

Australia's National Science Agency

# **Bioinformatics Student Exchange Program**

Start your career with a research project at one of the world's premier research organizations.





### 1 BSEP 2024

The Bioinformatics Student Exchange Program (BSEP) is aimed at giving overseas students the opportunity to contribute to world-class research and gain experience in an international research environment. Master and Honours students are invited to conduct original research as part of their University Thesis. This is an exciting opportunity to forge new collaboration and build up a strong international network.

### Why Australia?

Australia is the "most productive of all G20 nations" with respect to papers published [<u>nature Index</u>] and a recent <u>Nature article</u> says that "Scientists from across the world are attracted to the country, which competes internationally by focusing on its strengths".

<u>The Commonwealth Scientific and Industrial Research Organisation (CSIRO)</u> is Australia's Government Research Agency and one of the largest and most diverse scientific organisations in the world. By igniting the creative spirit of our people, CSIRO deliver great science and innovative solutions that benefit industry, society and the environment. **COVID-19:** International travel has been opened back up in Nov 2021. However, with volatility in cases future restrictions might prevent the 2023-participants to conduct all or any part of their studies in Australia. Catering for this, we also offer BSEP remotely.

University	Contact Person
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CSIRO	A/Prof. Dr. Denis Bauer Transformational Bioinformatics, eHealth, CSIRO Phone: +61 2 9325 3174 Email: <u>denis.bauer@csiro.au</u>

#### Key dates

Date	
June	CSIRO calls for project proposals
31 <sup>st</sup> July	Program Booklet sent to the Universities
Early August to early November	Students choose proposals and get in contact with CSIRO
Early August to early November	Deadline for PROMOS or equivalent funding application
Dec	Thesis committee assesses suitability of projects and identifies appropriate co-supervisor amongst the <u>faculty</u> .
Jan	CSIRO starts recruitment process (visa)
May*	Students commence research in Australia
Oct*	Students return home
Nov*	Students finalise reports and write master thesis with input from CSIRO researchers

\* Times for visit can be flexible

#### How to apply

Please choose the project you are interested in and get in touch with your contact person listed above. Your first step will be to organize funding by applying (see below). After a successful interview, CSIRO will issue a contract with a visa sponsorship number in January. It is crucial to apply for the Australian Visa quickly as it can take up to 3 months to be approved. CSIRO will guide you through the process but please have a look at:

#### Funding

Students are encouraged to apply for funding. Unless stated otherwise, the projects will not provide funding.

#### PROMOS

German funding through <u>PROMOS</u> (Deadline Early October to early November), which will cover from 300 to 500 EUR per month or traveling costs up to 1950 EUR

Note, PROMOS is not explicitly paying a health insurance, this hence needs to be covered by the student.

### DAAD

The DAAD offers Study Scholarships, such as Master Studies for All Academic Disciplines <u>https://www2.daad.de/deutschland/stipendium/datenbank/en/21148-scholarship-</u> <u>database/?detail=50026200</u> with funding between 10 and 24 months.

There are also other funding sources available such as <u>http://www.ranke-heinemann.de</u>.

### Other resources

Please choose the project you are interested in and get in touch with your contact person listed above. Your first step will be to organize funding by applying for PROMOS or equivalent sources (DAAD). After a successful interview in January, CSIRO will issue a contract with a visa sponsorship number. It is crucial to apply for the Australian Visa quickly as it can take up to 3 months to be approved. CSIRO will guide you through the process but please have a look at:

VISA:

The visa subclass 402 doesn't exist anymore, for us the Temporary Activity (subclass 408) visa – Research Activities applies now: <u>https://immi.homeaffairs.gov.au/visas/getting-a-visa/visa-listing/temporary-activity-408/research-activities#Overview</u>

Health insurance: https://www.studyinaustralia.gov.au/english/live-in-australia/insurance

German information on going to Australia: http://www.reisebine.de/

Official government website with information about studying and living in Australia <u>www.studyinaustralia.gov.au</u>



## 2 Experience Reports from previous students

#### 2023 – Julika Wenzel



I have been working with the Bioinformatics Products team at CSIRO for about a month now. However, my first interaction with them was almost a year ago. At first, I was surprised how easy the application process was, with just an informal email and meeting. Now I think the Transformational Bioinformatics group wants to offer the opportunity to have a great international research experience to a motivated student and therefore, make it easy to apply.

