VICTORIA UNIVERSITY PRESENTS THE...

2011 VCE LECTURE SERIES

HEALTH, ENGINEERING AND SCIENCE

VCE BIOLOGY
UNIT 4
AREA OF STUDY 1: HEREDITY

REVISION NOTES

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VICTORIA UNIVERSITY
MELBOURNE AUSTRALIA

BACHELOR OF SCIENCE (BIOMEDICAL SCIENCES) VTAC code 40181

You could play an important role in the search for cures of life threatening diseases, be involved in the marketing of these discoveries, or be the link between scientists and the public.

This appropriately tailored course qualifies students for entry to a broad range of careers including:

medical and scientific research, sales and marketing of biomedical products and health promotion. This degree leads on to postgraduate programs in medicine, dentistry, nursing, physiotherapy, dietetics and other allied health courses. This degree also offers a fourth year (Honours) program, with the possibility of Masters and PhD.

The Biomedical Sciences course aims to produce highly flexible but well-trained graduates who will be adequately equipped to adapt to a changing workforce environment. This course offers a range of core and elective units from biomedical sciences, as well as electives from other courses within the university, such as languages, sport and exercise, creative arts, psychology, chemistry, mathematics.

The core program consists of modern and traditional biomedical sciences including units of study such as: anatomy, physiology, biochemistry, microbiology, cell and molecular biology, immunology, pharmacology, pathophysiology, wellness and health management.

The Biomedical Sciences degree satisfies all the undergraduate selection requirements for post graduate medicine at all Australian Universities including prerequisites in anatomy, physiology and biochemistry. However, entry into medicine or other allied health related courses at other universities may depend on other specific entry requirements at these institutions.

Prerequisites: Units 3 and 4 – English (any). Middle Band: A study score of at least 25 in one or more health and human development, mathematics (any), physical education or science (any) = an aggregate 3 points higher per study, to a maximum 9 points.

BACHELOR OF SCIENCE (NUTRITION, HEALTH AND FOOD SCIENCES) VTAC code 44251

Nutrition, Food and Health Science involves the study of human nutrition, functional foods and nutraceuticals, safety and quality of food, food processing and their role in health. It includes the application of cutting-edge technologies in nutrition and food sciences. The course is designed to develop the knowledge and skills in the science of food, its safety and quality as required by today's nutritionists and food scientists. Increasing consumer awareness in regard to food related health and safety issues and the role of nutrition in the development and evaluation of food products have generated a rapidly growing need for graduates with a good understanding of food science, nutrition and health. The course has been specifically designed to meet the demand for such graduates. When you graduate, you will be qualified to contribute to the development of new foods and, to ensure their safety and provide advice and information to consumers.

Employers of our past graduates include:

- Kraft Foods Ltd
- Nestlé Australia
- Food Science Australia
- Australian Quarantine Inspection Service (AQIS)
- CSL Ltd
- Cadbury Schweppes Aust Ltd
- Heinz – Watties Aust
- McCains Foods (Aust) Pty Ltd

Prerequisites: Units 3 and 4 – English (any) and mathematics (any).

SCHOLARSHIPS ENTER AT 70

Available for Nutrition, Health and Food Sciences course for students with an ATAR (Enter) of 70 and above who meet the requirements. See www.vu.edu.au/hes for more information and application form.

ALTERNATIVE ENTRY FOR SCIENCE (VTAC code 41451)

Alternative entry program to science courses for students who have:

- Successfully completed year 12 with the required prerequisites, but may not have achieved the required study score in all prerequisites; or
- Have not studied the required mathematics prerequisite.

All admissions are on an individual basis. All applicants offered a place would be required to enroll in one or more subjects from the Foundation Year.

Prerequisites: Units 3 and 4 – English (any) and mathematics (any). ATAR (ENTER) 50+

AREA OF STUDY 1

HEREDITY

1. MOLECULAR GENETICS

THE HUMAN GENOME

- Refers to the total genetic material within a cell, individual or a species.
- Most genetic material is in the nucleus, but there is also DNA in mitochondria.
- Not all DNA codes for proteins - Only 25% of DNA codes for biological molecules – mainly proteins and some code for RNA.
- Only about 1.5% of our DNA actually codes for the production of functioning protein molecules.
- The rest is either never transcribed or never translated.

