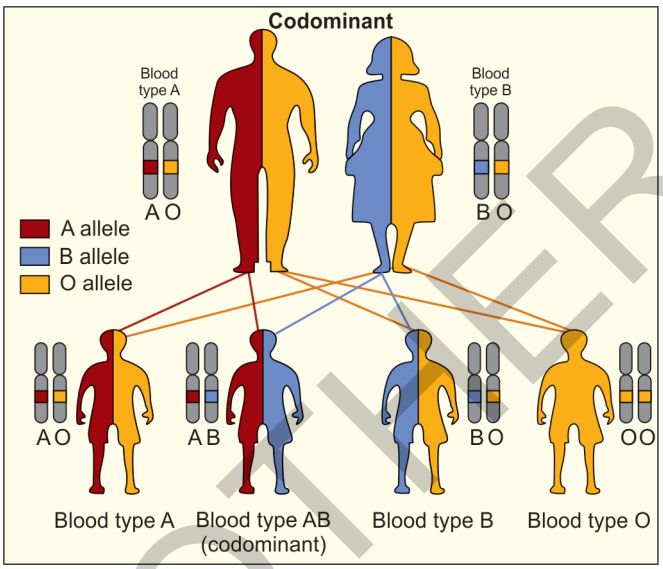

Fig. 3.5: Codominant inheritance.

- Each person has "two copies of genes" coding for their ABO blood group (one maternal and one paternal in origin).

GENETIC TESTING

Newborn Screening

Screening for carrier detection is essential for early recognition and management. Neonatal screening is routinely performed in developed countries. Recently, India has also initiated newborn screening services.

Diagnostic Test

A diagnostic test is useful for confirming the diagnosis that has been suspected on a clinical grounds, e.g. Duchenne muscular dystrophy (DMD) gene testing in a family with an index case of DMD.

Carrier Test

Carrier test is undertaken to determine the risk of an inherited disorder. The result has no implication for the health of the individual. E.g. North Eastern states of India have a high carrier status for β-thalassemia gene.

Predictive Test

A predictive test is undertaken in individuals who are normal but who have increased risk of developing the familial disease in the future.

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Preimplantation Genetic Diagnosis (PIGD)

Embryos are generated by in vitro fertilization (IVF).

During the 8–16 cell stage, a single cell is removed and tested for a specific genetic disorder.

This procedure is performed in embryos from IVF technique. The healthy embryos are taken for embryo transfer. Techniques are for genetic testing or for preimplantation genetic diagnosis (PIGD):

Karyotyping (Fig. 3.6)

Karyotyping is the method in which a photomicrograph is taken of an individual's chromosomes. The latter are arranged in a standard format that shows the number, size, and shape of each chromosome.

Fluorescent In-situ Hybridization (FISH)

Single-stranded DNA probes are used to identify complementary sequences in immobilized chromosomes in situ. Since, the probes are visualized with a fluorescent dye, the procedure is termed as fluorescent in-situ hybridization (FISH).

FISH is useful in the detection of microdeletion in chromosomes.

Prenatal Diagnosis

Prenatal testing aims at detecting various anomalies such as structural, chromosomal, and genetic disorders in the fetus or the embryo before its birth, while it is still in utero. Some of these anomalies include neural tube defects, chromosomal anomalies, genetic disorders (cystic fibrosis, Tay-Sachs disease, thalassemia, sickle cell anemia, etc.), and other anomalies such as spina bifida, cleft lip, etc. Prenatal diagnostic procedures can be divided into two categories: (1) invasive and (2) noninvasive procedures. Common noninvasive procedures include ultrasound examination and maternal serum screens (e.g. alpha-fetoprotein levels in the serum). The invasive tests include procedures such as amniocentesis, chorionic villus biopsy and cordocentesis. Both these procedures help in providing fetal cells which can be used for determining fetal karyotype.

