Understanding Mutations

Teacher Information Booklet

Contents

HARRY PERKINS INSTITUTE OF MEDICAL RESEARCH

education@perkins.org.au (08) 6151 0811

Lotterywest BioDiscovery Centre

About Us

About the Perkins

The Harry Perkins Institute of Medical Research, often referred to as the Perkins, is a leading Western Australian medical research centre, dedicated to tackling some of the world's biggest health issues.

With over 250 researchers located in three hospital campuses, our world-class teams accelerate the delivery of life-saving breakthroughs to improve the health of all Western Australians, today and for our future generations.

About the Lotterywest BioDiscovery Centre

The Lotterywest BioDiscovery Centre connects students, teachers, and members of the community to the world of medical science and the research happening at the Perkins. Through school visits, community tours, and teacher professional learning opportunities, we are invested in raising awareness of the importance of medical research throughout the Western Australian community.

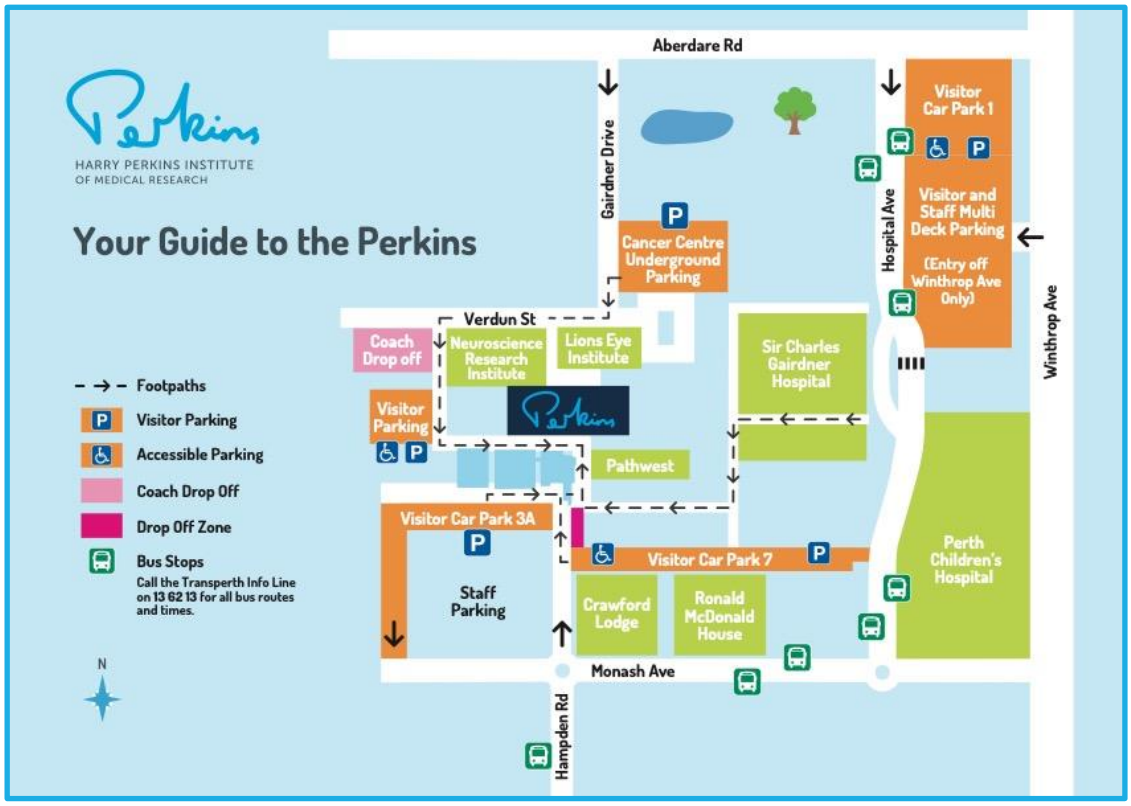
Where to find us

We are in **Nedlands** at 6 Verdun Street, 6009. See the building labelled "Perkins" on the map below.

Bus parking

Bus parking is organised through QEII parking. Should you need to park your school bus for the day please email Qeii.parking@health.wa.gov.au

Or call them on (08) 6457 7248.



