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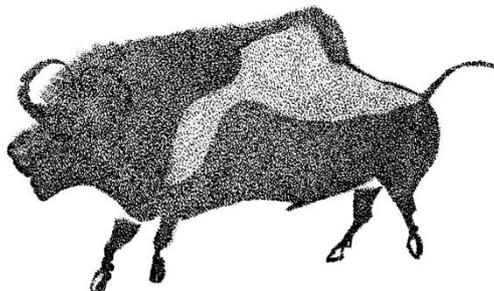
# **Evolution of Human Structure**

## **ANAT2521**

**Course Outline**

**Summer session 2016**

**January 4 to 28**



**Course Authority:**  
**Professor Ken Ashwell**

CRICOS Provider Code 00098G

Please read this outline in conjunction with the following pages on the [Medical Sciences website](#):

- [Advice for Students](#)
- [Learning Resources](#)

(or see "STUDENTS" tab at [medicallsciences.med.unsw.edu.au](http://medicallsciences.med.unsw.edu.au) )

Course authority: Prof. Ken Ashwell ([k.ashwell@unsw.edu.au](mailto:k.ashwell@unsw.edu.au), 9385 2482),  
Department of Anatomy, School of Medical Science, Room 447, Wallace Wurth

Lectures: **Wallace Wurth LG02**

Practical Classes: **Wallace Wurth 101 (Gross Anatomy Laboratory)**

Tutorials: venues to be advised.

## IMPORTANT NOTES

- Students must wear enclosed shoes (i.e. no thongs or sandals) in the Dissecting Room.
- No eating, drinking or smoking in the Dissecting Room.
- Mobile phones must be switched off during lectures and classes.

## COURSE AIMS

The aims of this course are to:

1. Provide the student with an understanding of the major biological (physical and evolutionary) attributes of non-human primates and humans.
2. Assist the student to develop a deeper appreciation of the place of humans in the natural world and their relationship to other primates.
3. Provide the student with some knowledge and skills from the field of biological anthropology.
4. Help the student to appreciate the importance and relevance of the study of human origins for an understanding of modern human structure, development and disease.

## STUDENT LEARNING OUTCOMES

Students should complete the course knowing (among other things):

1. Some basics of primate and human anatomy, especially of the skeleton, muscles and brain.
2. Anatomical features of the order primata and of major groups of primates.
3. The elements of evolutionary biology and the evidence for human evolution.
4. The broad patterns of evolution for the primates and humans, including major evolutionary trends.
5. The basis for human physical variation across the world and its effect on human diet and disease.
6. The evolutionary basis of modern human structure, with particular reference to the upper and lower limb, brain, birth canal and vocal apparatus.

The University of NSW has developed a list of graduate attributes (see <https://medicallsciences.med.unsw.edu.au/students/undergraduate/advice-students> ). This course and the required assessments will assist the student to develop skills in all of these areas.

## **ASSESSMENT**

### **one poster assignment, two spot tests, final theory exam**

- The poster/oral presentation is worth 20% of the final mark and will be assessed by the tutor. Students will be assessed both on the poster itself and the oral presentation. Each of these components will have equal weighting in determining the final mark for the paired tasks.
- The two spot tests will each be worth 20% of the final mark. The first spot test will be held at approximately 50% of the course duration and the second at course end. Each will cover the preceding half of the practical classes. Spot tests are held in 101 (Gross Anatomy Lab) of the Wallace Wurth Building.
- The final theory examination is worth 40% of the final mark and will be assessed by the course authority. The 2 hour examination will include 40 multiple choice questions and 3 short essay questions.

## **RESOURCES**

- The course will not require any special library resources. Students will be accessing eJournals to prepare their poster/oral presentations.
- All practical classes, tutorials and spot-tests will take place in the Gross Anatomy Laboratory (101) of the Department of Anatomy, School of Medical Sciences. All models and specimens required for the course are already available in the collections of the Department of Anatomy.
- Adaptive tutorials will be provided for the students to reinforce concepts introduced during lectures and practical classes. These will be developed by the course authority using Moodle and the SmartSparrow platform.

## **PREREQUISITES**

The course has been given a level 2 identifier, but can be taken at any level, even level 1. There are no prerequisites for the course because all necessary knowledge (e.g. elementary genetics and principles of evolution) is included within the course structure. This has been done to maximize the accessibility of the course for students with non-scientific backgrounds.