DNA (Deoxyribose Nucleic Acid)

- The inherited material that determines all characteristics of an organism.
- i.e. DNA is the genetic blue print of life. It determines what is made by a cell, when and where and by how much
- In eukaryotic cells, most DNA occurs in the nucleus but some occurs in mitochondria and chloroplasts.
- In prokaryotes, like bacteria, DNA occurs in a single circular chromosome in the cytoplasm, this is called a plasmid.
- The DNA molecule is made up of 2 strands held together with weak forces (hydrogen bonds).
- We say this is a double stranded molecule. The DNA model is called the Watson and Crick model

Diagram

- Each strand of DNA or chain is made up of nucleotides, which are the sub-units of DNA.
- A nucleotide consists of a sugar, a nitrogen base and a phosphate unit. Draw.
- The sugar is **Deoxyribose** and there are 4 different nitrogen bases that can be on each nucleotide. The symbols for each base are

A T C G

- One of the stands is called the template (sense) strand, while the other the complementary (antisense strand)
- The bases are said to be complementary.

- If the order on the template strand is as below, complete the complementary bases that make up the second strand (complementary strand)

T	T	A	A	C	G	T	A	C	G	T	T	T	C	G

- The sequence of bases on the DNA molecule is not the same in all DNA molecules i.e. the DNA sequence of one human is not the same as another human. The DNA is very closely matched but not identical. (DNA) will be identical for identical twins.
- DNA has the unique ability to replicate itself. Before a cell divides each DNA molecule can form an exact replicate.
- For a copy to be made the DNA uncoils, the weak hydrogen bonds are broken ad the 2 strands are pulled apart between their base pairs.
- The unpaired bases on each strand attract a complementary DNA nucleotide (from the cytoplasm) and bind to it. The enzyme involved is DNA polymerase.
- This is repeated for the length of the strand.

CHROMOSOMES

- Are made of a DNA molecule and its associated (histone) protein.
- These are found in the nucleus of most organisms
- Humans have 46 chromosomes in each **somatic** cell. This is call the Diploid or 2n number of chromosomes. The diploid number varies between different species. We have 26 pairs of chromosomes in each cell.
- Gametes have half the number of chromosomes. This is the n or Haploid number, which in humans is 23 chromosomes. This also varies between organisms.
- Humans have 22 pairs of homologous chromosomes called **autosomes** and 2 **sex chromosomes.**
- The 23rd pair of chromosomes in humans determines the sex of the individual. In humans - females are XX and males XY. In birds and reptiles it is the opposite - ZZ male WZ female
- Somatic cells are normal body cells
- Diploid cells have 2n chromosomes. We call these pairs, homologous chromosomes. Homologous chromosomes are the same length and have the same centromere positions and gene locations.
- We can view the chromosomes from a diploid cell in a **Karyotype**.

GENES

- Segments of chromosomes are called genes; therefore genes are made of DNA.
- Humans have large numbers of genes on each chromosome and each codes for a different characteristic or trait.
- Different genes are made of different bases. The coding region is called the exon and the non-coding section the intron.
- The position of a gene on a chromosome is called the Locus.
- Each gene directs the synthesis of one protein. Therefore a single gene is usually responsible for the inheritance of a single characteristic (monogenic).
- Different forms of one gene are called **alleles.** Alleles of one gene are usually represented by variations of one letter of the alphabet. Most genes have 2 alleles but some have more than 2 e.g. ABO blood group
- Gene Regulation
  - Sequences in the upstream region control the start of transcription and the rate at which protein is produced.
  - Sequences in the upstream region are called promoters.
  - Different genes are regulated in different ways.
  - Sometimes transcription factors bind to the upstream region, some turn genes on, others turn genes off.
  - Transcription factors are activated by intracellular or extracellular changes.

RNA (Ribonucleic acid)

- **A single stranded** nucleic acid made of nucleotides with a base, sugar and phosphate unit.
- The sugar is ribose and the 4 bases are C G A U
- Note **Uracil** instead of Thymine.
- Base pairing occurs- C – G and A - U
- There are three types of RNA :
  -Messenger RNA (mRNA) – which copies DNA in the nucleus and moves to the ribosomes
  -Transfer RNA (tRNA) – which attaches to a specific amino acid and brings it to the ribosome
  -Ribosomal RNA (rRNA) – part of a ribosome.

