# THE UNIVERSITY OF HONG KONG
# SCHOOL OF BIOLOGICAL SCIENCES

***Postgraduate Student Public Seminar***

# "EVOLUTIONARY POTENTIAL OF SMALL POPULATION ISOLATES"

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**Room 6N-11, Kadoorie Biological Sciences Building**

## Abstract

Small population isolates face severe evolutionary challenges due to the dual impacts of reduced effective population size ($N_e$) and restricted gene flow. Prolonged geographic isolation accelerates genetic drift and drives genome-wide inbreeding, often characterized by long runs of homozygosity (ROH). This process triggers genomic erosion by altering the dynamics of genetic variants, forcing recessive mutations out of a masked heterozygous state and allowing genetic drift to increase the burden of realized mutation loads. As ancestral variation becomes rapidly exhausted within these closed systems, *de novo* germline mutation emerges as the indispensable, primary fuel for future evolutionary adaptation, though precisely quantifying its per-generation rate remains a bioinformatic challenge. At the macroscopic level, this genetic decay is expected to break phenotypic buffering systems and directly compromise an organism's developmental stability under genetic or environmental stress.

To quantitatively investigate these consequences, in this thesis, I utilized the flat-headed loach (*Oreonectes platycephalus*) - a freshwater fish that lives in the headwaters of hill streams in southern China - as a highly replicated natural model system. First, a landscape survey across 16 populations confirmed that these loaches exhibit a high degree of genetic differentiation ($F_{ST}$), exceptionally low nucleotide diversity ($\pi$), as well as contemporary effective population sizes ($N_e^C$), signalling severe extinction risks. To overcome the technical barriers of non-model genomics, a high-quality, chromosome-level, haplotype-resolved reference genome was constructed based on PacBio HiFi long-read sequencing and Hi-C scaffolding technologies, providing a robust coordinate system for downstream genetic assessments. Further investigation revealed that elevated inbreeding coefficients ($F_{ROH}$) were tightly correlated with realized mutation loads. While natural selection demonstrated highly efficient purging of severely lethal loss-of-function (LoF) variants, mildly deleterious (missense) mutations systematically escaped and continuously accumulated via intense genetic drift. The fluctuating asymmetry (FA) of the lateral line system increased significantly alongside rising LoF loads within long-ROHs and decreasing $N_e^C$, providing direct evidence that recent inbreeding disrupts developmental stability. Finally, a precise *de novo* mutation rate ($\mu$) of flat-headed loach was estimated. By benchmarking family pedigree sequencing against PCR-verified mutations, long-read data was found to yield lower $\mu$ estimates than short-read data due to larger callable genome sizes.

To sum up, by leveraging a high number of replicate populations, this thesis demonstrates how long-term isolation drives genetic decay and phenotypic degradation in wild populations. Linking genomic erosion directly to physical asymmetry, these novel insights bridge the gap between theoretical population genetics and observable phenotypic fitness proxies. Ultimately, this work provides critical evolutionary parameters, methodological benchmarks, and framework solutions essential for the long-term conservation management and risk assessment of wild isolated species facing habitat fragmentation.

--- ALL ARE WELCOME ---