Timetable

All learning activities are in person unless otherwise indicated and attendance in compulsory

Lecture slides will be uploaded to the Amathuba site after the lecture (lectures will not be recorded)

A medical certificate will be required in the event of absence from assessments

* lecture content will be assessed via the Genetics, Individual Genomes and Population Genomes module assessments administered via Amathuba

♦ marks will be awarded for participation in learning activity

learning activity will be assessed via a written assignment/assessment

Genetics
Neurogenomics Lab
Psychiatry

Genetics

Week 1 (28 July - 1 Aug)

Monday 28 July	Teaching	Venue
11:00-11:45	**From Cells to Genes: The Foundations of Molecular Biology* (Dr Alina Esterhuizen)** focus: mitosis, meiosis and the central dogma *An overview of the essential processes of mitosis and meiosis, explaining how cells divide and generate genetic diversity. Introduction of the central dogma (DNA-RNA-protein) to lay the groundwork for understanding gene function and expression.*	Resource room 1 G floor, NI
11:45-12:45	**Caffeinated Conversations** *Curious about where a research career could take you? Join us for an informal coffee networking event with postgraduate students and researchers to chat about their journeys into academia and science.*	NI atrium
14:00-14:45	**How Genes Inherit and Misbehave: Patterns of Mutation and Inheritance* (Dr Alina Esterhuizen)** focus: inheritance patterns and mutational mechanisms *An overview of Mendelian and non-Mendelian inheritance patterns, alongside the mutational mechanisms that disrupt gene function and lead to disease. Clinical examples linking genetic theory to real-world conditions.*	Resource room 1 G floor, NI
Friday 1 Aug	**Teaching**	**Venue**
11:00-11:45	**Tools of the Trade: Core Techniques in Genetics and Genomics* (Dr Alina Esterhuizen)** focus: introductory cyto- and molecular technologies *Introduction to key molecular biology tools: PCR, Sanger sequencing, NGS, and others, explaining the principles and detection scope of these methods, as the backbone of modern genomics.*	Aadil Moerat Room on the ground floor of the Barnard Fuller Building (formerly known as "Conference Room 3")
12:00-12:45	**From Bench to Bedside: Molecular Diagnostics in Clinical Practice* (Dr Alina Esterhuizen)** (focus: diagnostic application of molecular technologies) *An overview of the application of molecular technologies in diagnostic practice versus research.*	Aadil Moerat Room on the ground floor of the Barnard Fuller Building (formerly

	Examples illustrating how molecular technologies are used to detect pathogenic variants and guide clinical decision-making (including precision medicine).	known as "Conference Room 3")
14:00-15:00	**Wet Lab Tour (Dr Alina Esterhuizen)**	NHLS/Division of Human Genetics

Individual genomes (part 1)

Week 2 (4 Aug - 8 Aug)

Monday 4 Aug	Teaching	Venue
11:00-11:30	**Practical Onboarding (Neurogenomics Lab student trainers)** Hands on support session for students needing assistance with software installation, website registrations, data and database access [see tutorial]	Room 32, F floor, NI
11:45-12:30	**What is Bioinformatics? From Tools to Pipelines* (Dr Mohammed Farahat)** *Introduction to the field of bioinformatics- the intersection of biology, computer science, and data analysis. We will explore how bioinformatics enables the interpretation of complex biological data, particularly genomic sequences. Starting with user-friendly, web-based tools, students will learn how biological questions can be addressed through computational approaches. The session will then build toward more advanced concepts, including writing and running code, executing analysis pipelines, and working with reproducible workflows. By the end of the lecture, students will have a practical sense of the spectrum of bioinformatics—from point-and-click applications to command-line-driven analysis—and how these tools support discovery in modern genomics.*	Resource room 1 G floor, NI [facilitated viewing of online lecture]
14:00-14:30	**Neurogenomics in the Clinic: Career Reflections* (Prof J Heckmann)** *A personal perspective on the evolution of neurogenomics and its growing impact on clinical care contrasting both phenotype-led (starting with the clinical picture) and genotype-led (starting with the DNA) approaches.*	IIDMM

