

Cellular Biology, Genetics & DNA



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CELL

The unit from which living organisms and tissues are built. At some stage in its development is capable of reproduction by mitosis. Composed of a nucleus, surrounded by cytoplasm and bounded by a cell membrane.

Cells are classified according to:

Function (secretory, nerve, germinal)

Shape (columnar, squamous)

Arrangement (single layer, stratified)

Contents (fat, pigment)

Special features (ciliated)

Each cell of a higher organism is composed of a jellylike layer of material, the cytoplasm, which contains many small structures called organelles. This cytoplasmic material surrounds a prominent body called the nucleus. Every nucleus contains a number of minute, threadlike chromosomes.

TISSUE

A group of associated similarly structured cells that perform specialised functions for the survival of the organism. Tissues form the basic anatomical and physiological components of a living organism, consisting of collections of cells, with one type predominating, e.g. muscle tissue, nerve tissue, bone tissue, scar tissue, connective tissue

ORGAN

Any differentiated part devoted to a specific function, e.g. heart, lung, bowel, liver, kidney, genitals, sense organs

VISCUS

(pl. viscera)

Internal organs, which lie within or closely, associated with a body cavity, e.g heart, lung, liver, spleen, kidney

Cell Structure and Function

Cells are composed primarily of oxygen, hydrogen, carbon, and nitrogen, the elements that make up the majority of **organic** compounds. The most important organic compounds in a cell are proteins, nucleic acids, lipids, and polysaccharides (carbohydrates). The "solid" structures of the cell are complex combinations of these large molecules. Water makes up 60 to 65 percent of the cell, because water is a favourable environment for biochemical reactions.

All cells are dynamic at some stage of their life cycle, in the sense that they use energy to perform a variety of cell functions: movement, growth, maintenance and repair of cell structure, reproduction of the cell, and manufacture of specialised cell products such as enzymes and hormones. These functions are also the result of interactions of organic molecules.

Division, Reproduction, and Differentiation

All cells are the products of the division of pre-existing cells. Simple cell division, or asexual reproduction, results in the production of two identical daughter cells, each containing a set of chromosomes identical with those of the parent cell. This process of cell division by which a new cell comes to have an identical number of chromosomes as the parent cell is called **mitosis**. Before the onset of division, a cell grows to roughly twice its original size. In doing so it duplicates its DNA, so that each chromosome is doubled. During division the duplicate sets are physically separated, following longitudinal splitting of each double chromosome, and are transported into opposite sides of the cell. The cell then constricts around its equator and pinches in two. This is the process by which complex organisms grow and replace worn-out tissue.

Sexual Reproduction

Sexual Reproduction is the mingling of the DNA of two different organisms of the same species to produce a cell, or cells, with a new combination of genes. Sexual reproduction requires the production of male and female germ cells or gametes (sperm and eggs respectively) by a process called **meiosis**. Meiosis in the male testes produces gametes called spermatozoa. Meiosis in the female ovaries produces gametes called ova (eggs). During this process a cell divides twice; but its chromosomes are duplicated only once. Thus, four germ cells are produced, each containing half the normal number of chromosomes (23 instead of 46). Meiosis differs from mitosis in one important way: in meiosis a single chromosome from each pair of chromosomes is transmitted from the original cell to each of the new cells. Thus, each gamete contains half the number of chromosomes that are found in the other body cells. A spermatozoa and an ovum unite during **fertilisation** to form a new cell, called a zygote, that has a complete set of chromosomes. The zygote has received half its genetic information from each parent, thus making it a new individual.

Chromosomes are tiny threadlike structure, composed of nucleic acids and proteins (chromatin), found in all plant and animal cells. Chromosomes vary in size and shape and usually occur in pairs. The members of each pair, called homologues, closely resemble each other physically. Most cells in the human body contain 23 pairs of chromosomes. Every chromosome in a cell contains many **genes**, and each gene is located at a particular site, or **locus**, on the chromosome.

Cromosomes

The chromosome contains a very long single strand of the nucleic acid **DNA**, which is divided into small units called **genes**. The genes determine the hereditary characteristics, such as colour and size, of the cell or organism. There are 2 alternative forms (**alleles**) for each gene, one occurring on each of the pair of homologous chromosomes. Normally the body cells of each species contain a specific number of chromosomes, which occur in pairs. Humans have 23 pairs of chromosomes.