Template

DNA	T	T	A	G	G	C	G	C	A	A	T	A	C
RNA													

CODONS

- These are sequences of three DNA bases, which code for a specific amino acid. For example CGA codes for the amino acid Alanine.
- Organisms use the genetic code to produce proteins in protein synthesis. This occurs in two stages.

GENE EXPRESSION – PROTEIN SYNTHESIS

OVERVIEW

- Messenger RNA forms against the template strand of DNA in the nucleus
- Messenger RNA moves to the ribosomes
- Anticodons in transfer RNA pair with complementary triplets (codons) in mRNA
- Each tRNA transports a specific amino acid
- The order of the codons in mRNA determines the order of amino acids in the polypeptide chain

TRANSCRIPTION

- The enzyme **RNA polymerase** binds to the gene to be expressed and that section of the chromosome unwinds from its helix.
- On one of the strands (the **sense strand)**, a complementary **messenger RNA** molecule (mRNA) forms i.e. the DNA is copied
- Transcription ceases when the "stop" codon is reached. Introns are cut from the mRNA and it moves from the nucleus into the cytoplasm while the DNA double helix reforms.

DNA Comp.																	
DNA Template	T	A	C	C	G	A	A	A	T	C	G	A	A	G	C	G	C
m RNA																	

TRANSLATION

- The mRNA, now in the cytoplasm, attaches to a **ribosome, which** translates the MRNA **anticodons.**
- **Transfer RNA** (tRNA) molecules, carrying specific amino acids attach to their matching anticodons, peptide bonds form between the amino acids and when translation is complete. The new protein is released.
- Cells produce many proteins to carry out numerous functions.
- The amino acids needed to make proteins are present in the cytosol**.**
- Cells produce thousands of proteins. Examples are enzymes, hormones, antibodies and parts of membranes.
- Proteins are made up of amino acids linked into chains called peptides. The bondings between the amino acids are called peptide bonds.
- 20 different amino acids exist so there are 1000s of combinations that can be made.
- Remember the instructions for the protein are in the DNA which is transcribed and then translated

2. CELL REPRODUCTION

CELL CYCLE

All cells arise from pre - existing cells. New cells are produced by mitosis (nuclear division) and cytokinesis (division of other cell components). The life cycle of a cell can be divided into phases:

- M phase – cell divides
- G1 phase – cell grows, organelles replicate
- S phase – DNA synthesis, chromosomes are replicated
- G2 phase – cells grow and prepare for the next cell division.

Different types of cells have different life spans – stomach cells for a few days skin cells for a few weeks, red blood cells for a few months and brain cells a life time.

APOPTOSIS

- This is the programmed death of cells.
- In healthy tissue, cell death is balanced by the production of new cells from mitosis.
- Too much apoptosis results in a reduction in cell numbers e.g. Alzheimer's disease
- Too little apoptosis leads to an increase in cell number and a tumour develops.
- This process is important in: developmental changes, removing old, infected or damaged cells, removing immune cells which attack self cells and removing cells which have DNA damage (if not removed cancers develop)

MITOSIS

- In the process of Mitosis, a eukaryotic cell divides to produce two identical daughter cells with DNA identical to that of the parent cell.
- Cells that undergo mitosis (not involved in gamete formation) are called **somatic** cells in animals and the growing regions of plants.
- The purpose of mitosis is for growth and repair.
- The phases are IPMAT

Stage 1 Interphase

Between divisions, each chromosome replicates so that it consists of two identical chromatids, held together by the centromere.

Stage 2 Prophase

The nuclear membrane breaks down; the double chromosomes shorten and thicken so they are visible under the light microscope. In animal cells, the centrioles move to opposite poles and anchor the spindle fibres.

Stage 3 Metaphase

Double chromosomes, attached to spindle fibres by the centromeres, line up in the middle of the cell

Stage 4 Anaphase

Centromeres divide and the daughter chromosomes (formerly chromatids) move to opposite poles.

Stage 5 Telophase

Two new nuclei form ad cell division is completed

MEIOSIS

- Occurs in gonads
- One diploid cell undergoes two cycles of division.
- In meiosis four haploid cells are produced each with half the number for chromosomes as the parent cell.
- The purpose of the process is to produce gametes for sexual reproduction.
- The 2 divisions are IPMAT and PMAT
- Produces gametes with random combinations of alleles of each gene.
- Leads to great variation in organisms
- Can produce gametes with new combinations of alleles from crossing over.