About Your Session – Understanding Mutations

Understanding Mutations

Genetic mutations are the driving force for evolution through all domains of life. Favourable mutations bring new characteristics into a population and improve potential for survival of the population in the struggle for life. Deleterious mutations can render organisms at disadvantage, ranging anywhere from reproductive challenges, or in worst case scenarios, life altering disease. At the Perkins, researchers are dedicated to understanding these deleterious mutations in human populations. Building an understanding of how these genetic mutations persist within a population, how they present themselves in patients and how they pass through successive generations.

The rare genetic diseases team at the Perkins, headed by Professor Nigel Laing AO and Associate Professor Gina Ravenscroft, have a particular interest on neurogenetic diseases, which includes diseases of the nerves and muscles, such as muscular dystrophies. Their work has led to the discovery of over 30 new disease genes, providing answers to families all around the world who, until now, could not have known the nature of the disease plaguing their families. This has important ramifications in family planning, diagnostics, and treatments; but also, in building the tools and resources to continue learning about the plethora of disease-capable genes yet undiscovered in the global human population.

One of the rare diseases studied by the neurogenetic diseases lab at the Perkins is **Myoglobinopathy** – a muscle disease effecting just six European families. Understanding the mechanisms and genetics of this disease will prove life-changing for these families, though given its rarity, research into the disease is limited. Myoglobinopathy is caused by a point mutation in the myoglobin gene.

Visit our website for more information about the rare genetic diseases team at the Perkins.

What this session covers:

Through interactive activities, your class will gain a deeper understanding of the causes of mutations, and what effect these mutations have on the survivability of an organism. Students conduct a hands-on 'simulation' activity, where different mutations are spread through a population, and students will discover how these mutations can persist in a population, and how natural selection and random genetic drift can remove mutations and alleles from a population.

In our purpose-built teaching lab, students will be challenged to use DNA technologies, such as PCR and Gel Electrophoresis, to understand which family members carry this gene mutation and predict the likelihood of the next generation to be impacted by the disease. Students will learn first-hand how advances in research conducted in Western Australia are directly impacting the lives of families around the world.

Understanding Mutations in Research

By having a firm understanding of how mutations are caused, and their effect on cell biology and human physiology, students are better equipped to tackle many of the modern challenges in research, such as:

- Precision medicine in developing personalised treatments for cancer
- Tracing movement of pandemic diseases, such as HIV and COVID-19
- Identifying biomarkers for diseases such as cystic fibrosis
- Contributing to ongoing development of gene therapies using gene technologies
- Tracking and predicting sizes of animal populations during conservation efforts.

Curriculum links		
Biology Year 12 Unit 3 - Continuity of species **Human Biology** Year 12 Unit 4 - Human Variation and Evolution		
SU Statements:	**SHE Statements:**	**SIS Statements**
Biology: Natural selection occurs when selection pressures in the environment confer a selective advantage on a specific phenotype to enhance its survival and reproduction; this results in changes in allele frequency in the gene pool of a population **Human Biology:** Mutations are the ultimate source of variation introducing new alleles into a population: new alleles may be favourable or unfavourable to survival	**Biology:** Technological developments in the fields of comparative genomics, comparative biochemistry and bioinformatics have enabled identification of further evidence for evolutionary relationships **Human Biology:** Developments in biotechnology, for example polymerase chain reaction (PCR), gel electrophoresis and DNA sequencing, have increased access to genetic information of species and provide evidence for evolution (the process of obtaining a DNA sequence is not required).	Conduct investigations safely, competently and methodically for valid and reliable collection of data

Booking Information

Price

Visits are priced at $50 per student for a group of 20 or more students. For a group of less than 20 students per day, a minimum fee of $1000 applies. If this is an issue for you please contact us to discuss how we may be able to assist by linking you in with another school.

For bookings over multiple days, each day will be priced independently.

Once we receive your online booking form, you will be invoiced for our $100 per session booking fee to confirm your place. This will later be deducted from your final invoice.

Numbers

At the time of making your booking, please provide an estimate of the number of students that will be attending. You will be emailed a reminder two weeks before your visit to confirm student numbers.