14:45-15:45	**Bioinformatics Crash Course Part 1: Alignment of NGS reads to a reference genome* (Dr Melissa Nel, Mr Gideon Wiafe, Mr Saifeldeen Elshahawy)** *This practical introduces the first key step in next-generation sequencing (NGS) data analysis: aligning short reads to a reference genome. We'll explore why alignment matters, how it works, and what tools are commonly used, laying the foundation for downstream genomic analysis.*	Resource room 1 G floor, NI
Friday 8 Aug	**Teaching**	**Venue**
11:00-13:00	**Ilifu facility visit* (Mr Dane Kennedy, Mr Mike Currin)** *An opportunity to explore the infrastructure and capabilities of a High Performance Computing (HPC) facility. Students will gain insight into how HPC supports advanced research across disciplines, with a particular focus on its role in genomic data analysis. The visit includes an orientation to the architecture of the HPC system, and discussions with technical staff on resource allocation, data storage, security, and job scheduling. By engaging directly with the facility, students will better understand how computational power is harnessed to process and interpret large-scale biological datasets, and how HPC integrates into the broader research ecosystem.*	ICTS on Main, 7 Main Road, Mowbray

Individual genomes (part 2)

Week 3 (11 Aug - 15 Aug)

Monday 11 Aug	**Teaching**	**Venue**
11:00-11:30	**From SNPs to Structural Variants: Understanding Genetic Variation* (Ms Michaela O'Hare)** *An overview of the different types of genetic variants—from single nucleotide polymorphisms (SNPs) to small insertions/deletions (indels) and large structural variants.*	Resource room 1 G floor, NI
11:30-12:15	**Bioinformatics Crash Course Part 2: Calling variants from aligned NGS reads* (Dr Melissa Nel, Ms Michaela O'Hare)** *This practical covers the next step in NGS data analysis: identifying genetic variants from aligned sequencing reads. We'll introduce key concepts in variant calling, explore commonly used tools, and discuss how raw data is transformed into interpretable genetic variation.*	Resource room 1 G floor, NI
13:15-14:00	**From Variants to Meaning – Annotating and Interpreting Genomic Data# (Dr Melissa Nel)** *Variant annotation involves linking raw genomic variants to known biological information- such as gene context, predicted functional impact, population frequency, and disease associations. Students*	Resource room 1 G floor, NI

	will be introduced to commonly used annotation tools and databases (e.g. VEP, ClinVar, gnomAD, OMIM, ClinGen)	
14:00-14:45	**Bioinformatics Crash Course Part 3: Manual Annotation of Genomic Variants[#] (Dr Melissa Nel, Mr Gideon Wiafe, Mr Saifeldeen Elshahawy, Ms Zama Mngadi, Ms Erin Kinghorn)** *In this hands-on session, students will practice the manual annotation and interpretation of genomic variants identified through NGS analysis. Working with a curated set of variants from a real patient case, students will use online databases and tools to investigate each variant's potential clinical significance. This will include:* ***Gene context and variant type** (e.g. missense, nonsense, splice-site)* ***Population frequency*** (e.g. gnomAD, 1000 Genomes) **Predicted functional impact** (e.g. SIFT, PolyPhen, CADD scores) ***Known disease associations** (e.g. ClinVar, OMIM)* ***Conservation and inheritance pattern*** *Each student will be paired with a trainer who will guide them through the process of interpreting and prioritizing variants based on these data. This manual approach provides a foundational understanding of how the pathogenicity of variants is assessed, before moving to automated annotation tools or pipelines.*	Resource room 1 G floor, NI
15:00-15:30	**Reflective Session[#] (Dr Melissa Nel)** *Through guided prompts and group discussion, students will evaluate their confidence and preparedness to apply bioinformatics skills—such as variant calling, annotation, and interpretation—to real-world genomic cases. The session will encourage critical thinking about the challenges of translating technical results into meaningful clinical or research insights.*	Resource room 1 G floor, NI
Friday 15 Aug	**Teaching**	**Venue**
10:00-11:00	**Genetics assessment**	Resource room 1 G floor, NI
11:00-14:00	**Workshop: Introduction to the Multidisciplinary Team Approach to Genomic Medicine♦ (Dr Melissa Nel on behalf of the NMDAfrika Initiative) [15min]**	Room 34, F floor, NI