Nucleic Acids are extremely complex molecules produced by living cells and viruses. Their name comes from their initial isolation from the nuclei of living cells. Nucleic acids have at least two functions: to pass on hereditary characteristics from one generation to the next (deoxyribonucleic acids or DNA), and to trigger the manufacture of specific proteins (ribonucleic acids or RNA). The backbones of both DNA and RNA molecules are shaped like helical strands. Their molecular weights are in the millions. The sugar in the backbone of RNA is ribose, and in DNA deoxyribose. In short, the structure of a DNA molecule or combination of DNA molecules determines the shape, form, and function of the offspring.

Gene Action: DNA and the Code of Life

DNA is made up of substances called **nucleotides**. Each nucleotide consists of a phosphate, a sugar known as deoxyribose, and any one of four nitrogen-containing bases. The four nitrogen bases in DNA are adenine (A), guanine (G), cytosine (C) and thymine (T). In RNA thymine is replaced by uracil (U).

In 1953, geneticists James Dewey Watson and Francis Harry Compton Crick worked out the structure of DNA. Watson and Crick found that the DNA molecule is composed of two long strands in the form of a Double Helix, somewhat resembling a long, spiral ladder. The strands, or sides of the ladder, are made up of alternating phosphate and sugar molecules. The nitrogen bases, joining in pairs, act as the rungs. Each base is attached to a sugar molecule and is linked by a hydrogen bond to a complementary base on the opposite strand. Adenine always binds to thymine, and guanine always binds to cytosine. To make a new, identical copy of the DNA molecule, the two strands need only unwind and separate at the bases (which are weakly bound); with more nucleotides available in the cell, new complementary bases can link with each separated strand, and two double helixes result. If the sequence of bases were AGATC on one existing strand, the new strand would contain the complementary, or "mirror image," sequence TCTAG. Since the "backbone" of every chromosome is a single long, double-stranded molecule of DNA, the production of two identical double helixes will result in the production of two identical chromosomes. This occurs during mitosis.

The DNA backbone is actually a great deal longer than the chromosome but is tightly coiled up within it. DNA has a "coiled-coil" configuration, like the filament of an electric light bulb.

Proteins are not only the major components of most cell structures, they also control virtually all the chemical reactions that occur in living matter. The ability of a protein to act as part of a structure, or as an enzyme affecting the rate of a particular chemical reaction, depends on its molecular shape. This shape, in turn, depends on its composition. Every protein is made up of one or more components called polypeptides, and each polypeptide is a chain of subunits called amino acids. Twenty different amino acids are commonly found in polypeptides. The number, type, and order of amino acids in a chain, which ultimately determine the structure and function of the protein, are encoded in the sequence of bases in the DNA molecule.

The Genetic Code

Proteins are the products of genes, and each gene is composed of sections of DNA strands. The sequence of the four nucleotide bases in the DNA directs the sequence of amino acids in the formation of polypeptides during protein synthesis. The Genetic Code is made up of combinations of three successive nucleotides. The sequence of the base triplets (or codons) within the gene thus defines the order of the amino acids in the polypeptide chain.

The DNA segment C-G-T-C-T-A-A-A-A-C-G-T

Translates into the triplet code [C-G-T] - [C-T-A] - [A-A-A] - [C-G-T]

Which corresponds to the amino acid sequence [Alanine] - [Aspartate] - [phenylalanine] - [Alanine]

Which in turn determines the type of structural protein or the enzyme synthesised by the cell.

BOOK ANALOGY: The Meaning of Life

All cells have the same 23 books (chromosomes) on their bookshelf (nucleus)

Each book is in two volumes (paired chromosome, one inherited from each parent)

Each book is composed of numerous chapters (genes)

Each chapter is composed of a long sequence of words (triplet base sequences or codons)

Each word is composed of any of 26 letters (codons comprise 3 of the 4 bases).

Although all the cells in the body cells have access to the full reference collection of 23 x 2 books (46 chromosomes) different cells will only bother to 'read' the relevant chapters in the relevant volumes, according to their own specific structure and function. E.g. skin cells would concentrate on chapters relating to the synthesis of keratin, to produce hair and nails, red blood cells on the synthesis of haemoglobin, white blood cells on the synthesis of antibodies, gut lining cells on the synthesis of digestive enzymes, pancreatic cells on the synthesis of insulin.