My project involves developing and testing a tool to identify epistasis-like interactions between proteins. My tasks also include finding suitable data, pre-processing and statistical analysis of protein data. In weekly meetings with my supervisor, we discuss current and next steps, but I can also ask him questions at any time. During the weekly brainstorming meetings, I learn about other

projects in the group and can experience and participate in scientific discussions and brain-picking, which gives me insight into the world of research. Another thing I really appreciate is the opportunity to create learning experiences for myself that may not be directly related to my project. The great thing about Denis' group is that so many facets of bioinformatics, biology and computer science are covered by the team members, which means that there is probably someone who can educate me on what I am interested in. A great opportunity has been the chance to experience a major conference here in Sydney.

Thus far, I am enjoying my time at CSIRO and believe it will be a great overall experience that will help shape my future in the field of bioinformatics.

## 3 Project

Projects can be altered to fit the students interests and skills. The Transformational Bioinformatics group at CSIRO has a very broad spectrum of activities, ranging from human health to biosecurity; from basic science to real-world applications. We highly encourage you to check our webpage (<u>https://bioinformatics.csiro.au/</u>) for our activities and approach us with your **own project ideas**.

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	BSEP14 Data augmentation techniques for non-image data (genetics)	
	BSEP15 Application of Semantic Similarity Tools for Novel Disease Gene Analysis	
	BSEP16 Differential Privacy for Polygenic (risk) Models	
	BSEP17 LLM-powered chatbots for local documents (Informed consent materials)	

Project Title	BSEP01 Predicting disease-risk from genomic data using machine learning
Brief description	There is a significant genetic component to disease risks in humans. Can we
of the project	predict these risks early? Find out with Machine Learning (ML) and the world's
highlighting	largest genomic datasets. The student will compare the predictive
expected	performance of existing genomic risk models against ML algorithms for
outcomes	common (polygenic) diseases (e.g., Alzheimer's, cardiovascular diseases). At
	the end of the project, the student will determine the most suitable algorithm
	for generating polygenic risk scores, considering computational efficiency and
	its applicability across different ethnicities.

Duties/Tasks	<ul> <li>The student will perform</li> <li>Trait prediction analysis using VariantSpark</li> <li>Use HAIL/AWS with python notebooks for large scale data analysis</li> <li>Use ML algorithms to build predictive models with genetic data</li> <li>Compare currently available PRS models (e.g., LD-pred2, LDAK-SumHer, PRS-CS, PRSice) with ML algorithms</li> </ul>
Relevant field/s of	Machine learning
study	Bioinformatics
Supervisor	Letitia Sng
	Anubhav Kaphle
Contact Details	Letitia.sng@csiro.au
	Anubhav.kaphle@csiro.au
Location	Remote or in person in Sydney

Project Title	BSEP02 Science Writing/Communication for Blog posts
Brief description	The student will work with science experts to write up complex scientific
of the project	concept into communications pieces that are understandable by the general
highlighting	public. For IT project, the student will document the functionality, algorithms
expected	and optimisations in a markdown format with clear diagrams. Where
<u>outcomes</u>	applicable, the student will create non-scientific diagrams to aid the
	understanding. The resulting blog post will be published on
	bioinformatics.csiro.au.
Duties/Tasks	The student will perform
	Compile easy to understand blog articles
	<ul> <li>System analysis and produce software diagrams including, but not</li> </ul>
	limited to;
	<ul> <li>System Sequence Diagrams</li> </ul>
	<ul> <li>Architecture Diagrams</li> </ul>
	<ul> <li>Implement reproducible boilerplates (terraform/ serverless/</li> </ul>
	CloudFormation)
Relevant field/s of	Science communication
study	Bioinformatics
	Graphic design
Supervisor	Anuradha Wickramarachchi
Contact Details	Anuradha.Wickramarachchi@csiro.au
Location	Remote or in person in Adelaide

