

An Indian-Australian research partnership

**Project Title:**

Application of Artificial Intelligence in human genomics

**Project Number**

IMURA0806



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## Research Clusters:

## Research Themes:

Highlight which of the Academy's CLUSTERS this project will address? <i>(Please nominate JUST <u>one</u>. For more information, see <a href="http://www.iitbmonash.org">www.iitbmonash.org</a>)</i>		Highlight which of the Academy's Theme(s) this project will address? <i>(Feel free to nominate more than one. For more information, see <a href="http://www.iitbmonash.org">www.iitbmonash.org</a>)</i>	
1	Material Science/Engineering (including Nano, Metallurgy)	1	Advanced computational engineering, simulation and manufacture
2	Energy, Green Chem, Chemistry, Catalysis, Reaction Eng	2	Infrastructure Engineering
3	Math, CFD, Modelling, Manufacturing	3	Clean Energy
4	CSE, IT, Optimisation, Data, Sensors, Systems, Signal Processing, Control	4	Water
5	Earth Sciences and Civil Engineering (Geo, Water, Climate)	5	Nanotechnology
6	Bio, Stem Cells, Bio Chem, Pharma, Food	6	Biotechnology and Stem Cell Research
7	Semi-Conductors, Optics, Photonics, Networks, Telecomm, Power Eng	7	Humanities and social sciences
8	HSS, Design, Management	8	Design

## The research problem

*Define the problem*

Single nucleotide polymorphisms, called SNPs (snips), are the most common type of genetic variation among people. SNPs occur once in every 300 nucleotides on average in human genome. There are roughly 10 million SNPs in the human genome, and most commonly, these variations are found in the DNA between genes. In addition to snips, DNA-rearrangements, gene silencing, and satellites are also known to cause diseases. All of these can act as biological markers, helping scientists locate genes that are associated with disease.

## Project aims

*Define the aims of the project*

- Catalogue of genes, SNPs, and DNA elements associated with high – probability of diseases (literature review, and data base construction).
- Developing specific DNA-marker database(s) with special emphasis on cancer (/diabetics/ cardiovascular diseases): Global and Indian population SNP map, and their comparison.
- GWAS to estimate disease risk: Development and application of genome-based strategies for the early detection, diagnosis, and treatment of disease.
- Development of robust bio-informatics pipeline to study genes and DNA elements on a large scale genomic data analysis.
- Integration of big-data analytics tools and artificial intelligence (AI) for human genome analysis.
- Use of AI and machine learning (ML) in early diagnosis of diseases, through advance data processing (viz. image data analysis), and correlation with that of genome data.

## Expected outcomes

*Highlight the expected outcomes of the project*

- Novel biomarker discovery in human disease.
- Artificial Intelligence/Machine Learning methodologies in human genomics

### **How will the project address the Goals of the above Themes?**

*Describe how the project will address the goals of one or more of the 6 Themes listed above.*

The project will combine the knowledge of Artificial Intelligence/Machine Learning and human genomics and integrate them to discover novel biomarkers of disease.

### **Capabilities and Degrees Required**

*List the ideal set of capabilities that a student should have for this project. Feel free to be as specific or as general as you like. These capabilities will be input into the online application form and students who opt for this project will be required to show that they can demonstrate these capabilities.*

Artificial Intelligence/Machine Learning: preferred

Expertise in computer science and engineering

Knowledge of human biology and disease: preferred

Knowledge of genomics and biology: preferred

Competencies in computational biology: preferred

### **Potential Collaborators**

*Please visit the IITB website [www.iitb.ac.in](http://www.iitb.ac.in) OR Monash Website [www.monash.edu](http://www.monash.edu) to highlight some potential collaborators that would be best suited for the area of research you are intending to float.*