HARRY PERKINS INSTITUTE OF MEDICAL RESEARCH

Lotterywest BioDiscovery Centre

Year 12: Understanding Mutations using PCR

Teacher Information Booklet

Contents

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About the Perkins

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The Harry Perkins Institute of Medical Research, commonly known as The Perkins, is the largest medical research institute in Western Australia working on diseases such as cancer, diabetes, heart disease, and rare genetic diseases.

With over 250 researchers located in three hospital campuses, the Perkins is uniquely positioned to fast track the development of new discoveries and treatments. Researchers at the Perkins are driven to find new ways of diagnosing and treating diseases.

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 events, and educator resources, we are dedicated to ensuring all Western Australians become more active participants in medical research.

Where to find us

We are located in Nedlands at **6 Verdun St, Nedlands 6009**. See the map below.

Bus parking

Bus parking is organized through QEII parking. Should you require to park your school bus for the day please contact:

- Nicolette.Staal@health.wa.gov.au,
- Qeii.parking@health.wa.gov.au

Or alternatively, call on: (08) 6457 7248

Parking Map


Your Guide to the Perkins
HARRY PERKINS INSTITUTE OF MEDICAL RESEARCH
Footpaths leading to the Perkins
Visitor Parking
Accessible Parking
Drop Off Zone
Bus Stops
Aberdare Rd
N
Gairdner Drive
Cancer Centre Underground Parking
Visitor Car Park 1
Visitor and Staff Multi Deck Parking
(Entry off Winthrop Ave Only)
Hospital Ave
Winthrop Ave
Verdun St
Parking
Neuroscience Research Institute
Lions Eye Institute
Visitor Parking
Main Entrance
Sir Charles Gairdner Hospital
Drop off Zone
Pathwest
Visitor Car Park 3A
Staff Parking
Caledenia Cres
Staff Parking
Crawford Lodge
Ronald McDonald House
Perth Children's Hospital
Monash Ave
Hampden Rd

About your Session

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 in 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 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.

What this session covers:

Students will be introduced to a case-study of one of the major challenges Perkins researchers have in understanding the origins and inheritance of genetic diseases. Looking at a population experiencing a genetic disease, and a family with unknown inheritance, students will be challenged to develop an understanding of mutations and identify them using Pedigrees; and the laboratory techniques used to investigate

them. Students will experience a hands-on activity to build an understanding of polymerase chain reaction before conducting their own in our laboratory.

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 the Myoglobinopathy gene mutation, predict whether the mutation was inherited the patients' parents or occurred through random chance, and determine the likelihood for the next generation to be impacted by the disease. Students will learn first-hand how advances in research conducted in Western Australia is directly relevant to the lives and wellbeing of families around the world.

Curriculum Links

Year 12 Biology Curriculum Unit 3: Continuity of Species		
Science Understanding:	**Science as a Human Endeavour:**	**Science Inquiry Skills**
Pedigree charts can be used to reveal patterns of inheritance and assist in determining the probability of inheriting particular alleles in future generations DNA sequencing enables mapping of species genomes; DNA profiling identifies the unique genetic makeup of individuals; processes such as PCR (to amplify minute samples of DNA to testable amounts) and gel electrophoresis can be used to facilitate DNA sequencing of genomes	Technological developments in the fields of comparative genomics, comparative biochemistry and bioinformatics have enabled identification of further evidence for evolutionary relationships	Conduct investigations safely, competently and methodically for the collection of valid and reliable data.

Year 12 Human Biology Curriculum Unit 4: Human Variation and Evolution		
Science Understanding:	**Science as a Human Endeavour:**	**Science Inquiry Skills**
Mutations are the ultimate source of variation introducing new alleles into a population: new alleles may be favourable or unfavourable to survival	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 the collection of valid and reliable data.

Terms and Conditions of Booking

Price

Visits are priced at $50 per student for a group of 20 or more students. For a group of less than 20 students, a minimum fee of $1,000 applies. Feel free to contact the education team to organise school sharing opportunities. Cost will be shared proportionately if multiple schools attend a singular day.

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

Once your booking has been confirmed, a $100 per session booking fee will be issued within 10 business days. For bookings in the next calendar year, this booking fee will be issued in the new year, unless requested otherwise.

Numbers

At the time of making your booking, 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 school can opt-out of this arrangement through the booking confirmation form, or through written notice to the education team at any time before or after the excursion.

Timing

Sessions run from 9:30 am – 2:30 pm. 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.

Cancellations will not be accepted with less than one week's notice.

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 to the number students attending.

Duty of Care

School staff are to always remain with students. Staff at the Harry Perkins of Medical Research do not hold duty of care over school 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 must bring their own food and drink. Pens and workbooks are provided.

Note: On-site cafés are not accessible for students or staff during their visit.

PC2 Compliance

All teachers and students must comply by expectations for our Physical Containment Level 2 (PC2) as set by the Institutional Biosafety Committee (C-IBC). This means students and staff must wear fully enclosed shoes, and all hair/fringes tied back and off 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 demonstrator staff have a current Working With Children Card.