Project Title	BSEP03 Programming CRISPR scoring methods
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Brief description of the project	The student will learn how to 1) utilise existing CRISPR scoring algorithms (MIT, Tuscan); and 2) perform research into emerging CRISPR scoring
highlighting	methods (e.g. Cas-13) to develop reusable high-performance computing
expected	software libraries within a cloud-computing development environment. The
<u>outcomes</u>	resulting score-function will be included into the GT-Scan web service.
Duties/Tasks	The student will perform
	<ul> <li>Programming Python/C++/AWS</li> </ul>
	Translation of biological/genome-editing research into cloud software
Relevant field/s of	Bioinformatics
study	Software Programming
	Cloud computing
Supervisor	Tracey Wright
Contact Details	tracey.wright@csiro.au
Location	Remote or in person in Brisbane

Project Title	BSEP04 Machine Learning Model Selection for genetic data	
Brief description of the project <u>highlighting</u> <u>expected outcomes</u>	You will learn to execute machine learning (ML) methods on DNA samples and analyse the results. Some of the diseases of interest are Diabetes, Alzheimer's, Rheumatoid Arthritis, and ALS. We will provide you with some pointers on how to evaluate the model but also the freedom to pursue your own ideas. At the end of the project the aim is to have a comprehensive set of <u>evaluation methods for quantifying the performance</u> <u>of the ML algorithms</u> .	
Duties/Tasks	<ul> <li>The student will perform</li> <li>VariantSpark analysis (random forest)</li> <li>AWS + python (R is also possible)</li> <li>Identify and pursue metrics to quantify the model's performance</li> <li>Interpret and summarise results</li> </ul>	
Relevant field/s of study	<ul> <li>Bioinformatics</li> <li>Computer science</li> <li>Epidemiology</li> </ul>	
Supervisor	Roc Reguant	
Contact Details	Roc.reguant@csiro.au	
Location	Remote or in person in Sydney	

Project Title	BSEP05 Identification of epistatic biomarkers in a cancer dataset	
Brief description of	Metastatic cancer, cancer that has spread from its primary site, poses	
the project	significant challenges in terms of treatment and often carries a high risk of	
highlighting	mortality. For this project, we propose conducting a comprehensive	
expected outcomes	analysis of cancer data from a systems perspective. By incorporating	
	multiple variables (i.e., whole genomic sequencing, metadata) and their	
	interactions, we can potentially uncover novel insights that could	
	contribute to improved diagnostics and treatment strategies for metastatic	
	cancer. The work will be based on publicly available data	
Duties/Tesks	https://www.nature.com/articles/s41586-023-06054-z	
Duties/Tasks	The student will perform	
	VariantSpark analysis (random forest)	
	Epistasis detection tools	
	AWS + python	
	Interpret and summarise results	
Relevant field/s of		
study	Bioinformatics	
Supervisor	Roc Reguant	
	Mitchell O'Brien	
	Letitia Sng	
Contact Details	Roc.reguant@csiro.au	
	mitchell.o'brien@csiro.au	
	Letitia.sng@csiro.au	
Location	Remote or in person in Sydney	

Project Title	BSEP06 Database solutions for COVID-19 metadata analysis	
Brief description of the project <u>highlighting expected</u> <u>outcomes</u>	Millions of genomic samples of the COVID-19 virus are collected around the world and the cumulative data is reaching the limits of current infrastructure. The student will explore different database solutions i.e. relational and non-relational <u>with the aim to better incorporate</u> <u>metadata (e.g. sample information) to the genomic pipeline.</u>	
Duties/Tasks	<ul> <li>The student will</li> <li>Understand COVID19 genomic analysis</li> <li>Explore database solutions for metadata</li> <li>Incorporate database to existing genomic pipeline</li> </ul>	

Relevant field/s of study	<ul><li>Bioinformatics</li><li>Database management</li></ul>
Supervisor	Yatish Jain, Carol Lee
Contact Details	Yatish.Jain@csiro.au Carol.Lee@CSIRO.au
Location	Remote or in person in Sydney