Some sequences of nucleotides are repeated many times throughout the genetic material; these are called **Variable Number of Tandem Repeats (VNTR)**. VNTR repeat units comprise 150 - 300 bases repeated many times creating sequences measuring 1 000 - 10 000 bases.

Since all human cells require to produce the same human proteins and enzymes the DNA sequences are almost identical in all individuals. Only about 10% of the DNA molecule is used for genetic coding. Between these coding genes are long repetitive non-coding segments, which show marked individual variations and are unique to that person. The chances of two unrelated individuals sharing the same sequence is estimated at one in a million billion and even amongst siblings only one in ten thousand million.

Codon (individual amino acid) comprises 3 bases

Restriction enzyme site comprises 4-6 base pairs (bp)

Typical VNTR repeated sequence comprises 15-75 bp

Typical non variable repeat comprises 150-300 bp

Typical VNTR locus comprises 1-10 kilobases (kb)

Typical gene comprises 20-100 (kb)

Average human chromosome comprises 120 million bp

The total human genome comprises 3 billion base pairs (bp)

Knowing how protein is made allows scientists to understand how genes can produce specific effects on the structures and functions of organisms. This does not explain, however, how organisms can change in response to changing environmental circumstances, or how a single-celled zygote can give rise to all the different tissues and organs that make up a human being. Most of the cells in these tissues and organs contain identical sets of genes but nevertheless

make different proteins; clearly, in the cells of any one tissue or organ some genes are acting but others are not. Different tissues have different arrays of genes in the active state. Thus, part of the explanation for the development of a complex organism must lie in the ways by which genes are specifically activated. The processes of gene activation in higher organisms are still obscure.

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Human Heredity

Genetics is the scientific study of how physical, biochemical, and behavioural traits are transmitted from parents to their offspring. Geneticists are able to determine the mechanisms of inheritance because the offspring of sexually reproducing organisms do not exactly resemble their parents, and because some of the differences and similarities between parents and offspring recur from generation to generation in repeated patterns. Most physical characteristics of humans are influenced by multiple genetic variables as well as by the environment. Some characteristics, such as height, have a relatively large genetic component. Others, such as body weight, have a relatively large environmental component. Still other characteristics, such as the blood groups and the antigens involved in the rejection of transplanted organs, appear to involve entirely genetic components; no environmental condition is known to change these characteristics.

Susceptibility to various diseases has an important genetic element. These diseases include schizophrenia, tuberculosis, malaria, several forms of cancer, migraine headaches, and high blood pressure. Many rare diseases are caused by recessive genes and a few by dominant genes. The human genome contains approximately 50,000 to 100,000 genes, of which about 4000 may be associated with disease.

The Transmission of Genes

The union of gametes brings together two sets of genes, one set from each parent. Each gene—that is, each specific site on a chromosome that affects a particular trait—is therefore represented by two copies or alleles, one coming from the mother and one from the father. Each copy is located at the same position on each of the paired chromosomes of the zygote. When the two alleles are identical, the individual is said to be **homozygous** for that particular gene. When they are different—that is, when each parent has contributed a different allele of the gene—the individual is said to be **heterozygous** for that gene. Both alleles are carried in the genetic material of the individual, but if one is dominant, only that one will be manifested.

For example, the ability of a person to form pigment in the skin, hair, and eyes depends on the presence of a particular allele (A), whereas the lack of this ability, known as albinism, is caused by another allele (a) of the same gene. For convenience, alleles are usually designated by a single letter; the dominant allele is represented by a capital letter and the recessive allele by a small letter. The effects of the A allele are dominant over a, the recessive allele. Therefore, heterozygous persons (Aa), as well as persons homozygous (AA) for the pigment-producing allele, have normal pigmentation. Persons homozygous for the allele that results in a lack of pigment (aa) are albinos.

Each child of a couple who are both heterozygous (Aa) has a probability of one in four of being homozygous AA, one in two of being heterozygous Aa, and one in four of being homozygous aa. Only the individuals carrying aa will be albinos. Note that each child has a one-in-four chance of being affected with albinism; it is not accurate to say that one-quarter of the children in a family will be affected. Both alleles will be carried in the genetic material of heterozygous

offspring, who will produce gametes bearing one or the other allele.

