The Genomic Bioinformatics Workshop

28th September-1st October 2013

The RCPA, AACB and HGSA are proud to announce the dates for an inaugural four-day Genomic Bioinformatics workshop, which specifically targets the CPD requirements of genetic pathologists, medical genetic scientists, and trainees working towards gaining Fellowship qualifications.

The workshop is an intermediate level workshop with a practical focus, which has been developed to meet the vocational training needs of experienced genetic laboratory professionals requiring fluency in the practical applications of genomic bioinformatics tools. This workshop will be recognised with accreditation of CPD points for the RCPA.

The four day workshop will be structured around daily themes, with the first three days focusing on core competencies, as follows:

Day 1 – Ensembl Workshop – Advanced browser navigation, custom annotation and data-mining.

Day 2 – Pipeline biostatistics – pipeline validation, pipeline quality monitoring and management, massively parallel data interpretation and common errors.

Day 3 – Introduction to informatics – introduction to algorithms and computational methods in a set of classic bioinformatics problems: String matching, sequence alignment and assembly, and variant calling.

Day 4 – Genome data analysis, including a half-day Galaxy workshop demonstrating common analyses and operations on genomic data, building a basic workflow to perform variant analysis, and sharing, describing and publishing analyses and generated datasets.

The workshop's computing requirements limits participation to a maximum of 50 registrants.

Applicants will be expected to have the following assumed knowledge:

An advanced understanding of:

Mutational mechanisms of disease, inheritance patterns and genetic models, diagnostic laboratory practices, mutation/variant detection strategies, interpretation and reporting the clinical significance of genetic test data

A working understanding of:

Bayes' theorem, basic diagnostic statistics (e.g. Type I/Type II errors, positive/negative predictive values, sensitivity and specificity), familiarity with major genome browsers and online databases (NCBI, EMBL), evaluation of data quality

Applications will be accepted on a first-come, first-served basis with preference given to RCPA Fellows and Trainees for the first two weeks of enrolment. Given the number of placements is strictly limited, early enrolment is advised.

Registrations will open on the 19th June 2013, and will be found on the RCPA website: http://www.rcpa.edu.au/Continuing/CalendarOfEvents/September2013.htm