	Rare Disease Case Presentations♦ (Dr T Europa) [15min] *In this session, we'll explore real-world rare disease cases to understand how clinical features and family history guide the diagnostic journey from bedside to genome.* **Genetic Pre-Test Counselling in Practice♦ (Ms Nabeelah Peerbhai) [60min]** *This interactive session uses role-play to simulate genetic pre-test counselling conversations. Students will take turns acting as clinicians and patients, gaining practical experience in communicating complex genomic concepts, setting expectations, and exploring ethical considerations before genetic testing.* **BREAK [30min]** **Describing Disease: An Introduction to Human Phenotype Ontology (HPO) * (Dr Sergio Alves) [20min]** *This talk introduces the Human Phenotype Ontology (HPO), a standardized vocabulary for describing clinical features of genetic conditions. We'll explore how HPO terms improve phenotype-driven analysis, support rare disease diagnosis, and enable more precise links between patients and genes.* **Franklin Practical: From Clinical Notes to Genomic Insights♦ (Dr Tarin Europa/Dr Sergio Alves/Dr Melissa Nel) [40min]** *In this interactive session, students will use the Franklin platform to enter clinical information and annotate cases with Human Phenotype Ontology (HPO) terms. Through hands-on analysis, they'll explore how structured phenotypic data supports variant prioritization and rare disease diagnosis using AI.*	

Individual genomes (part 3)

Week 4 (18 Aug - 22 Aug)

Monday 18 Aug	Teaching	Venue
11:00-11:45	**Evaluation and classification of genetic variants in a clinical setting and associated challenges* (Ms Nabeelah Peerbhai)** *This session explores how genetic variants are evaluated and classified in a clinical diagnostic context, using internationally recognized frameworks such as the ACMG/AMP and ClinGen guidelines. Students will be exposed to the systematic process of assessing variant pathogenicity—taking into account population frequency, computational predictions, segregation data, functional studies, and clinical correlations. The session will highlight the real-world challenges faced in clinical genomics, including limited data in underrepresented populations, variants of uncertain significance (VUS), and evolving evidence that may reclassify a variant over time. Through case examples, students will gain insight into how multidisciplinary teams (clinicians, genetic counsellors, bioinformaticians) collaborate to make evidence-based decisions that directly impact patient care.*	Resource room 1 G floor, NI
12:00-12:45	**HiFi Genome Sequencing: Accuracy Meets Long Reads* (Acclaim Moila)** *PacBio HiFi sequencing is transforming the field of genomics by combining long read lengths with exceptional base accuracy—unlocking access to the hardest-to-sequence parts of the genome. This lecture will showcase HiFi technology, what sets it apart from other long-read and short-read platforms, and how it enables better detection of structural variants, repeat expansions, and complex disease-associated loci.*	Resource room 1 G floor, NI
14:00-14:45	**Hands-On Practical: Comparing Exome, Short-Read Genome, and Long-Read Genome Data* (Dr Melissa Nel, Mr Gideon Wiafe, Mr Saifeldeen Elshahawy, Mr Waheed Amanjee)** *In this interactive session, students will explore real sequencing datasets to understand the strengths and limitations of different approaches to genome analysis. Exome sequencing, short-read whole genome sequencing (WGS), and long-read (HiFi) genome data will be compared in terms of coverage, variant detection, and resolution of complex regions.*	Resource room 1 G floor, NI
Friday 22 Aug	**Teaching**	**Venue**
09:30-10:15	**Reflective Session (Dr Melissa Nel) [45min]** *Students will reflect on their experience analysing real rare disease cases using the Franklin platform. From variant prioritisation to identifying pathogenic variants, this session invites students to share what challenged, surprised, or excited them during the process.*	Room 34, F floor, NI

10:30-11:30	**Bioinformatics crash course part 4: The trouble with repeats...* (Dr Melissa Nel, Ms Zama Mngadi)** *Repetitive DNA sequences are a major challenge in genome analysis—from confounding short-read alignments to hiding disease-causing variants in plain sight. In this session, we explore the biology of genomic repeats, the technical hurdles they pose for variant detection, and the specialized tools used to tackle them. Students will gain insight into how repeat expansions are interpreted in a clinical genomics context.*	
11:30-12:30	**Genetic Post-Test Counselling in Practice# (Ms Nabeelah Peerbhai) [60 min]** This interactive session uses role-play to simulate genetic post-test counselling conversations. Students will take turns acting as clinicians and patients as they navigate the delivery of genomic test results and explore the ethical and psychosocial challenges that arise.	