Table 1: Autosomal Recessive Inheritance (Albinism)

		Normal heterozygous parent (Aa)	
		A	a
Normal heterozygous parent (Aa)	A	AA normal	Aa unaffected heterozygote
	a	Aa unaffected heterozygote	aa affected

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Other autosomal recessive conditions: sickle cell anaemia, phenylketonuria, thalassaemia

Table 2: Autosomal Dominant Inheritance (Huntingdon's chorea)

		Affected parent (Hh)	
		H	h
Normal parent (hh)	h	Hh affected	hh normal
	h	Hh affected	hh normal

Other autosomal dominant conditions: achondroplasia, spherocytosis, Marfan's syndrome

A distinction is made between the appearance or outward characteristics, of an organism and the genes and alleles it carries. The observable traits constitute the organism's **phenotype**, and the genetic makeup is known as its **genotype**.

Sex and Sex Linkage

Sex is usually determined by the action of a single pair of chromosomes. Abnormalities of the endocrine system or other disturbances may alter the expression of secondary sexual characteristics, but they almost never completely reverse the genetically determined sex. 22

pairs of chromosomes are alike in both males and females; these are called **autosomes**.

The remaining pair of chromosomes are called the **sex chromosomes**. Females have two X chromosomes, males have one X and one Y chromosome. When gametes are formed, each egg produced by the female contains one X chromosome, but the sperm produced by the male can contain either an X or an Y chromosome. The union of an egg, which always bears an X chromosome, with a sperm also bearing an X chromosome produces a zygote with two X's: a female offspring. The union of an egg with a sperm that bears a Y chromosome produces a male offspring.

Table 3: Determination of sex

		Male (XY)	
		X	Y
Female (XX)	X	XX daughter	XY son
	X	XX daughter	XY son

The human Y chromosome is approximately one-third as long as the X, and apart from its role in determining maleness, it appears to be genetically inactive. Thus, most genes on the X have no counterpart on the Y. These genes, said to be sex-linked, have a characteristic pattern of inheritance. Haemophilia, for example, is usually caused by a sex-linked recessive gene (h). A female with HH or Hh is normal; a female with hh has haemophilia. A male is never heterozygous for the gene because he inherits only the gene that is on the X chromosome. A male with H is normal; with h he has haemophilia. When a normal man (H) and a woman who is heterozygous (Hh) have offspring, the female children are normal, but half of them carry the h gene—that is, none of them is hh, but half of them bear the genotype Hh. The male children inherit only the H or the h; therefore, half the male children have haemophilia.

Table 4: Haemophilia

		Normal male (XY)	
		X	Y

Carrier Female (X^hX)	X^h	X^hX carrier daughter	X^hY affected son
	X	XX normal daughter	XY normal son

Table 5: Haemophilia

		Affected male (X^hY)	
		X^h	Y
Normal Female (XX)	X	X^hX Carrier daughter	XY normal son
	X	X^hX Carrier daughter	XY normal son

Thus, in normal circumstances a female carrier passes on the disease to half her sons, and she also passes on the recessive h gene to half her daughters, who in turn become carriers of haemophilia.

Other sex-linked conditions: red-green colour blindness, hereditary nearsightedness, night blindness, and ichthyosis (a skin disease).

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IDENTIFICATION BY DNA PROFILING

The DNA present in hair root bulbs, spermatozoa or any nucleated tissue can be profiled. It is noteworthy that red blood cells lose their nuclei early in their life span but white blood cells remain nucleated. A few hair roots on a blunt instrument found in the possession of a crime suspect can be matched against an autopsy blood sample. Similarly seminal fluid in the vagina of a rape victim can be compared with DNA in the hair or white blood cells of a suspect. Since every cell within an individual contains the same DNA there is no need to match DNA from semen with semen or from blood with blood. Samples commonly examined for DNA in forensic cases include liquid blood, blood stains, semen, saliva, cellular tissue, hair root bulbs and mouth

swabs. In Dundee all prisoners charged with a serious arrestable offence have cells scraped from the cheek lining; these are profiled and entered into a DNA database.