DIFFERENCES BETWEEN MEIOSIS AND MITOSIS

- At Metaphase 1, the double chromosomes line up in **homologous** (or matching) pairs. **Crossing over** (exchange of genetic material) can occur at one or more places between adjacent chromatids from different chromosomes. The point where crossing over occurs is a **chiasma.**

- The centromeres DO NOT separate at Metaphase 1 and double chromosomes (each with two chromatids) move towards each pole at Anaphase 1.

- In the second division cycle. All cells (gametes) are now haploid (half the usual chromosomal complement). As a result, many of the chromosomes present in the four haploid cells will contain one or more chromosome segments from both of the original homologous chromosomes and thus be identical to neither of them. Chromosomes different from those in the parent cells are called **recombinant** chromosomes. This exchange of genetic material results in greater diversity of offspring and increases the species' ability to adapt to change.

ERRORS IN MEIOSIS

- ## ANEUPLOIDY

  - Refers to the inheritance of too few or too many chromosomes.
  - Occurs when a centromere fails to separate at Metaphase 1 (non-disjunction).
  - The most common is the inheritance of 3 copies of a homologous chromosome rather than 2.
  - The most common is Trisomy 21 or Down syndrome
  - This can occur in autosomes and sex chromosomes.
  - Other examples include; trisomy 13 - Patau Syndrome, XXY – Klinefelter Syndrome and XO – Turner Syndrome

- ## POLYPLOIDY

  - Refers to the inheritance of additional full sets of chromosomes.
  - Most common in plants e.g. apples mostly 3N

ASEXUAL AND SEXUAL REPRODUCTION

- Asexual reproduction

  - Occurs in bacteria, most simple plants and animals and some complex plants.
  - Does not involve gametes
  - New organisms arise by mitosis and therefore are exact copies of the parent.
  - Asexual reproduction includes:
    Binary fission – spitting into two
    Budding – new individuals arise off the parent
    Fragmentation – splitting of a parent into two or more organisms.

- Sexual reproduction

  - Involves the fusion of two gametes produced in meiosis.
  - Each gamete has N number of chromosomes (half the genetic material of the new organism)
  - In most cases gametes come from a male and a female.

3. VARIATION

MUTATIONS

- Any change in the arrangement or amount of DNA in a cell or virus.
- Can occur in somatic cells or in germ line cells.
- Only germ line cell mutations can be passed on.
- Mutagens can cause mutations. These include X radiation, UV radiation and many chemicals.
- If the sequence of bases in DNA is altered, the likely outcome is that the new code will result in the production of an altered protein, which may have a different biological function to the one coded for originally.
- Types of mutation:
  (a) Deletion
  If one base is deleted, all the codons from that point to the end of the gene will undergo a "frame shift". Most (or all) of the amino acids coded for will be different.
  (b) Addition
  If one base is added, the result is similar to a deletion.
  (c) Substitution
  If one base is replaced, only one amino acid may be affected (unless this occurs in a "start" or "stop" codon.
  (d) Chromosomal abnormality
  Part, or a whole chromosome may be altered with a consequent alteration to many different genes.
  Most mutations result in the organism being affected adversely, although occasionally the new protein confers an advantage to the organism.

GENOTYPE

- Refers to the particular combination of alleles at one or more gene loci of an organism.
- For genes located on autosomes, an organism has two copies of each gene.
- If the alleles are identical the organism is homozygous e.g. AA, aa, TT
- If the alleles are different the organism is heterozygous e.g. Aa. Tt.