## Lecture and Practical/Tutorial Schedule

WEEK 1		
<b>Day 1</b>	<b>Monday 4<sup>th</sup> January (5 hours)</b>	
10-11	Lecture 1	Introduction to Primate Biology (KA)
11-12	Lecture 2	Elements of Genetics (CL)
12-1	Lecture 3	Diversity and Evolution (CL)
2-3	Lecture 4	Ethics of Human Remains and Forensic Anthropology (CL)
3-4	Film	Ape and Human Behaviour (KA)
<b>Day 2</b>	<b>Tuesday 5<sup>th</sup> January (5 hours)</b>	
10-11	Lecture 5	Principles of Paleoanthropological Techniques (KA)
11-1	Practical 1	Primate Musculoskeletal Anatomy (KA)
2-3	Lecture 6	The Origin and Early Evolution of Primates (KA)
3-3.30	Lecture 4 cont.	Interpreting human bones (CL)
<b>Day 3</b>	<b>Wednesday 6<sup>th</sup> January (5 hours)</b>	
10-11	Lecture 7	Early Hominins (KA)
11-12	Lecture 8	<i>Homo ergaster</i> and <i>Homo erectus</i> (KA)
12-1	Films	Portrayals of Human Ancestors (KA)
2-3	Lecture 9	Archaic <i>Homo sapiens</i> (KA)
3-4	Tutorial 1	Group Orientation and Choosing of Poster Topics (KA)
WEEK 2		
<b>Day 4</b>	<b>Monday 11<sup>th</sup> January (5 hours)</b>	
10-11	Lecture 10	Modern <i>Homo sapiens</i> (KA)
11-1	Practical 2	Cranial Anatomy of Australopithecines and Early Humans (KA)
2-3	Lecture 11	Humans in Australia (CL)
3-4	Lecture 12	Humans in the Americas (KA)
<b>Day 5</b>	<b>Tuesday 12<sup>th</sup> January (5 hours)</b>	
10-11	Lecture 13	Evolution of Human Behaviour (KA)
11-12	Lecture 14	Origin and Mechanics of Bipedalism (CL)
1-3	Practical 3	The Human Lower Limb and Bipedal Locomotion (CL)
3-4	Lecture 15	Human Sexuality and the Problems of Human Childbirth

		(CL)
<b>Day 6</b>	<b>Wednesday 13<sup>th</sup> January (5 hours)</b>	
10-12	Practical 4	Human Childbirth (CL)
1-2	Lecture 16	The Comparative Anatomy and Function of the Hand (CL)
2-4	Practical 5	The Human Hand and Tool Use (CL)
<b>WEEK 3</b>		
<b>Day 7</b>	<b>Monday 18<sup>th</sup> January (5 hours)</b>	
10-11	Spot test 1 (based on practical classes 1 to 4)(KA)	
11-12	Lecture 17	The Hominin Brain (KA)
1-3	Practical 6	The Human Brain (KA)
3-4	Lecture 18	Language, Speech and the Human Face (lecture/film)(KA)
The remainder of week 3 is allowed for poster preparation.		
<b>WEEK 4</b>		
<b>Day 8</b>	<b>Monday 25<sup>th</sup> January (5 hours)</b>	
10-12	Practical 7	The Human Face and the Functional Anatomy of Language (KA)
12-1	Lecture 19	Variation and Adaptation of Modern Humans (CL)
2-3	Lecture 20	Changing Patterns of Disease during Human History (KA) Food and Diet (CL)
3-4	Lecture 21	Syphilis, Tuberculosis and HIV/AIDS (KA)
<b>Day 9</b>	<b>Wednesday 27<sup>th</sup> January (5 hours)</b>	
10-11	Lecture 22	Malaria and Human Variation (CL)
11-1	Tutorial 2	Presentation of Group Posters (KA)
2-4	Tutorial 3	Presentation of Group Posters (continued)(KA)
<b>Day 10</b>	<b>Thursday 28<sup>th</sup> January (3 hours)</b>	
10-12	Final examination (KA)	
3-4	Spot test 2 (based on practical classes 5 to 7)(KA)	

KA – Prof Ken Ashwell

CL – Dr Carol Lazer

# Lecture 1 Introduction to primate biology

## Specific Objectives:

1. To be able to list the key characteristics of primates.
2. To know the main groups of living primates.
3. To be able to explain the key features of primate ecology.
4. To be able to summarize the main threats to the biodiversity of primates.