The traditional method of DNA analysis involved producing an autoradiograph, which has the appearance of the familiar supermarket bar code. The technique of determining the sequences is extremely complex and involves cutting the DNA at predetermined points using biochemical scissors known as restriction enzymes.

The DNA fragments so produced are loaded into wells at one end of a gel. They are then separated by electrophoresis, which involves applying an electrical current to a gel. The fragments race along the gel according to their size and charge; the smallest fragments, less constrained by the gel, move fastest and furthest from the origin and the larger fragments will move more slowly. Fragments are then fixed into position to which they have moved by blotting onto a nylon membrane (Southern blotting). The position to which each fragment has moved can be visualised by use of a probe, a synthetically produced piece of DNA whose chemical sequence structure recognises specific repeated chemical sequences within the fragments to be identified. The probe is labelled with a radioactive isotope or chemical marker. The marker probe binds or hybridises to the fragments of interest. The position of the probes and hence the fragments of interest is identified as a series of black bands where the radioactive isotope have exposed the X-ray photographic plate. This is called an auto-radiograph.

Single locus probes detect a single segment of DNA located on a single chromosome. This results in a DNA pattern containing two bands, one for each DNA segment recognised on each member of a chromosome pair, one being inherited from each parent. Running samples from father, mother and child alongside each other will rapidly visualise whether the child's bands could have been inherited from the father or whether the father is unrelated. By selecting probes with different chemical structures it is possible to look at different regions along the threads of DNA. A series of probes is generally used, each identifying fragments from a different region. If the same sized fragment is inherited from both parents only one band will be seen where the two fragments of the same size overlap. The patterns are thus visualised as a one or two band bar-code

Multi-locus probes detect multiple DNA segments located on many chromosomes and results in numerous bands. With CellmarkTM diagnostic technology 45-60 different segments are recognised giving the appearance of a supermarket bar-code. This can provide many points of comparison between individuals and are highly specific giving rise to a '**DNA fingerprint**'. A DNA fingerprint can positively show whether or not a man is the biological father of a child since the odds that two unrelated people share the same DNA fingerprint has calculated to be on average thirty billion to one. The earth's population is only five billion and only half of these are males. With such impressive statistics it is easy to become overawed by the significance of DNA evidence. However some caution is required since laboratory techniques must be meticulous to prevent contamination of the samples and to ensure the authenticity of sample materials. There has been considerable legal argument about the strength of DNA evidence in recent years (Journal of the Forensic Science Society 1993; 33(4): p204-241).

POLYMERASE CHAIN REACTION (PCR)

Using this technique it is possible to copy small fragments of DNA in a test-tube resulting in a many million fold increase in the DNA available for testing. Polymerase enzyme unzips the double helix of DNA and builds new complimentary strands by adding complimentary bases and backbone units opposite each exposed strand of DNA. During successive generations huge numbers of chemical copies of the original DNA are produced. This technique can be carried out even on very small, degraded or old samples. Enough DNA can be propagated from a licked envelope or stamp or from a single hair root.

The DNA to be duplicated or amplified is denatured by heating to 94°C. This separates the double stranded DNA into 2 single strands. **Primers** are added to mark the beginning and end of the base sequence to be duplicated. The primer consists of a short sequence of DNA which binds or hybridises to the complimentary sequence at the desired points on the DNA chain. This effectively lays the first and last bricks and gives the Polymerase the starting point to commence duplication of DNA. PCR allows selective replication of the specific portions of DNA which contain the regions of greatest variability. In the second stage, Polymerase enzyme and nucleotide units of A, T, C & G are added to commence rebuilding the complimentary base sequence one by one. In this way the desired portion of each strand of DNA is duplicated within 2 minutes. Repeating this process 25-30 times produces a million copies of the DNA present in the initial sample. Chemically labelled complimentary base sequences or **probes** are added to mark the position of the fragments. The chemical colour changes are then detected by computerised laser scanning and converted into a numerical code which can be rapidly compared between suspect and evidential samples. In many laboratories these high throughput automated techniques have largely superseded the older, more cumbersome techniques of Southern blotting and production of auto-radiographs.