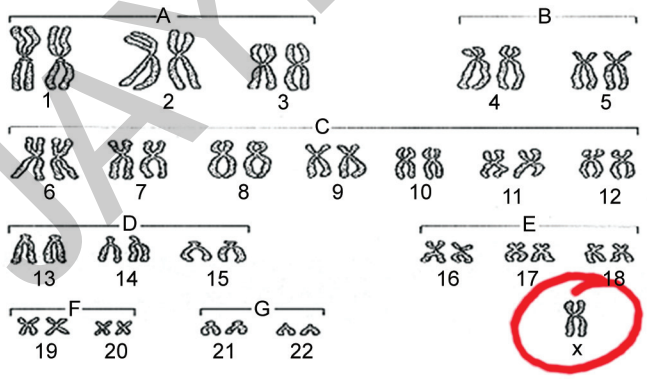

Fig. 3.6: 45-XO karyotype of Turner's syndrome.

Mutation Analysis

Direct analysis of DNA is used for specific detection of deletions and mutations. Microarray and comparative genomic hybridization are the other molecular diagnostic methods. Currently, genome sequencing has revolutionized cancer chemotherapy, inherited disorders, and birth defects (Boxes 3.1 and 3.2).

SELECTED CHROMOSOMAL DISORDERS

Few of the selected chromosomal disorders have been discussed in Box 3.1.

Down's Syndrome

Down's syndrome is the most common and best known human chromosomal disorder. The overall incidence is

Box 3.1: Chromosomal disorders.

- Chromosome numerical alteration 47XY + 21
- Deletion—single gene, microdeletion
- Duplication
- Inversion
- Unbalanced translocation
- Ring chromosome
- Uniparental disomy

Box 3.2: Causes of birth defects.

- Heredity
- Intrauterine infections (TORCH)
- Maternal diseases—diabetes mellitus, hypertension
- Maternal drug injection phenytoin
- Smoking and alcohol injection
- Advanced paternal and maternal age
- Environmental toxin
- Maternal exposure to harmful radiation.

[TORCH: toxoplasmosis, other (syphilis, varicella-zoster, parvovirus B19), rubella, cytomegalovirus, and herpes]

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approximately 1 in 1,000 live births, but markedly varies with increasing maternal age (Table 3.1).

Causes

Majority (90%) of Down babies occur due to non-disjunction during maternal oogenesis. Two percent occur as a result of Robertsonian translocation, while another 2% is due to mosaicsm.

Clinical Features (Figs 3.7A to C)

Facial Features: Small, lowset ears, upward slanting eyes, prominent epicanthic folds, brush fields spots apparent in eyelids (white spot in the iris), flat facial profile, small oral cavity with protruding tongue, short nose with depressed nasal bridge, and brachycephaly.

Hypotonia, single palmar crease, fifth finger clinodactyly.

Cardiac: 40–50% have congenital heart disease. Endocardial cushion defect, ventricular septal defects (VSD), atrial septal defect (ASD), Fallot's tetralogy, and isolated posterior descending artery (PDA).

Gastrointestinal tract: Duodenal atresia, Hirschsprung's disease, trachea esophageal fistula, and imperforated anus.

Table 3.1: Likely incidence of Down's syndrome with increasing age.

Maternal age	*Incidence*
20	1 in 1,530
30	1 in 900
35	1 in 385
37	1 in 225
40	1 in 109
44	1 in 41

Eye

- Congenital cataract
- Glaucoma.

Genitourinary system

- Hypospadias
- Cryptorchidism

Hematological

- Iron-deficiency anemia
- Leukemia

Endocrine

- Acquired hypothyroidism

Deafness

- Both sensorineural and conductive

Orthopedic

- Developmental hip dysplasia
- Atlantoaxial dislocation

Increased risk of infection.

Diagnostic Investigation

A clinical diagnosis requires cytogenetic studies, karyotyping of the infant. In translocated Down's syndrome, parents also need karyotyping.