## The key characteristics of primates

These are set out in detail in the class notes for practical 1. Primates are, of course, mammals, which means that they possess body hair, milk (mammary) glands and maintain relatively high body temperature by internal metabolic activity (endothermy). Many of the features that characterize primates are actually 'generalized' features. In other words these mammals have not developed a specialized form in response to evolutionary pressures as have horses or dolphins, but have retained many of the features of the early mammals. This has also meant that primates have been able to evolve in a number of different directions, which explains the great diversity seen among living primates. This is readily seen in the primate upper limb, which has retained some bones that have been lost in other mammals (e.g. the clavicle or collar bone). Primates have a generalized limb structure, which allows them to engage in a range of locomotor activities (e.g. walking on the ground on four limbs, or quadrupedalism, as seen in baboons, or swinging through trees beneath branches, brachiating or arm-swinging, as seen in gibbons). Other important features include a tendency towards an upright body posture, flattened nails (in some, but not all primates), binocular vision, colour vision, grasping hand (some with opposable thumbs or big toes), a general lack of dietary specialization, enlarged brain, a prolonged period of dependency of the infant on the mother and a tendency to live in social groups. Several of these features (binocular vision, grasping hand, vertical posture) are excellent adaptations to life in the trees leading to an **arboreal hypothesis** for primate origins. On the other hand, it has also been noted that binocular vision would have been of great benefit if early primates stalked and captured fast-moving insect prey using vision (**visual predation hypothesis**).

Primate teeth are generalized in form, meaning that these animals can eat many different types of food (e.g. fruit, leaves, insects, birds, amphibians and occasionally other mammals). Primates from the Old World (Africa, Europe and Asia) have 2 incisors, 1 canine, 2 premolars and 3 molars on each side of each dental arch. This is known as the **dental formula** and is seen in humans as well as Old World monkeys and apes. New World monkeys have three premolars, whereas the strepsirhines (prosimians) have varying dental formulae.

Most primates are active by day (**diurnal**) whereas the strepsirhines tend to be active by night (**nocturnal**). Primates have shifted from olfactory dependence (as seen for many other terrestrial mammals, e.g. dogs, rats) to visual dependence with colour vision. The ability to detect colour is beneficial in assessing the ripeness of fruit and in detecting insect prey against foliage (see below).

## The main groups of living primates

Primates are usually divided into two suborders: **Strepsirhini** (strepsirhines, including lemurs, galagos and lorises) and **Haplorhini** (haplorhines, including tarsiers, monkeys, apes and humans). The haplorhines have enclosed orbits (eye sockets), enlarged brains and fused frontal bones and mandibles. Within the **Haplorhini** there is a subdivision into three groups: Superfamily **Ceboidea**,

containing new world monkeys, **Cercopithecoidea** containing Old World monkeys, and **Hominoidea** including **apes (gibbons, orangutans, gorillas, chimpanzees and bonobos)** and humans.

We shall review the main points of the different primate types in turn:

#### **A) Prosimians**

- Most similar of all primates to the early mammalian ancestor.
- Include lemurs (Madagascar) and lorises (Africa, India, SE Asia).
- Pronounced reliance on olfaction – reflected in the moist fleshy pad (rhinarium) at the end of the nose.
- More laterally placed eyes than other primates.
- Different uterine structure from other primates.
- Dental comb specialization of the lower incisors and canines.
- Grooming claw on second toe.

#### **B) Tarsiers**

- All live in southeast Asian islands.
- Possess both retained ancestral traits (grooming claw and unfused mandible) and advanced traits (absence of rhinarium and dental comb).
- Large eyes dominating the face.
- Lower limbs adapted for leaping.
- Believed to occupy an evolutionary position between prosimians like the lemurs, and haplorhines like the monkeys and apes.

#### **C) New World Monkeys**

- Found in central and south America.
- Almost exclusively arboreal.
- Broad, widely flaring noses with outward facing nostrils (platyrrhine – flat-nosed).
- Include four families: Atelidae – howlers and spider monkeys; Callitrichidae - marmosets and tamarins; Cebidae – capuchins; Pitheciidae – sakis.
- 3 premolars in each dental quadrant instead of 2 as in Old World monkeys.

#### **D) Old World Monkeys**

- Found in Africa, Asia, India.
- Most are quadrupedal and primarily arboreal.
- Noses are downward facing (catarrhine).
- Females of some species exhibit pronounced cyclical changes in the appearance of the external genitalia (associated with estrus).
- Sexual dimorphism (differences in size between genders) is typical of some terrestrial species and is particularly pronounced in baboons.

#### **E) Hominoids**

- 3 families (**Hylobatidae** - gibbons and siamangs; **Pongidae** – orangutans, gorillas, bonobos and chimpanzees; **Hominidae** – humans).
- usually larger than monkeys.
- no tail, shorter trunk than monkeys.
- more complex brain and behaviour than monkeys.
- increased infant dependency.