Some newer techniques are aimed at detection of shorter but highly variable portions of the DNA chain, known as **Short Tandem Repeats (STR)** systems. The repetitive segments detected are short and exhibit less variability in fragment length. There are also fewer possible lengths of fragment, typically 6-12, compared to more than 100 with SLP & MLP systems. This means that STR has lower discriminating power and larger numbers of STR test systems must be used to achieve similar levels of discrimination. However, the smaller number of fragments separated can be sized with considerable accuracy and their categorisation allows results to be expressed as simple numerical values or types. These numerical DNA types are readily compared between individuals and easily compiled for storage on a database.

DNA fingerprinting was first developed as an identification technique in 1985. Originally used to detect the presence of genetic diseases, DNA fingerprinting soon came to be used in criminal investigations and forensic medicine. The first criminal conviction based on DNA evidence in the United States occurred in 1988. In criminal investigations, DNA fingerprints derived from evidence collected at the crime scene are compared to the DNA fingerprints of suspects. The DNA evidence can implicate or exonerate a suspect.

Generally, courts have accepted the reliability of DNA testing and admitted DNA test results into evidence. However, DNA fingerprinting is controversial in a number of areas: the accuracy of the results, the cost of testing, and the possible misuse of the technique.

The accuracy of DNA fingerprinting has been challenged for several reasons. First, because DNA segments rather than complete DNA strands are "fingerprinted," a DNA fingerprint may not be unique; large-scale research to confirm the uniqueness of DNA fingerprinting test results has not been conducted. In addition, DNA fingerprinting is often done in private laboratories that may not follow uniform testing standards and quality controls. Also, since human beings must interpret the test, human error could lead to false results. DNA fingerprinting is expensive. Suspects who are unable to provide their own DNA experts may not be able to adequately defend themselves against charges based on DNA evidence. Widespread use of DNA testing for identification purposes may lead to the establishment of a DNA fingerprint database. This database could potentially be used for unauthorized purposes, such as identifying individuals with stigmatizing illnesses such as Acquired Immune Deficiency Syndrome (AIDS).

The addresses of several useful websites on the subject of DNA can be found on our [Links Page](#)

BLOOD TYPES

Blood typing is a classification of red blood cells by the presence of specific surface protein substances (antigens) on their surface. Typing of red blood cells is a prerequisite for blood transfusion. In the early part of the 20th century, physicians discovered that blood transfusions often failed because the blood type of the recipient was not compatible with that of the donor. In 1901 the Austrian pathologist Karl Landsteiner classified blood types and discovered that they were inherited from the parents.

Blood grouping, using the word in its widest sense, can be used:

1. To show whether blood stains on weapons, clothing or elsewhere could have come from a particular suspect or victim.
2. In matching fragmented human remains.
3. To help resolve parentage and inheritance disputes.

In the last few years the use of DNA profiling has overtaken serological blood testing. However its cost and unavailability in certain areas means that serology remains an important method of forensic analysis. The major blood group system is called the ABO system and results from the expression of A and/or B antigens on the red blood cells and the presence of anti-A and/or anti-B antibodies in the serum. Antibodies to the ABO groups are available commercially for use in laboratory testing of unknown blood samples.

80% of people secrete water soluble blood group antigens in their sweat, saliva, semen, gastric or other body fluids. Such secretors may be identified from seminal stains and even the saliva deposited on a bite.

The four main blood types are known as A, B, AB, and O. Blood type A contains red blood cells that have a substance A on their surface. This type of blood also contains an antibody directed against substance B, found on the red cells of persons with blood type B. Type B blood contains the reverse combination. Serum of blood type AB contains neither antibody, but red cells in this type of blood contain both A and B substances. In type O blood, neither substance is present on the red cells, but the individual is capable of forming antibodies directed against red cells containing substance A or B. If blood type A is transfused into a person with B type blood, anti-A antibodies in the recipient will destroy the transfused A red cells. Because O type blood has neither substance on its red cells, it can be given successfully to almost any person (group O individuals are universal donors). Persons with blood type AB have no antibodies and can receive any of the four types of blood (group AB individuals are universal recipients).