Project Title	BSEP07 UI/UX DevOps for Genome Editing Platform
Brief description of the project <u>highlighting</u> <u>expected outcomes</u>	Genome Editing holds promise for a wide range of application areas, from human health to biosecurity. However, the key to unlocking this potential lies in enabling researchers to use the powerful but complex data analysis engine GT-Scan. The student will hence work on an intuitive user interface that empowers researchers to design genome editing experiments with ease.
Duties/Tasks	<ul> <li>The student will perform</li> <li>Conducting design interviews with users for desirable features (wireframe prototypes</li> <li>Implement the Frontend on AWS cloud</li> <li>Create visuals to advertise the tool</li> </ul>
Relevant field/s of study	<ul> <li>DevOps</li> <li>UX/UI experience</li> <li>cloud experience</li> </ul>
Supervisor	Denis Bauer
Contact Details	Denis.Bauer@CSIRO.au
Location	Remote or in person in Sydney

Project Title	BSEP08 Cluster analysis of pathogenic COVID-19 strains
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Brief description of the project <u>highlighting</u> <u>expected outcomes</u>	This project will build on a recent paper published in the lab regarding pathogenic and protective mutations in SARS-CoV-2 viral genomes. We will aim to cluster >12,000,000 viral samples in GISAID to determine patterns and whether pathogenic and protective strains cluster together and delve into the features shared in these clusters. The outcomes of this project will include analysis which could form the basis of a follow-up paper of our publication, and we will collaborate with RONIN and Intel to optimise our solutions.
Duties/Tasks	<ul> <li>The student will:</li> <li>Help build the optimal EMR configurations for the analysis of millions of datapoints</li> <li>Evaluate different cluster methods to find the optimal solution for our viral genome dataset</li> <li>Explore feature extraction methods</li> </ul>
Relevant field/s of study	<ul> <li>Experience with machine learning and feature extraction methods</li> <li>cloud experience</li> </ul>
Supervisor	Priya Ramarao-Milne
Contact Details	priya.ramarao-milne@csiro.au
Location	Remote or in person in Brisbane

Project Title	BSEP09 Identification of feature interactions for continuous variables
Brief description of the project <u>highlighting</u> <u>expected outcomes</u>	In biology, different systems interact and modulate each other. Current methods assume that one biomarker has one effect on our body. However, that is an oversimplification. We would like to improve current methods and develop a pipeline to quantify the additive effects interactions between biomarkers have on diseases. If successful, your name will be on a scientific publication.
Duties/Tasks	<ul> <li>The student will: <ul> <li>Implement well known Machine learning methods and algorithms</li> <li>AWS + python (R also possible)</li> <li>Identify interacting model features</li> </ul> </li> </ul>

	- Interpret and summarise results
Relevant field/s of	Bioinformatics
study	Computer science
Supervisor	Roc Reguant
	Priya Ramarao-Milne
Contact Details	Roc.reguant@csiro.au
	priya.ramarao-milne@csiro.au
Location	Remote or in person in Sydney

Project Title	BSEP10 Exploring the background on our identified COVID-19 Variants of Concern
Brief description of the project <u>highlighting</u> <u>expected outcomes</u>	The SARS-CoV-2 genome comprise of numerous mutations of varying significance and many of these are understudied. The student will use known signature mutations and other relevant mutations of SARS-CoV-2 strains to explore the adaptive advantage provided by a combination of these mutations. This will focus on the Alpha, Beta, Gamma, Delta, and Omicron Variants of Concern but may involve other strains of importance or high frequency. The project will allow us to gain a better understanding of the genetic make-up of SARS-CoV-2 and characteristics enhancing virulence.
Duties/Tasks	<ul> <li>The student will perform</li> <li>Comparisons between and within major SARS-CoV-2 strains</li> <li>Investigations into co-evolving mutations and their frequencies</li> </ul>
Relevant field/s of study	<ul><li>Bioinformatics</li><li>Phylogenetics</li></ul>
Supervisor	Carol Lee
Contact Details	<u>Carol.lee@csiro.au</u>
Location	Remote or in person in Sydney

Project Title	BSEP11 Enhancing genomic searching using metadata
Brief description of the	sBeacon presents a novel and scalable method to rapidly search
project <u>highlighting</u>	multiple genomic datasets for the presence and frequency of mutations
expected outcomes	of interest. Students will expand the capabilities of the service allowing,
	for example, the ability to search not just by genotype, but by
	phenotype, and to use the associated metadata to perform analysis
	across the phenome-genome space of millions of samples.
Duties/Tasks	The student will:

	<ul> <li>Add new search functionality over sample metadata</li> <li>Optimise the query pipeline to handle the increased data throughput</li> <li>Perform sample analysis to showcase capability</li> </ul>
Relevant field/s of study	<ul> <li>Computer Science</li> <li>Bioinformatics</li> <li>Cloud Experience</li> </ul>
Supervisor	Brendan Hosking
Contact Details	brendan.hosking@csiro.au
Location	Remote or in person in Sydney

Project Title	BSEP12 Identify long-term COVID possibility for different SARS-CoV-2 variants
Brief description of the	Genomes of different variants of SARS-CoV-2 are available in GISAID,
project <u>highlighting</u>	however the effects of protein fragments that remain after infection in
expected outcomes	the body is unknown. This project entails cataloging different peptides
	that can be generated from different SARS-CoV-2 variants and
	obtaining their pathogenicity score via pepbank. Overlaying this
	information with omics data from people with Long COVID may shed
	light on the molecular bases of this condition.
Duties/Tasks	<ul> <li>The student will:</li> <li>Write scripts to generate peptides of variable lengths</li> <li>Compare the generated peptides against peptide databases like pepbank or peptide atlas.</li> <li>Generate visualisations to present the metrics for different variants</li> <li>Submit analysis to https://virological.org/</li> </ul>
Relevant field/s of	Bioinformatics
study	Computer Science
	Proteomics
Supervisor	Yatish Jain
Contact Details	Yatish.jain@csiro.au
Location	Remote or in person in Sydney

Project Title	BSEP13 Text mining of scientific articles to create an epistasis database
Brief description of the	The number of scientific publications has been increasing exponentially
project <u>highlighting</u>	over the past years. Humans can barely keep up with all the novel
expected outcomes	scientific articles; therefore, computer-assisted tools to mine all the
	information available are on the rise. Currently, there are existing tools
	that mine how genes affect diseases; however, the potential

	interactions are oftentimes overlooked. Text mining of scientific articles
	to create an epistasis database The goal of this project is to create a
	tool that finds genetic interactions found in the literature.
Duties/Tasks	The student will:
	<ul> <li>Develop a script to scrape scientific articles.</li> </ul>
	<ul> <li>Apply ML algorithms to the extracted text</li> </ul>
	Create a database to store the results
Relevant field/s of	Bioinformatics
study	Computer Science
	Machine learning
Supervisor	Roc Reguant
Contact Details	Roc.reguant@csiro.au
Location	Remote or in person in Sydney

Project Title	BSEP14 Data augmentation techniques for non-image data (genetics)
Brief description of the project <u>highlighting</u> <u>expected outcomes</u>	There have been many efforts to perform data augmentation, especially in images, to improve deep learning models' performances. Data augmentation plays a significant role to increase data availability and balancing class distribution. In addition, it enhances the model's performance by improving generalization, mitigation of overfitting, and increasing model robustness. In this project, the student will implement data augmentation techniques to genetic data and assess if the above- mentioned benefits can be successfully transferred to this novel
Duties/Tasks	domain. The student will: • Run machine learning methods • Augment the data • Compare results
Relevant field/s of study	<ul> <li>Bioinformatics</li> <li>Computer Science</li> <li>Machine learning</li> </ul>
Supervisor	Roc Reguant
Contact Details	Roc.reguant@csiro.au
Location	Remote or in person in Sydney

Project Title	BSEP15 Application of Semantic Similarity Tools for Novel Disease Gene Analysis
Brief description of the	Rare diseases often pose challenges in clinical diagnosis and treatment
project <u>highlighting</u>	due to limited information on the associated genes and phenotypes.
expected outcomes	Semantic similarity tools, such as Phenolyzer, have shown promise in
	matching genes with known phenotypes. In this proposal, we aim to
	explore the application of these tools in analysing novel rare disease
	genes. By comparing their performance in known disease-causing
	genes and novel ones, we can assess the effectiveness of these tools
	and their potential for identifying new disease-gene associations
Duties/Tasks	The student will:
	Research tools for trial
	<ul> <li>Augment data from ClinVar and rare disease cohorts</li> </ul>
	Compare results
Relevant field/s of	Bioinformatics
study	Computer Science
Supervisor	Mitchell O'Brien
Contact Details	Mitchell.o'brien@csiro.au
Location	Remote or in person in Sydney