Interestingly, chimpanzees, bonobos (pygmy chimpanzees) and humans share more genetic similarity than do zebras and horses, or goats and sheep. On this basis it would be appropriate to group humans and chimpanzees within the same family and perhaps the same genus (*Homo*). Humans have 46 (i.e. 23 pairs of) chromosomes, while chimpanzees have 48 (i.e. 24 pairs of). The banding pattern of human chromosome 2 corresponds to those of two much smaller chimpanzee chromosomes (chromosomes 12 and 13). This finding has led to speculation that in an ancestral hominin (human), these two chromosomes fused to produce what became human chromosome 2.

### **Primate chromosomes, proteins and DNA**

Relationships between primates may be revealed by analysis of biochemical and cytogenetic features. These include:

- Karyotyping – the analysis of chromosome shape, size, number and banding patterns.
- Amino acid sequencing – e.g. of blood proteins such as hemoglobin. The more distantly related the primate, the more amino acid substitutions in a given protein.
- DNA hybridisation – testing for differences and similarities between DNA sequences from different primates. Evolutionary similarities/differences are calculated from the number of mismatched base pairs along a hybrid DNA sequence from two primates.
- DNA sequencing – involves determining directly the sequence of nucleotides along a strand of DNA. This data is already available for humans as part of the Human Genome Project, but is not yet available for all primates.

### **Primate ecology**

Most primates are herbivores, existing on a diet of leaves and/or fruit. Some strepsirrhines eat insects and the tarsiers are exclusively carnivorous, living on insects, lizards, frogs and other small animals. Fruit eating primates (**frugivores**) must be able to assess when fruit is ripe (colour vision) and cope with the seasonal availability of their food. Leaf eating primates (**folivores**) must cope with the poor nutrient and calorie supply in their diet. Folivores prefer the youngest leaves and shoots because these have the highest ratio of nutrients to indigestible fibre. Plants also produce toxic compounds (tannins, phenols and alkaloids) in their leaves to discourage attacks by folivores. Baboons, chimpanzees and humans supplement their diet with the flesh of other animals.

The diet of a primate greatly influences its anatomy and physiology. Folivores often have large gastrointestinal tracts, sometimes with specialized stomachs for fermentation of the cellulose in plant food. Insectivorous primates tend to be small, because insect eating is more costly in time and energy for large primates, who tend towards folivory.

All mammals live in defined regions called **home ranges**, which must contain all the resources needed by a primate group (water, food, shelter, mates). That part of the home range used most intensively is the **core area**.

### **Primate conservation**

Many primates live in habitats that are under threat from human activities. There are three reasons for the decline in primate numbers worldwide:

- Habitat depletion
- Hunting
- Live capture for export or local trade

## Lectures 2 & 3 Elements of Genetics / Diversity and Evolution

These lectures introduce concepts and terminologies (marked in bold) that are used in many of the subsequent lectures and practical classes throughout the course. The detailed notes are intended as reference and general background for non-biologists.

It should not be necessary to take additional notes so you can relax and concentrate on the images and concepts. What you need to know is included in the lecture notes.

### Lecture 2 Elements of genetics

#### Specific Objectives

1. To describe the location, structure and function of the genetic material (DNA), including the processes of the molecule copying itself (replication) and reading of the genetic code to make proteins (transcription and translation).
2. To list types of coding errors (mutations) with examples of their effects.
3. To know basic terminology and patterns of inheritance of simple genetic traits.

#### Introduction

Evolution is a process that changes the form of **populations** (groups of breeding individuals) over successive generations. To understand the mechanics of this process it is necessary to know some basic molecular and general genetics concepts and terminology.

All organisms (plants, animals, bacteria, etc) are made of **cells**, which contain their own organs (organelles), are able to copy themselves, and can produce the molecules necessary for survival. An important group of molecules, called **proteins**, perform numerous functions including: structural (*fibres, like collagen, elastin, etc*), chemical (*enzymes that act as catalysts for chemical reactions*), communication (*hormones*), transport (*carriers for other molecules, like oxygen and fats*), buffers (*to maintain constant blood pH*), etc. All proteins are constructed according to coded instructions stored in the genetic material, which is another important group of molecules, called **nucleic acids**. There are two forms of nucleic acid, DNA and RNA.

Every organism inherits the specific details to make all its proteins. This comprises the **genetic code** or **genome**. All organisms which share common codes for their proteins form distinct species, whereas slight variations in parts of this code make all individuals unique. How these variations are inherited in a population forms the basis of the process known as **evolution**.