It is the responsibility of the organising teacher to confirm final numbers at least one week prior to their booking. A decrease in the number of participants will not be accepted after this point and schools will be invoiced accordingly.

Photo Consent Policy

The Harry Perkins Institute of Medical Research may use photos taken during the session for publicity purposes unless otherwise agreed upon. The onus is on the school to obtain permission for students to be photographed.

Timing

Sessions run from 9:30 to 2:30. Alternative times must be arranged ahead of the session.

Cancellation Policy

The Lotterywest BioDiscovery Centre at the Harry Perkins Institute for Medical Research requests one month's notice for the cancellation of a school's booking. If this notice is not given, the school will be charged at 50% of the original cost of the excursion or session. The $100 booking fee is not refundable. It is the responsibility of the organising teacher to confirm final numbers one week prior to their booking.

Shared Classes

If the total number of students in a shared class drops below 20, all schools attending that day will split the $1000 minimum fee proportionally, based on the number students attending.

Duty of Care

School staff are to always remain with students. External providers do not have the same duty of care relationship with students and are not responsible for personally caring for students. Should there be any attendee with a medical condition, disability, mobility issue or special learning requirements, the school is required to attend with two adults who can accept responsibility for duty of care.

On the Day

What your students need to bring

Students will need to bring their own food and drink. Pens and workbooks are provided.

NB: Access to the café is not possible for students or staff during their visit.

PC2 Rules

All teachers and students must abide by PC2 (Physical Containment level 2) rules to be able to enter the lab. This means students and staff must wear fully enclosed shoes, and all hair/fringes tied back and off the face. Phones, bags, food or drink (including water) are not permitted in the PC2 lab area.

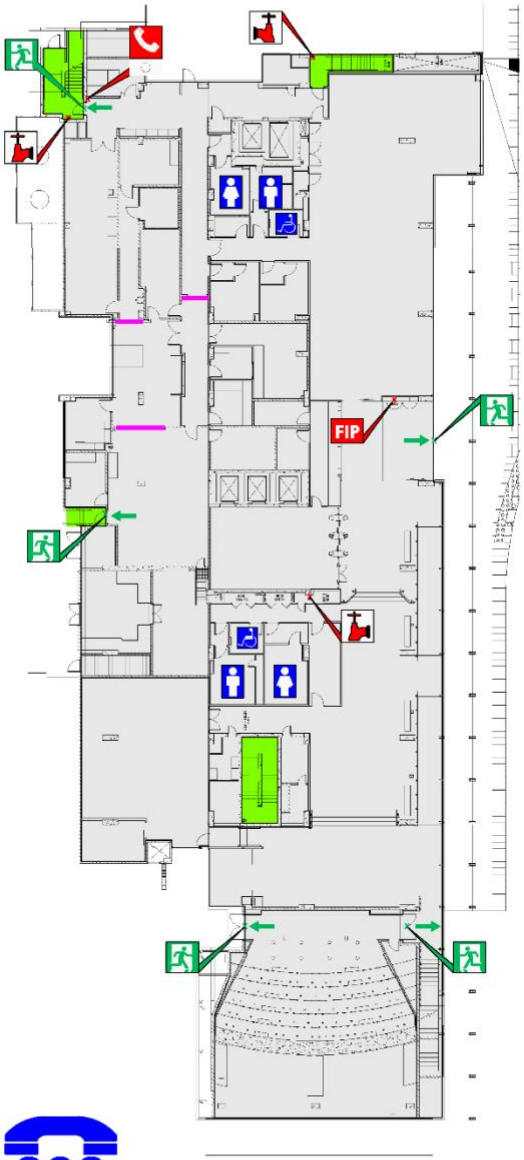
Working With Children Check Confirmation

It is a condition of employment at the Lotterywest BioDiscovery Centre that all staff have a current *Working With Children Card*.