Population genomes (part 1)

Week 5 (25 Aug - 29 Aug)

Monday 25 Aug	Teaching	Venue
11:00-11:45	**Understanding Complex Disorders I: Genetic Architecture, Heritability, and Polygenicity (Dr Megan Campbell)** focus: foundational concepts in complex trait genetics, an introduction to the genetic principles underlying common complex disorders. *This lecture explores how complex disorders differ from rare, Mendelian diseases by focusing on polygenicity, gene-environment interactions, and the nature of common variant contributions. Concepts of SNP-based heritability, how it differs to twin and family based estimates and its estimation will be introduced to frame the genetic architecture of traits like psychiatric and substance use disorders.*	Resource room 1 G floor, NI
12:00-12:45	**Understanding Complex Disorders II: Genetic Overlap and the Search for Missing Heritability (Dr Megan Campbell)** focus: trait interconnectivity and interpretation of GWAS results	Resource room 1 G floor, NI

	This session deepens the discussion of complex genetic traits by exploring genetic correlation, pleiotropy, and the concept of missing heritability. Students will be introduced to the challenges of detecting genetic signal, the limitations of GWAS, and the role of cross-trait architecture. The lecture sets the stage for the upcoming session on genome-wide association studies, connecting theory to the practical sessions covered in the rest of the module.	
14:00-14:45	**Population Genetics in the Context of Complex Traits and Psychiatric Genomics (Dr Mary Mufford)** Focus: Understanding how population genetic principles inform the design, interpretation, and generalizability of genetic studies of complex traits, especially psychiatric disorders. *This lecture introduces key population genetic principles essential for understanding human genomic variation and its relevance to complex trait research. Students will explore how evolutionary forces, such as allele frequency shifts, linkage disequilibrium, genetic drift, migration, and population structure, shape genomic diversity. These dynamics present important considerations for genome-wide association studies (GWAS), heritability estimation, and the interpretation of genetic risk across diverse groups. The session will emphasize the critical importance of ancestry diversity in genetic studies, with a focus on situating African genomic data within global research efforts.*	Resource room 1 G floor, NI [facilitated viewing of online lecture]
15:00-15:45	**Genome-wide Association Studies (Dr Megan Campbell)** focus: methodology, interpretation, and post-GWAS analysis *An introduction to genome-wide association studies (GWAS), covering foundational principles including study design, population stratification, statistical thresholds, and signal detection. The session highlights how GWAS identifies genomic regions linked to complex traits and outlines key considerations when interpreting these associations. Students will also be introduced to post-GWAS strategies and the importance of integrating results with other omic data. This lecture provides the conceptual groundwork for the GWAS tutorials and downstream analyses explored later in the module.*	Resource room 1 G floor, NI
Friday 29 Aug	**Teaching**	**Venue**
09:30-10:30	**Individual Genomes Assessment**	Room 34, F floor, NI
11:00-13:00	**GWAS Tutorial[#] (A/Prof Shareefa Dalvie & Dr Megan Campbell)** focus: interpreting GWAS findings and visualizing association signals	Dumo Baqwa (it's the first venue on your

	This tutorial guides students through the interpretation of a published GWAS paper, focusing on how to critically read summary statistics, QQ plots, and Manhattan plots. Key concepts such as population stratification, multiple testing, and linkage disequilibrium will be discussed to help students make sense of real-world genetic association results.	right when you enter through the Barnard Fuller security desk)
[lecture must be viewed before the start of week 6]	**Polygenic risk scores (Dr Lerato Majara)** focus: interpreting genetic risk through polygenic frameworks. An introduction to the development, application, and limitations of polygenic risk scores (PRS) in complex trait prediction. This lecture builds on foundational concepts to explain how cumulative common variant effects are aggregated into PRS. It will explore methodological considerations, population-specific challenges, and interpretability in clinical and research settings. Examples from psychiatric and substance use traits will illustrate practical applications and current limitations in diverse populations.	pre-recorded lecture

Population genomes (part 2)

Week 6 (1 Sep - 5 Sep)

Monday 1 Sep	Teaching	Venue
11:00-11:45	**Phenotypes in Psychiatric Genetic Studies (Dr Kristien van der Walt)** focus: understanding how phenotype definition shapes genetic discovery. *This lecture highlights the critical role of phenotyping in psychiatric genomics, using the Schizophrenia in the Xhosa (SAX) study as a focal example. It explores how phenotyping strategies impact the validity of genetic findings. Discussions will include phenotypic factors such as diagnostic complexity, comorbidity, symptom overlap (including transdiagnostic approaches), disease severity and phenotype heterogeneity, with emphasis on the implications for study design, interpretation, and translatability across diverse populations.*	Resource room 1 G floor, NI
12:00-12:45	**Psychiatric Epigenomics and Transcriptomics (A/Prof Nastassja Koen)** focus: foundational concepts in psychiatric epigenomics and transcriptomics *This lecture will provide an introduction to epigenomics (with a focus on DNA methylation) and transcriptomic mechanisms (including microarray data and RNA-sequencing), in the context of*	Resource room 1 G floor, NI