#### The genetic material

The genetic code is carried on long molecules of DNA, known as **chromosomes**, which are found in the nucleus of cells. The molecular structure of DNA and RNA explains both how the code is conserved and how it is read.

The DNA molecule is made up of atoms of carbon, hydrogen, oxygen, nitrogen, phosphorus, etc, to form two spiral strands called a **double helix**. Each strand is a chain of alternating sugar and phosphate molecules linked together by pairs of molecules, called **bases**. A unit of sugar, phosphate and base is called a **nucleotide**. Each long chain in a nucleic acid comprises a string of joined nucleotides. The cross-links between the strands are made by four bases which link in specific pairs: Adenine always pairs with Thymine (two hydrogen bonds A=T) and Cytosine always pairs with Guanine (three hydrogen bonds C≡G). **DNA**

(deoxyribonucleic acid) is a double helix (two stranded) with deoxyribose as the sugar molecule. **RNA** (ribonucleic acid) is a single helix (one stranded) with ribose as the sugar molecule and the base Uracil instead of Thymine (A=U).

The genetic material is conserved, in that exact copies are made automatically (**replication**) each time a cell divides. An enzyme splits the hydrogen bonds between the bases separating the two strands of each DNA molecule. Nucleotides present in the nucleus match up with the unpaired bases like jigsaw pieces to re-establish the complementary strands using each split original as a template.

The order of the bases along the DNA molecule is the **genetic code**. DNA holds the whole code in the nucleus but a piece of code copied to an RNA molecule (**transcription**) can be carried from the nucleus to ribosomes in the cytoplasm, where the sequence of bases in the RNA will be “read” to assemble a specific protein molecule (**translation**).

To achieve this, a section of the DNA of a chromosome is split open by an enzyme, as in replication, and a string of code (**gene**) on one strand is copied, or transcribed, as RNA to carry the **complement** of that code from the nucleus into the cell. This messenger RNA (**mRNA**) attaches to ribosomes, the sites where it is decoded, or translated, and protein construction occurs.

**Proteins** are made of strings of a group of chemicals called **amino acids**. There are 20 different amino acids and their sequence determines the properties of the protein, such as how the molecule will fold and interact with other molecules. The final shape of the protein is critical to its proper function.

Floating in the fluid cytoplasm of the cell are specially folded RNA molecules that carry amino acids to the ribosomes for protein assembly. This transfer RNA (**tRNA**) has three unpaired bases at one end of the molecule and the particular amino acid coded by the complement of that sequence at the other end. Hence, the genetic code is read in “words” of three “letters” (triplets) from an “alphabet” of four bases. A code of doublets would allow 4x4 or 16 combinations, which is not enough for 20 amino acids. A code of triplets enables 4x4x4 or 64 combinations to provide coding redundancy as well as start and stop codes. Each **triplet** (three bases) on the mRNA is called a **codon** and the complimentary set of three bases on the tRNA is called the **anticodon**. Only one anticodon will pair with the next codon on the mRNA at the binding site on the ribosome, so only the tRNA carrying the next correct amino acid will be brought alongside the growing amino acid chain to link with it. As the chain gets longer each disconnected tRNA molecule is released and recycled to collect its specific replacement amino acid in the cytoplasm. This process produces the primary structure of a protein, the amino acid sequence. It will then fold into simple helix or pleated sheet sections to form the secondary structure, which will further fold into a complex 3-D shape to form the tertiary structure. Some proteins form a quaternary structure where two or more tertiary protein chains bind to each other. An example is haemoglobin in red blood cells which is made of four molecules, two  $\alpha$  chains and two  $\beta$  chains.

If a change occurs in the genetic code, often as an error in replication, it will result in misreading of the codons with a subsequent change to the final protein. This is called a **mutation**. Typical errors include: deletion (loss of a base), addition (an extra base), or substitution (replacement of the correct base by another). One or more mutations may change the ability of a protein to function correctly. This might have no obvious effect, or cause an illness if it occurs in body cells (benign tumour or cancer), or an inherited condition if it occurs in sex cells (abnormality or genetic disease).