Project Title	BSEP16 Differential Privacy for Polygenic (risk) Models
Brief description	"Oh, the double-edge sword of genomic data privacy!"
of the project	
highlighting	Obtaining individual-level genetic datasets is challenging due to ethical and
expected outcome	privacy reasons; however, GWAS summary data are readily available, and many genomic tools increasingly utilise summary data nowadays. Nevertheless, sharing GWAS summary data can also result in privacy risks. To address this, differential privacy mechanisms have been proposed to perturb the data before public sharing. But with these mechanisms applied, how valuable are the shared data?
	This project aims to assess the impact of changing privacy settings for GWAS summary data on the effectiveness of constructing genomic prediction models. The student will conduct experiments by artificially perturbing SNP effect sizes in locally created GWAS summary data using differential privacy mechanisms and parameters. They will then develop trait prediction models and compare their performance by measuring the difference in R2 values or area under the curve (AUC) between models using the original and modified data. The objective is to evaluate the sensitivity of prediction models to privacy parameters and determine the usefulness of constructing prediction models using publicly shared GWAS summary data with privacy measures applied.

	Useful resources: <u>GitHub - IBM/differential-privacy-library: Diffprivlib: The IBM Differential</u> <u>Privacy Library</u> <u>GitHub - OpenMined/PyDP: The Python Differential Privacy Library. Built on</u> <u>top of: https://github.com/google/differential-privacy</u> <u>Privacy-Preserving Data Sharing for Genome-Wide Association Studies - PMC</u> <u>(nih.gov)</u>
Duties/Tasks	<ul> <li>The student will</li> <li>Generate in-house summary GWAS statistics (using tool such as PLINK, LDAK)</li> <li>Perturb the summary statistics using differential privacy mechanisms (assess Laplace mechanism with changing epsilon (ε) and delta (δ)).</li> <li>Create trait prediction models (using e.g., LDAK-SumHer, PRSice) based on the original and privacy-perturbed summary statistics.</li> <li>Test the prediction accuracy of the polygenic risk models based on R2, or area under the curve at different privacy parameters.</li> </ul>
Relevant field/s of	Machine learning
study	Data Privacy
	Bioinformatics
Supervisor	Anubhav Kaphle
Contact Details	Anubhav.kaphle@csiro.au
Location	Remote or in person in Melbourne

Project Title	BSEP17 LLM-powered chatbots for local documents (Informed consent materials)
Brief description	
of the project	This project focuses on addressing the challenges that participants in
highlighting	genomics research face when trying to understand and agree to study consent
<u>expected outcome</u>	documents, which are often lengthy and complex. To provide a more natural and interactive solution, we aim to develop a ChatApp using LangChain and OpenAI's GPT-3/4 APIs. The student involved in the project will work on developing and integrating the app with our informed consent management system. The ChatApp will enable participants to learn about the study and consent in a simple and user-friendly manner, while their understanding will be evaluated through quizzes. If possible, the project will also assess the

	chatbot's performance based on criteria such as accuracy, relevance, clarity, and user satisfaction. <b>Reference</b> : <u>https://dl.acm.org/doi/10.1145/3544548.3581252</u>
Duties/Tasks	<ul> <li>The student will perform these tasks:</li> <li>Data collection and pre-processing from consent information materials as well as relevant documents from online</li> <li>Chatbot development and training using LangChain and OpenAl's GPT-API</li> <li>Integration of the app to Dynamic consent management system</li> <li>Chatbot testing and evaluation using human subjects and quantitative and qualitative methods</li> </ul>
Relevant field/s of	Artificial Intelligence
study	Application development
	Chatbots
Supervisors	Anubhav Kaphle
	Anuradha Wickramarachchi
Contact Details	Anubhav.kaphle@csiro.au
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Location	Remote or in person in Melbourne



As Australia's national science agency and innovation catalyst, CSIRO is solving the greatest challenges through innovative science and technology.

CSIRO. Unlocking a better future for everyone.

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