Judi Lane
Community Education Manager
Lotterywest BioDiscovery Centre
Harry Perkins Institute of Medical Research

Evacuation Map: Ground Floor

EMERGENCY EVACUATION DIAGRAM
QE II PERKINS INSTITUTE
GROUND



1. **When the ALERT ALARM(Beep, Beep, Beep) sounds,** Stop what you are doing and prepare for evacuation. Listen for instructions.Follow directions from your warden.
2. **When the EVACUATE tone (Whoop, Whoop, Whoop) Sound,** under the instruction of the Wardens evacuate the building via the Emergency Exit. If no wardens are present, evacuate the building immediately via your nearest exit.
3. Do not panic. Move quietly and calmly to the primary **Assembly Area** on the **footpath in Verdun Street.**
4. Do not return to the building until given the"All Clear" by the Chief Warden or Emergency Services.

LEGEND

FIP	Fire Indicator Panel
	W.I.P
	Fire Hydrant
	Emergency Alarm Button
	Emergency exit
	Assembly Area
	Fire isolation roller doors

000 EMERGENCY

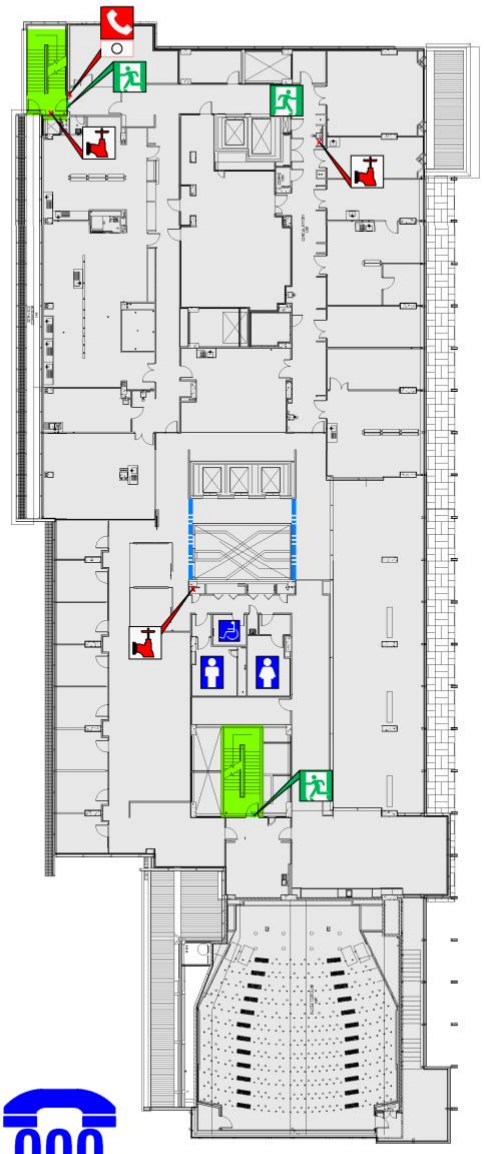
ISSUE DATE NOVEMBER 2013
VALID DATE: NOVEMBER 2022

Issued in compliance With AS3745-2010

IN THE EVENT OF A FIRE RING 000 TO ENSURE FIRE SERVICE RESPONSE

Evacuation Map: Level 1

EMERGENCY EVACUATION DIAGRAM
QE II PERKINS INSTITUTE
LEVEL 1



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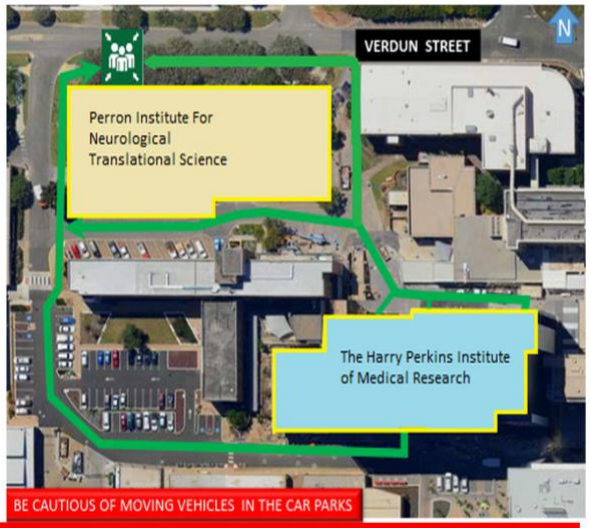
LEGEND

- W.I.P
- Fire Hydrant
- Emergency Alarm Button
- Emergency exit
- Assembly Area
- Fire Curtin



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