The influence of mutation can be quite varied depending on the protein affected. A change to a structural protein can affect its function, such as a single

base substitution in the  $\alpha$  chain of haemoglobin to produce sickle cell anaemia. Abnormal enzymes interfere with metabolic pathways. Inheritance of different ineffective enzymes causes very different illnesses, even when they belong to the same biochemical pathway. A defect in any enzyme along the path, such as the conversion of food from the diet to the pigment molecule, melanin, will cause the process to stop at different points. For example, if the enzyme that converts phenylalanine (an amino acid from protein in our food) into tyrosine (another amino acid) is defective, then phenylalanine will build up to toxic levels in a baby and cause brain damage with mental retardation, a disease known as phenylketonuria. If a different enzyme that converts a metabolic product of tyrosine to melanin is defective, the individual will lack pigment, a condition known as albinism. Note that sufferers of phenylketonuria have the pale appearance of an albino, since the metabolic pathway has been blocked further back than the chemical product needed by the other normal enzyme to produce melanin

These concepts and processes form the underlying mechanics of genetics and evolution.

### **Inheritance of characteristics**

During most of the time in the cell cycle the individual, long molecules of DNA (**chromosomes**) cannot be seen (interphase) but they become highly coiled and shortened during cell division (**mitosis**), which occurs in several stages (prophase, metaphase, anaphase, telophase) and are easily seen under the microscope. They are often shown in photographs as stained chromosomes spread out from the metaphase stage of cell division. With special stains the chromosomes show consistent patterns of light and dark bands. The photographed images of these chromosomes are arranged and numbered in order by size and the position of the constriction (**centromere**) to produce a **karyotype**. All the chromosomes in a nucleus form an individual's karyotype, which is typical for the species. In humans there are 46 chromosomes, being two sets of 23 chromosomes, one from each parent. Manipulation of the physical properties of DNA in the laboratory shows that chimpanzees and humans share around 98% of their DNA (the number varies slightly with sources) but some of it appears to be distributed differently. Human chromosome 2 corresponds in banding pattern to the two smaller chimp chromosomes 12 and 13. This would explain why humans have 46 chromosomes and chimps have 48, suggesting fusion in the hominid line of two chromosomes that remained separate in the ape line (or vice versa).

The base sequence on each chromosome codes for many different proteins and the code for any one protein is called a **gene**. The gene for each protein will occupy the same location (**locus**) on the same chromosome in all individuals. Since chromosomes occur in duplicate there are two copies of the same gene for each protein. Each copy is not always identical with slight code variations (from mutation), called **alleles**. An individual can carry two copies of the same allele (**homozygous**) or two different alleles (**heterozygous**) inherited from their parents. When two different alleles occur together the outward appearance will usually be of only one of them and that one is called **dominant**. The other is called **recessive**. The alleles carried (**genotype**) produce the outward appearance (**phenotype**). As a simplified example, genes for the pigment molecules of eye colour code for pigment proteins that can vary in size in the iris of the eye. The small molecules scatter the light to give a blue colour, while large molecules absorb light to give a brown colour. By convention the variants (**traits**) are identified by a letter from the alphabet with the dominant trait capitalized. Brown eyes (large molecules) are dominant to blue eyes (small molecules), so the genotypes homozygous brown (BB), homozygous blue (bb)

and heterozygous Bb will have the phenotypes of brown eyes, blue eyes and brown eyes respectively.

Knowing this allows understanding of the patterns of inheritance. A blue eyed parent must be bb and a brown eyed parent could be BB or Bb. If a child has blue eyes (bb), the brown eyed parent must be Bb and the probability of blue eyes is 0.5 or 50% for each child. If two brown eyed parents have a blue eyed child, then they must both be heterozygous (Bb) with a 0.25 or 25% probability for each child having blue eyes.

## Lecture 3 Diversity and evolution

### Specific Objectives

1. To list several sources of variation and diversity.
2. To describe the principles of mitochondrial and Y chromosome inheritance.
3. To be able to discuss key concepts of population change and the process of evolution.

### Generating Diversity

The process of DNA replication occurs whenever the cells of the body divide (**mitosis**) or when the sex cells of the body are formed (**meiosis**). In mitosis each cell division produces two identical daughter cells, each with the same DNA. In meiosis the genetic material is rearranged and the DNA content halved through two cell divisions. The two sets of chromosomes (maternal and paternal chromosomes) are randomly separated (**independent assortment**) with some exchange of complementary genetic material between maternal and paternal chromosomes (**recombination**). In meiosis four sex cells are produced, where each has half the chromosome number and none are genetically identical. This ensures that genetic variability is maintained between the generations independently of new mutations. Note that the **purpose of sex** is to contribute a genetic remix from each parent to produce new genetically related but unique individuals.

This can be illustrated by another simplified example using two genes, eye colour and hair colour. Hair works in the same manner as eye colour with the allele for dark hair (D) being dominant to fair hair (d). A blue eyed / dark haired parent (bbDd) and a brown eyed / fair haired parent (Bbdd) could have the following children: blue eyes / dark hair (bbDd), brown eyes / fair hair (Bbdd), both looking like one of their parents, or blue eyes / fair hair (bbdd), brown eyes / dark hair (bBDd), both unlike either parent with new phenotypes.

