**UQ Summer or Winter Research Project Description**

Please use this template to create a description of each research project, eligibility requirements and expected deliverables. Project details can then be uploaded to each faculty, school, institute, and centre webpage prior to the launch of the program.

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| **Project title:** | **G**enetic causes of cleft lip (CL) and cleft palate (CP) |
| **Project duration:** | 10 weeks |
| **Description:** | **Background**  Cleft lip and cleft palate is also known as oral-facial cleft. Actually, it is a group of conditions, cleft lip (CL), cleft palate (CP) or a combination (CLP). When the cleft sits in the upper lip of patients, the condition is call CL; When the cleft sits in the roof of mouth, the condition is called CP; When the cleft sits in both positions, the condition is called CLP. These conditions frequently cause feeding difficulties, speech problems, hearing problems and some other problems ([1-3](#_ENREF_1)). The risk factors include genetic factors and environmental factors. The environmental influences include maternal smoking, maternal alcohol abuse, diabetes, obesity, age of parents, et al ([4-7](#_ENREF_4)). People with certain variants of some genes are with higher occurrence risk of these conditions. It has been suggested that these conditions might be a results of interplay of genes and environmental influences ([8](#_ENREF_8)). Cleft lip (CL), cleft palate (CP), and cleft lip with cleft palate (CLP) are among the most common birth defects in humans with prevalence between 1 in 500 and 1 in 2,500 live birth, with ethnic and geographic variation ([9](#_ENREF_9)). These condition results in about 3300 deaths globally in 2013, comparing 7600 deaths in 1990 ([4](#_ENREF_4)). These conditions can be treated with surgery with good outcome. However, they bring heavy financial burdens and cause psychosocial issues in kids with these conditions ([10](#_ENREF_10),[11](#_ENREF_11)). Though several genes and many loci have been suggested relevant to CL, CP and CLP, more studies needed to validate loci that have been reported relevant to these conditions.  **Resources**  This study will require collaborative efforts between Professor Adam Ye’s lab, School of Dentistry, UQ and Professor Jian Yang’s lab, Institute for Molecular Biology, UQ. Multi-discipline knowledge and skills are essential for the completion of this project. The knowledge and skills on bioinformatics will largely be provided by Professor Jian Yang’s lab, and biological and clinical knowledge and skills will be provided by Professor Adam Ye’s lab. We have been collecting essential genome data from public database, such as dbGap (<https://www.ncbi.nlm.nih.gov/gap>), and from collaboration parties. Professor Jian Yang’s lab is a strong lab on bioinformatics (<http://researchers.uq.edu.au/researcher/13615>), with more than enough facilities and resources to complete bioinformatics part of this project. Professor Adam Ye’s lab (<http://researchers.uq.edu.au/researcher/2713>) is a clinical, biological and nano-biomaterial research lab with access to facilities and resources for molecular biology, cell biology, biochemistry, imaging, animal experiments, et al, to complete functional validation parts of this project.  **Methods**  A method called SMR published by Professor Jian Yang’s group ([12](#_ENREF_12)) will be used to discover new candidate genes that integrates summary-level data from GWAS with data from expression quantitative trait locus (eQTL) studies to identify genes whose expression levels are associated with a complex trait. Molecular biology, cell biology and animal model experiments might be used in Professor Adam Ye’s lab to validate the candidate genes.  **Aims**   1. We attempt to uncover genes that are responsible for onset of CL, CP and CLP and have been or have not been reported to sit in loci that are relevant to these conditions from the efforts of Genome-Wide Analysis Studies. 2. We will validate results from Aim 1 with functional assays. |
| **Expected outcomes and deliverables:** | It is expected that the applicants could learn practical skills/techniques and acquire experiences on molecular biology, cell biology, light imaging (such as confocal fluorescence microscopy) and animal handling, et al. It is encouraged for the applicants to write scientific reports and to present the research output orally. |
| **Suitable for:** | *This project is open to applications from 3-4 year students enrolled at UQ with a background in molecular biology, bioinformatics and/or cell biology.* |
| **Primary Supervisor:** | Professor Adam Ye |
| **Further info:** | Please contact Professor Adam Ye at a.ye@uq.edu.au or +61 7 336 58078 for further information before you apply for this position. |

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3. Freitas, J. A., Garib, D. G., Oliveira, M., Lauris Rde, C., Almeida, A. L., Neves, L. T., Trindade-Suedam, I. K., Yaedu, R. Y., Soares, S., and Pinto, J. H. Rehabilitative treatment of cleft lip and palate: experience of the Hospital for Rehabilitation of Craniofacial Anomalies-USP (HRAC-USP)--part 2: pediatric dentistry and orthodontics (2012) *J Appl Oral Sci* **20**, 268-281

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11. Snyder, H. T., Bilboul, M. J., and Pope, A. W. Psychosocial adjustment in adolescents with craniofacial anomalies: a comparison of parent and self-reports (2005) *Cleft Palate Craniofac J* **42**, 548-555

12. Zhu, Z., Zhang, F., Hu, H., Bakshi, A., Robinson, M. R., Powell, J. E., Montgomery, G. W., Goddard, M. E., Wray, N. R., Visscher, P. M., and Yang, J. Integration of summary data from GWAS and eQTL studies predicts complex trait gene targets (2016) *Nat Genet* **48**, 481-487