Table 6:

<i>Phenotype</i>	<i>Possible genotypes</i>	<i>Antigen on blood cells</i>	<i>Antibody in serum</i>	<i>Compatible Donor groups</i>	<i>Incompatible Donor</i>
A	AA, AO	A	Anti-B	A, O	B, AB
B	BB, BO	B	Anti-A	B, O	A, AB
AB	AB	both A & B	neither antibody	A, B, AB, O	none

O	O	neither A nor B	Anti-A & Anti-B	O	AB, A, B
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Table 7:

<i>Phenotype</i>	<i>UK</i>
A	42%*
B	8%
AB	3%
O	47%

*43% in Southern England, 34% in Scotland

Other hereditary blood-group systems have subsequently been discovered. The hereditary blood constituent called Rh factor is of great importance in obstetrics and blood transfusions because it creates reactions that can threaten the life of new-born infants. Blood types M and N have importance in legal cases involving proof of paternity.

PRINCIPLES OF BLOOD GROUP INHERITANCE

1. Blood group genes (and the antigens for which they code) are passed on from parent to offspring.
2. One gene of an allelic pair is derived from each parent who has one of two genes to donate at random.
3. A blood group antigen cannot be present in the child unless it is also present in at least one parent.
4. If one parent possesses a homozygous allelic pair (e.g. AA or BB) then that gene must be present in all of his or her offspring.

Blood group remains constant throughout life except for transient disturbances after blood transfusion. The pair of genes responsible for the expression of blood group antigens are inherited, one from each parent.

Table 8:

<i>Parent 1</i>	<i>Parent 2</i>	<i>Possible offspring</i>	<i>Impossible offspring</i>
O	O	O	A, B, AB

O	A (AA, AO)	O, AO	B, AB
O	B (BB, BO)	O, BO	A, AB
A (AA, AO)	A (AA, AO)	O, AA, AO	B, AB
A (AA, AO)	B (BB, BO)	O, AA, AO, BB, BO, AB	None impossible
B (BB, BO)	B (BB, BO)	O, BB, BO	A, AB
O	AB	AO, BO	O, AB
A (AA, AO)	AB	AA, AO, BO, AB	O
B (BB, BO)	AB	BB, BO, AO, AB	O
AB	AB	AA, BB, AB	O

Red Cell Antigen systems which can be typed: MNS, Rh, ABO, Duffy, Kidd, Kell, Luthran,
 Serum Proteins systems which can be typed: Haptoglobins (Hp), Gc, C3, Gm,
 Red Cell Enzymes systems which can be typed: Erythrocyte acid phosphatase, Glutamate
 pyruvate transaminase, Glyoxylase, Phosphoglucomutase, Esterase D, Adenylate kinase,
 Adenosine deaminase. 6-Phosphogluconate dehydrogenase 2.5%

The total chance of exclusion using all the above systems combined is 93%. Thus, the more systems tested the more likely a wrongly accused father is to be positively excluded from paternity. Such blood grouping can exclude a man being the father but can never totally confirm his paternity.

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DISPUTED PARENTAGE

In practice this means disputed paternity since maternity is rarely in doubt. Disputes may arise under the following circumstances:

1. A married man may allege that his wife has committed adultery, and that he is not the father of one or more of her children.
2. A women who has illegitimate offspring may allege that a certain man is the father of that child in order to obtain an affiliation order against him and to ensure financial assistance with the child's upbringing.

In either event the court may seek medical assistance in determining the paternity of the child under the Family Law Reform Act 1969, which applies to England and Wales but not Scotland, a court may direct that blood tests are used, upon application by one of the parties to the

proceedings. Submission to blood testing is usually agreed between the parties and court orders are seldom required. Consent is required, the age of consent being 16 but the court may draw inferences from the fact that a person has failed to comply with blood testing. However since serological blood testing can only exclude parentage, the man has little to lose and perhaps everything to gain from submitting to blood testing; the mother of the child has by contrast comparatively little expectation of advantage. Use of DNA profiling for such purposes is increasing since it more positively discriminating than exclusion by serology. The practicalities of blood testing involve adequate identification of the parties involved by their legal representatives and by means of photographs. Blood is taken from the mother, the child and the putative father or fathers who must be identified to the doctor taking the samples either by them all attending together or being separately identified by solicitors acting in the case. 5 ml of venous blood is taken from adults and 1 ml by heel or ear prick from infants who should be at least 6 months old. No party should have had a blood transfusion within 3 months of sampling.

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