Some genes have more than two alleles but only a maximum of two will occur in any individual. For a gene with two alleles only two phenotypes are possible. For a gene with three alleles more variations are available. One of the red blood cell groups is coded by a gene with three alleles ( $I^A$ ,  $I^B$ , i), two being equally dominant (codominant) and one recessive, to produce four phenotypes (A, B, AB, O). In the case of tissue types, several genes are involved (with 4-5 routinely tested), and each has multiple alleles (6-12, or more). It is easy to see why it is so difficult to find compatible tissue matched donors for transplants when so many available combinations make each individual effectively unique. Inherited variation can be even more complex than this. Some phenotypes result from the interaction of multiple genes and environmental influences, known as **multifactorial inheritance**. Two examples are height and intelligence, where the combined genetic potential of many genes can be influenced by such factors as nutrition, childhood illness, education, etc. This makes the allocation of characteristics to racial types very tenuous.

### The female line (mitochondrial Eve) and the male line (Y chromosome)

There are two kinds of DNA in a cell. Most is found in the nucleus (**nuclear DNA**) but a small amount is present in an organelle called the mitochondrion (**mitochondrial DNA**). Mitochondria are the site of cellular respiration and are thought to have been bacteria that now live exclusively inside cells. They have maintained their own DNA for self-replication and to produce the enzymes necessary for the conversion of sugar and oxygen to energy.

Sex in most species is determined by two special chromosomes, a medium sized X and a small Y chromosome. In humans, the 46 chromosomes comprise 22 pairs of chromosomes common to both sexes (**autosomes**) and one pair of **sex chromosomes**. Females have two X chromosomes while males have an X and a Y. The egg will carry one of the two maternal X's while the sperm can carry either an X or a Y, so it is the father that determines the sex of the offspring.

The egg is a gigantic cell, while the sperm is tiny in comparison. All the cytoplasm and organelles in the cells of our bodies originated from the cytoplasm of the egg, whereas the only contribution of the sperm is half the nuclear DNA. This means that all our mitochondria and its DNA came from our mothers. Similarly, the Y chromosome, common to all men, could only be carried by the sperm and so all Y chromosomes in men came from their fathers.

Stable DNA avoids the processes that generate diversity and changes very little over time. It includes mitochondrial DNA that does not undergo meiosis, or genes unique to the Y chromosome that do not take part in recombination during meiosis. Studies of the diversity of genes of stable DNA are used compare the relative ages of populations. Older, ancestral populations accumulate more genetic changes, through non-lethal mutations, than recently separated populations. If all mitochondrial DNA is inherited through the female line we can theoretically "look back" to the original mother of the line, the so called **mitochondrial Eve**. The same principle applies through the male line by looking at the **Y chromosome segment** that is unique and does not pair with the X chromosome.

### **How evolution works**

Physical form is strongly related to function and both are influenced by environmental conditions. Evolution, often referred to as **Darwinian evolution**, is a process that acts on the form and function of individuals in the short term and changes populations in the long term. Genetic variation within populations (**gene pool**) provides the basic material through which evolution acts by changing gene (allele) frequencies. In this context the populations are called **species**, which are true breeding groups that cannot successfully interbreed with other groups. Any isolated breeding group, such as separated colonies of a species, will be subject to local evolutionary pressures. If such an isolated group diverges sufficiently from the original it becomes a new species (**speciation**).

More individuals are born than will survive. In a varied population individuals will be removed from the gene pool through predation, illness, competition for nesting sites or mates, non-viable offspring, etc. Associated with environmental change, a few individuals may fare better than the rest to leave many offspring to the next generation. This process where an individual's ability to breed is affected by the environment is called **natural selection**.

Mutation is often cited in popular fiction as the source of variation which drives evolution. This is not the case as most mutations are disadvantageous (*naked neck chicken*) or lethal (*sickle cell anemia*), a few are relatively benign (*albino*), and almost none will bestow advantage in the current environment. It is **sexual reproduction** with the independent assortment and recombination of meiosis that rearranges the allele combinations and creates much of the variation in a population.