Fluorescent in situ hybridization test or quantitative fluorescence PCR (QFPCR) may provide rapid diagnosis.

Prenatal Screening

Screening is offered to women of all ages:

- A nuchal translucency scan is performed at 11–13 weeks of pregnancy
- Maternal serum markers: Serum alpha fetoprotein, unconjugated estriol, and total human chorionic gonadotropin (hCG) can be measured between 10 weeks and 14 weeks of gestation.



Figs 3.7A to C: Down's syndrome: (A) Upslanting eyes—mongoloid slant; (B) Brachycephaly—flat occiput; (C) Simian crease and clinodactyly of fifth finger.

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Prenatal Diagnostic Tests

Diagnostic test is indicated to screen. The initial screening tests include:

- Chorionic villus sampling
- Amniocentesis is ideally performed after 15 weeks of gestation followed by QFPCR or full karyotyping.

Management

- Genetic counseling is essential to prevent recurrences. Recurrence risk for trisomy 21 is 1% and de novo translocation is 2–3%. The risk is higher (10–13% for carrier mother and 2–3% in carrier father) for a Robertsonian carrier parent.
- Cardiac assessment, hip ultrasound, and audiology are minimum base-line investigations.
- *Early stimulation:* Early stimulation is suggested for all the children suffering from Down syndrome and should be started as soon as possible. Preferably, a child psychologist might be recommended for the stimulation process. Parents should be actively involved during the stimulation process. The primary intention of the early stimulation process includes:
  - Individualizing the training program
  - Imparting basic information and modifying the unfavorable attitude of parents toward children suffering with Down syndrome
  - Implicating parents for efficiently teaching the appropriate skills in diverse areas of development.
- *Investigation*:
  - Cardiac echo
  - Thyroid function test
  - Immunoglobulin level
  - Ophthalmological examination
  - Hearing test.

Central nervous system: Moderate to severe learning difficulty is characteristic of Down syndrome.

Psychiatric Disorders

Autism, attention deficit hyperactivity disorder (ADHD), obsessive compulsive disorder, and Alzheimer-like dementia may develop early. Many children need speech and language therapy.

Eye

Strabismus, blepharitis, cataract, need correction. Annual ophthalmological and audiological evaluation is necessary.

Endocrine

Short stature, obesity, hypothyroidism, diabetes, decreased fertility.

Skeletal

Atlantoaxial instability can lead to spinal cord compression. Hence, cervical radiography is essential to detect atlantoaxial instability every 3 years.

Blood

Acute leukemias (AML and ALL) are common.

ENT

- Obstructive sleep apnea
- Chronic otitis media
- Hearing loss.

Long-term follow-up should ideally be by a multidisciplinary team led by a pediatrician with special expertise, such as a developmental pediatrician working in a child developmental center. Physiotherapy to improve tone and posture is often required.

Prognosis: Outlook is much improved with better integrity and longevity. Majority can live semi-independently with supervision. Congenital heart disease is the main cause of early mortality.

Alzheimer's like dementia may occur in Down's syndrome.

Edwards Syndrome (Trisomy 18)



Meiotic non-disjunction contributes to 95% of Edwards syndrome. 5% occur due to translocation or mosaics.

Clinical Features
(Figs 3.8A to D and Box 3.3)

- Intrauterine growth retardation
- Dysmorphic and characteristic features
  - Micrognathia
  - Narrow palpebral fissures
  - Prominent occiput
  - Ptosis
  - Hyper-flexed wrist
  - Ocular hypertelorism

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Figs 3.8A to D: Edward syndrome:. (A) Short stature—Turner syndrome; (B) Webbed neck—Turner syndrome; (C) Overlapping of fingers; (D) Baby with cleft lip palate.

Box 3.3: Prevention of birth defects

1. Avoid consanguineous marriages
2. Periconceptional folic acid
3. Appropriate antenatal care
4. Avoid exposure to radiation
5. Maternal ingestion of drugs to be monitored
6. Healthy eating and adequate exercise during pregnancy.