PHENOTYPE

- Refers to the expression of the genotype in the physical, biochemical and physiological characteristics of an organism
- Environmental factors can interact with the genotype to produce different phenotypes under different conditions

INHERITED VARIATIONS

- Variations may be due to the action of a single gene, called **monogenic** traits.
  - Typically have two or three phenotypes e.g. attached ear lobes or not.
- May be due to the action of many genes and called p**olygenic** traits
  - Typically have a continuous range of phenotypes e.g. human height
- Changes in the chromosome number in an organism

4. PATTERNS OF INHERITANCE

- We inherit our characteristic from our parents i.e. the genes that code for all our traits come from our parents. These genes are part of the chromosomes that we get from each parent i.e. we get 23 from mother and 23 from our father.
- Early research such as Mendel's concentrated on single gene traits. Thus, the trait was controlled by one gene and there were two alleles (forms of one gee) carried on the organism's chromosomes.
- The genotype is the alleles present for a particular characteristic.
- The phenotype is the expression of the genes.
- Terminology used in describing patterns of inheritance includes:

Homozygous - having two identical alleles for a trait (e.g. AA)

Heterozygous - having two different alleles for a trait (e.g. Aa)

Dominant - expressed in the heterozygote

Recessive - expressed only in the homozygote

Incomplete dominance - the heterozygote has an intermediate phenotype

Co-dominance – when both alleles are expressed in a heterozygote.

Multiple alleles - more than two forms of a gene exist. The ABO blood system is an example with three alleles.

Lethal allele - a homozygote doesn't live to a reproduction age.

Linkage - two or more genes on the same chromosomes

Sex linkage - Inheritance of genes occurring on a sex chromosome. With X- linked recessive traits.

All males with an affected allele show the trait but only homozygous females show the trait.

Autosomes - chromosomes not involved in sex determination

P - parental generation

F1 - first generation

F2 - second generation of offspring

MONOHYBRID CROSS

- A cross between two organisms involving the alleles at a single gene locus.
- If the genotypes of parents are known, it is possible to predict the genotypes of offspring and the expected ratios.
- Can be shown on a Punnet square or by the Algebraic method.
- Example: Aa X Aa

	½ A	½ a
½ A	¼ AA	¼ Aa
½ a	1/4Aa	¼ aa

1/4AA , ½ Aa and ¼ aa

DIHYBRID CROSS

- - A cross involving alleles at two gene loci between two organisms
- If the genes are on different chromosomes they are said to be unlinked.
- Example two organisms heterozygous for two particular traits AaBb x AaBb usually a 9:3:3:1 ratio of offspring phenotypes.

TEST CROSS:

- Also called a back cross

PEDIGREES

- These are a schematic way of representing inheritance patterns in a family
- Males are represented by squares, females by circles
- Individuals showing a trait have the square/circle shaded.
- They can be used to determine if a trait is dominant or recessive.
- They can be used to determine if a trait is autosomal or sex-linked
- Pedigrees can be used to determine if at trait is
  - Autosomal Dominant
  - Autosomal Recessive
  - X- linked Dominant
  - X- linked Recessive

5. GENE TECHNOLOGY

- This involves the manipulation and use of DNA in somatic (non- reproductive) or germ line (gamete forming) cells. Somatic modifications are not passed on in sexual reproduction. Enzymes are important in all gene technologies.
- The roles of enzymes include:

Enzyme	Role in Gene Technology
Reverse transcriptase	Used to synthesize single stranded DNA from mRNA. Used to make copies of genes because mRNA isolated from cells already has its non-coding introns removed.
Restriction Enzymes	These are used to cut DNA at a particular sequence. Each specific restriction enzyme cuts DNA at a particular restriction site. Isolated from bacteria. Useful to isolate genes for copying, preparing DNA for sorting in gel electrophoresis and creating sticky ends on target and host DNA.
DNA ligases	Can be used for the insertion of cut DNA. Will join DNA fragments with sticky ends.
DNA polymerase **Taq polymerase**	Used to form double stranded DNA from single stranded DNA Used in PCR to join individual nucleotides to produce new DNA strands. Used in DNA sequencing

TOOLS AND TECHNIQUES OF GENE TECHNOLOGY

PCR (Polymerase Chain Reaction)

- This technique is a method of producing many copies of the same piece of DNA. Uses the enzyme DNA polymerase.
- PCR is used for:
  Amplification of small amounts of fossil DNA
  Making copies of genes for insertion into other cells
  Amplification of samples for forensic testing
  Making copies of artificially mutated DNA for further study.

MAKING A COPY OF DNA

- mRNA can act as a template for the formation of a single stranded DNA molecule called cDNA.
- The sequence of bases in cDNA is complementary to the mRNA
- Reverse transcriptase is the enzyme used to catalyse the joining of nucleotide subunits to form the cDNA
- DNA polymerase is the enzyme used to make a double stranded DNA molecule.