Even so, potentially fatal phenotypes do turn up in populations. Dead or disadvantaged individuals will not breed and pass their genes to the next generation but these mutations, although rare, always occur to recreate alleles that are being lost. The population frequency of a lethal allele will be close to the mutation rate. Recessive deleterious or lethal alleles are often masked by the normal dominant alleles. The frequency of hidden alleles for abnormal traits in a population is called

the **genetic load**. With a change in the environment, a normally deleterious or benign allele may confer advantage, so this load is a potentially important source of hidden variation. For example, drug resistant properties in the chemical makeup of a few bacteria in a population may be of no value, or even a slight disadvantage, till the appropriate drug is encountered in the environment. Then the resistant bacteria will have a selective advantage (they will not be dead) and so produce most of the offspring of the next generation to become a drug resistant strain.

The concept of fitness is another Darwinian idea that has been corrupted by popular fiction. **Fitness** does not refer to the physical strength or aggression of an individual but to how many offspring that individual leaves to grow up and reproduce. Only those individuals well suited to the environment will successfully produce offspring and pass their genes (allele combinations) to the next generation. For example, rare white rabbits in woodland are easily seen by predators and so have a low fitness (no offspring) compared to the other well camouflaged, brown rabbits. A change to the environment where snow becomes predominant would confer a much higher fitness to any white rabbits that are now better camouflaged than the brown rabbits. With increased reproductive success the allele frequency for white rabbits, and subsequently the number of white rabbits, will increase in the population.

It should be noted that evolution can never “design” a perfect or novel organism. New structures cannot be created but existing structures can be modified to fit a new function (**divergence**). For example, the basic body structure is four limbs, so wings can develop from modified forelimbs, such as a bird or bat, but another pair of limbs cannot be produced from nothing to form a four legged and winged dragon. Change can only build on what is already present (**homology**) to give the best fit to the environment at the time with the best **compromise**.

If the environmental challenges are the same the solutions will be similar, leading to **convergent evolution**. Two examples are adaptations to exploit air and sea. More than one group of plants and animals have independently developed flight with membranous extensions common to winged seeds, flying squirrels and gliding possums, and unrelated modifications of the forelimb to become wings in flying prehistoric reptiles, birds and bats. The penguin, seal and whale have undergone similar changes for a life dependent on swimming with conversion of limbs to paddles and rudders and a body shape similar to fish. These structures look similar because they are the best responses to the common needs imposed for staying airborne or moving efficiently underwater. Another less extreme example is the tendency for many species to develop large, heavy jaws and teeth associated with a high proportion of tough vegetable fibre in the diet.

Populations are shaped by their environment but there is no such thing as the perfect population. Environments will always change forcing population change, a process known as **dynamic equilibrium**. A documented example for a species of moth in England illustrates this principle well, where colour change occurred in moth populations in response to environmental change. A population of light coloured moths had evolved to camouflage against pale lichens on the bark of trees. Black moths would occasionally occur but these had low fitness as they were easily seen by birds and eaten. The industrial revolution covered the trees with black soot. The light moths, now easy to see, had reduced fitness while the fitness of the rare, dark moths increased. Over a several generations the population frequency of the allele for dark colour increased, to produce a population of dark moths. Occasional light moths would continue to occur with low fitness. Conservation efforts eventually forced industries to clean up with a drop in the level of pollution. With less soot the pale lichens were able to re-colonise the tree trunks. The dark moths were now at a selective disadvantage, while the rare light moths survived to produce many young

and so change the gene frequency again; this time to favour the allele for light colour. Over several generations, the moth population changed back to its original pale colour with the occurrence of occasional dark moths.

Highly specialised species can be too closely tied to their environment. It can spell disaster if the environment changes too radically or too rapidly, as it takes generations to effect population change. For example, koalas eat leaves from a limited range of trees and risk extinction wherever their forests become threatened. The possum has a very flexible diet and easily adapts to new environments, like the urban landscape that replaces their native vegetation. It is no accident that a generalised animal, like the shrew, was the forebear of the primates as it formed an uncomplicated “blank slate” for physical adaptations to exploit life in the tree tops.

There is **no direction** in evolution. There is no such thing as an evolutionary hierarchy to a superior form over an inherently primitive form. This notion is another legacy of popular culture which suggests that humans are the pinnacle of primate evolution. It is often projected further to predict a future, mentally superior being with a small body and very big head, much like most popular representations of aliens. Evolution cannot produce a final form, only a current form with the best fit to the current environmental conditions.

It can be argued that modern humans are the only exception to the evolutionary process as they tend to modify their environment to suit themselves rather than be modified by their environment. For example, humans have rapidly spread into a wide variety of climatic extremes without modification of form. Any other species would need generations of time to develop major adaptations to a new environment, such as thick fur to protect from extreme cold. However, there are many examples of human variation that has been influenced by evolutionary pressures.