	psychiatric -omics research. Select cross-generational research globally, as well as emerging data from South Africa, will also be presented.	
14:00-16:00	**Written assignment (A/Prof Shareefa Dalvie & Dr Megan Campbell)** *This assignment is directly linked to the GWAS tutorial session. A published GWAS paper will be circulated to students on the Friday before the assignment. In the assignment, you will interpret the paper's results using the same logic and methods demonstrated in the tutorial- working through results, interpreting figures, and considering factors such as population stratification, multiple testing, and linkage disequilibrium. You will then answer a set of structured questions based on the paper.*	Resource room 1 G floor, NI
Friday 5 Sep	**Teaching**	**Venue**
09:00-10:00	**Written assignment (A/Prof Shareefa Dalvie & Dr Megan Campbell)** *Students will be assessed on material covered in Introduction to Complex Traits, Population Genomics, and Genome-Wide Association Studies. The assessment will include a combination of multiple-choice and short-answer questions designed to test both conceptual understanding and practical interpretation skills.*	Room 34, F floor, NI
10:00-12:00	**Workshop: Ancestry, Admixture, and Population Descriptors in Genomic Research** **(Dr Yolandi Swart, Ms Erin Kinghorn)** *This workshop explores the complexities of ancestry and population structure in genomic research, with a focus on South African population groups. Students will engage with concepts such as global and local ancestry inference, admixture mapping, and the interpretation of diverse genetic backgrounds in the context of health and disease. Students will reflect on responsible and context-aware use of population* descriptors in research and reporting.	Room 34, F floor, NI

Population genomes (part 3)

Week 7 (15 Sep - 19 Sep)

Monday 15 Sep	**Teaching**	**Venue**
11:00-11:45	**What is a variant? A philosophical workshop and debate♦ (Dr Melissa Nel)** *We use the term variant every day in genomics—but what does it really mean? In this thought-provoking session, students will step beyond the technical definitions to interrogate the concept of*	Resource room 1 G floor, NI

	variation itself. Is a variant simply a deviation from the reference genome? Who defines what is "normal," and how do context, population, and technology shape our interpretation?	
12:00-12:45	**Human Pangenome Reference Graphs* (Dr Mohammed Farahat)** *The human reference genome has long been a foundational tool in genomics—but it represents only a small fraction of global human genetic diversity. In this session, we introduce the concept of pangenomes and explore how reference graphs are reshaping our approach to genome analysis. We'll also discuss the computational challenges and tools involved in building and using genome graphs, and examine their potential to reduce reference bias and improve equity in genomic research.*	Resource room 1 G floor, NI [facilitated viewing of online lecture]
Friday 19 Sep	**Teaching**	**Venue**
10:00-14:00	**Genomic Medicine in Practice (Dr Armin Deffur)** *Students will learn how genomic medicine and digital health tools are being applied in real-world psychiatric care. Through clinical case discussions, the session will highlight how precision approaches are transforming diagnosis, treatment, and patient outcomes in mental health.* **case presentations and discussion [2hrs]** **working lunch [1hr]** **assessment [30min]**	*Site visit to InDiGen Africa Precision Psychiatry Unit, Cure Day Hospital*

Genomes in Society

Week 8 (22 Sep - 26 Sep)

Monday 22 Sep	**Teaching**	**Venue**
11:00-11:45	**Genomic data accessibility and sharing: ethical considerations and challenges (Dr Melissa Nel)** *This lecture explores the balance between advancing science through open sharing of genomic data and safeguarding the rights and interests of research participants. It highlights ethical considerations such as privacy, consent, data ownership, and equity in access, while also examining the practical challenges of ensuring secure yet meaningful data use. The session will encourage critical reflection on how to responsibly maximize the benefits of genomic research while minimizing risks to individuals and communities.*	Resource room 1 G floor, NI

14:00-14:45	**A disability activist living with type 3 spinal muscular atrophy shares her story (Ms Thembelihle Ngcai)**	Resource room 1 G floor, NI [facilitated viewing of online lecture]
Friday 26 Sep	**Teaching**	**Venue**
09:30-10:30	**Population Genomes Assessment**	Room 34, F floor, NI
10:45-12:15	**Workshop: How the media shapes public perception of genomics (Ms Nabeelah Peerbhai)** *This interactive workshop examines how media narratives influence public understanding and attitudes toward genomics. Participants will analyze real-world examples of media coverage, identify biases or misconceptions, and reflect on how framing shapes trust, expectations, and ethical debates in genomic science.*	Room 34, F floor, NI