ELECTROPHORESIS

- This is a technique for sorting DNA fragments of different lengths.
- A mixture of fragments of DNA is loaded in a well in a gel
- An electric current is passed through the gel (Negatively charged fragments move to the positive end)
- The shorter the fragment of DNA the quicker it moves through the gel.
- Can be used for example to diagnose a number of genetic disorders when a sample of DNA is available

DNA PROFILING

- We differ in the alleles we carry for a particular gene but we also differ in the base sequences of DNA between genes. There are often sequences of non-coding bases repeated many times between genes.
- These regions of base repeats vary from person to person. (Often called STRs, short tandem repeats). These repeated sequences are inherited like other alleles.
- The repeated sequences revealed by radioactive or fluorescent probes is called **genetic profiling** or **genetic fingerprinting.**
- Genetic profiles can be used to distinguish individuals.
- This process can be used to:
  Establish parentage – used where the identity of the father is disputed.
  Establish other family relationships.
  Analyse biological evidence from a crime scene such as blood or semen and compare this to suspects.
  Establish the level of genetic variation in threatened species of animals or in zoo breeding programs.
  Establish evolutionary relationships between groups of organisms

DNA SEQUENCING

- Is the identification of the exact orders of nucleotide bases in a DNA molecule.
- The sequencing of the human genome has now been completed. This has been possible due to the development of sequencing machines in which the process is automated.
- The process involves:
  The use of different coloured fluorescent dyes each binding to a specific base.
  Using a single stranded copy of DNA to be sequenced for a template
  Making complementary copies of the template.
  Processing the material by computer and producing a print out of the nucleotide sequence.

VECTORS

- Vectors are special carriers that can transport DNA fragments into cells
- These can be bacteria, viruses or other small particles that can invade the tissue of target organisms.
- A plasmid is one type of vector. It is a small circular molecule of DNA.
- Many bacteria cells contain plasmids
- Insertion of a foreign gene into a plasmid and into a bacterial cell can lead to **gene cloning** as the bacteria divides.

GENE PROBES

- This is a small piece of single stranded DNA or RNA with a label so that it can be located.
- The label can be either a radioactive or fluorescent marker.
- A gene probe can locate a particular piece of DNA from among many small fragments of DNA.
- The base sequence of a gene probe must be complementary to a portion of one of the strands of the DNA target

RECOMBINANT DNA TECHNOLOGY

- Often called genetic engineering
- Involves the removal of DNA from one organism to be placed in another of the same or different species.
- Organisms with the foreign gene are called **transgenic organisms.**
- Involves the use of **ligases** to join pieces of DNA.
- Steps involved:
  DNA is cut from donor chromosomes using **restriction enzymes** or DNA is produced from mRNA using r**everse transcriptase**
  The DNA is copied by PCR.
  DNA is added to a **vector** that invades the tissue of the target organism. DNA is often added to bacterial plasmids and these plasmids are inserted into bacterial cells.
  DNA can also be directly injected into eggs, zygotes or embryonic tissue or plant tissue culture.
- Applications include:
  a) Insertion of genes into micro organisms to allow them to produce useful proteins eg bacteria producing insulin
  b) Insertion of genes into crop plants to gain favourable characteristics eg disease resistance, herbicide resistance, firmer fruit
  c) Insertion of genes into animals to allow them to express some desired characteristic.
  d) Insertion of a gene into individuals who have a genetic disorder in order to allow the production of a missing or faulty protein.

VICTORIA UNIVERSITY 2011

CHANGE OF PREFERENCE 16-21 DECEMBER

DROP INTO VUHQ - CITY FLINDERS CAMPUS, ONLINE CHAT SERVICE & COURSE HOTLINE: 1300 VIC UNI

Friday 16	8:30AM - 5:30PM
Saturday 17	11:00AM - 4:00PM
Sunday 18	11:00AM - 4:00PM
Monday 19	8:30AM - 5:30PM
Tuesday 20	8:30AM - 5:30PM

GENERAL INFORMATION SESSIONS

Monday 19	4:00PM - 7:00PM
Tuesday 20	4:00PM - 7:00PM

Footscray Park Campus
Ballarat Road, Footscray
Melways Ref. Map 42 C2

CAMPUS TOURS

Footscray Park tours will operate after each information session.

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