"Take care of your genes and they will take care of you."

  - Microcephaly
  - Cleft lip/palate.

Low set or malformed ear

- Skeletal abnormalities
  - Rocker bottom feet
  - Over-riding fingers
  - Clenched fist
  - Hemivertebrae
- Cardiac and renal anomalies.

Diagnosis: Prenatal diagnosis is by triple test and targeted ultrasonography.

Prognosis

- 50% die in utero
- For live-born children, 50% live up to 2 months.

Turner's Syndrome (Fig. 3.9)

Incidence: 1 in 2,500 live birth.
Karyotype: 45,XO.

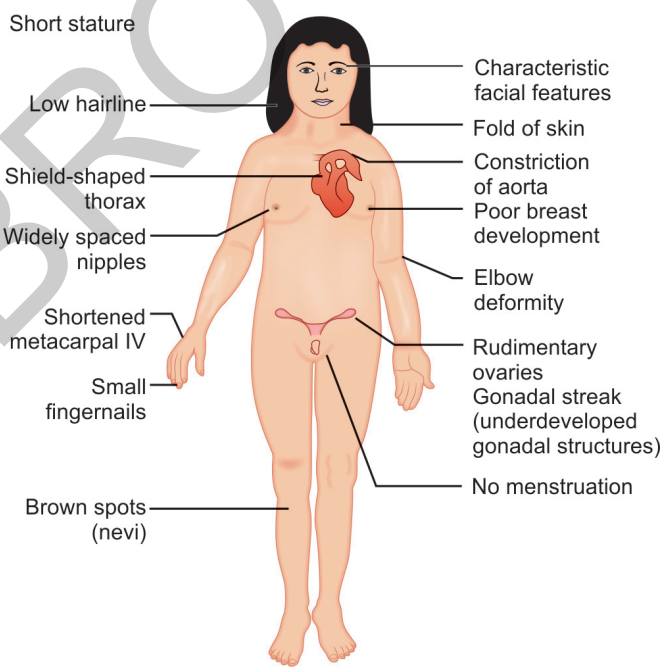

Fig. 3.9: Turner's syndrome.

Etiology

In 80% of cases, the paternal X chromosome is missing. Other variants of karyotypes include mosaics: 46,XX/45,X or 46,XY/45,X.

Clinical Features

Newborn: Phenotypic features of Turner's syndrome are:

- Webbed neck
- Low posterior hairline

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- Shield chest
- Widespaced nipples
- Coarctation of aorta
- Loose folds of skin particularly in neck
- Lymphedema (Bonnevie-Ullrich syndrome).

Childhood

- Short stature
- Delayed puberty.

Skeletal

Cubitus valgus, short fourth metatarsal or metacarpal, shield chest and hip dislocation.

Cardiovascular

Coarctation of aorta and bicuspid aortic valve.

Eyes and Ear

Ptosis, strabismus, amblyopia, and cataract.

MANAGEMENT OF GENETIC DISEASES

Genotype–phenotype variation leads to a wide variety of conditions in the affected children. Mild defects can be treated appropriately whereas the severely affected children need prolonged care. Multisystem involvement requires the services of a multidisciplinary team: Pediatrician, geneticist, cardiologist, neurologist, pediatric surgeon, physiotherapist and psychologist. Parents need counseling. Drugs, enzyme replacement therapy, surgery, physiotherapy, and counseling play a role in rehabilitating these children in the society. Family screening is vital to help other members to deal with hereditary problems. Periodic screening and surveillance is recommended if cancer is anticipated.

Key Points

1. Genes control all bodily functions and are responsible for the way we look and behave.
2. Loss or gain of chromosomes result in disease.
3. Environmental factors also play an important role in disturbing the developing fetus.
4. If one organ system is affected, involvement of other systems must be probed.
5. Karyotyping is performed first in children presenting with ambiguous genitalia and short stature.

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