Linking genotype, phenotype, and fitness: Comparing genetic, behavioral, ecological, and morphological data in primatology

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The use of primates as a model organism is appealing to researchers as their close evolutionary relationship to humans provides insight into our own evolution. Primate work has produced large genetic, behavioral, ecological, and morphological data sets. Dr. Brenda Bradley’s Lab at The George Washington University takes advantage of this in their work to determine the genetic basis for adaptive molecular evolution in wild primate populations. Dr. Bradley’s lab uses both a top-down comparative genomic approach to search for evidence of selection, as well as a candidate gene approach to identify genes involved in phenotypes of interest. During her talk at the April 6th, 2018 Duquesne University Department of Biological Sciences Seminar, Dr. Bradley focused on her lab’s efforts to better understand the molecular evolution of certain genes involved in color vision, camouflage, and communication traits across primates.

Color vision is thought to be an adaptive advantage in foraging in many primate species. Individuals better able to discriminate color differences can more easily locate foods of different colors relative to background. The color vision in primates however is highly variable and many individuals have dichromatic (red-green color blind) or full trichromatic color vision. To better understand the genetic basis for this color-vision phenotypic
adaptation, the Bradley Lab focused on X-linked opsins, genes which code for opsin proteins necessary for light-sensitivity in the cones of the eye.

Primate eyes contain three types of light sensitive cones, which have short, medium, and long wavelength sensitivity and are autosomal (short) or x-linked genes (medium and long). Using a comparative genomics approach, 429 individuals from 8 genus and 18 species, it was determined there are only three sites that are different between the medium and long genes. If apes or humans only have the medium or long wavelength recognition gene that individual will be red-green colorblind. Phylogenetic comparative analysis by habitat and activity showed great variation in the distribution of polymorphisms by geographic location/habitat at the species, population, community, and organismal levels.

In the Bradley lab, the genetic basis of pigmentation and pelage (fur color) is being studied concurrently with color vision. DNA samples, geographical, and morphological data from 400 pelts/hairs preserved in museums were analyzed. DNA and RNA was extracted and DNA sequencing, mRNA/miRNA/whole transcriptome sequencing determined the distribution and expression of pigment cells across the body using the melanocyte marker gene MITF. Complete migration of pigment cells results in all black coloration, while white spots lack melanocytes. While changes in pigmentation and pelage can have direct adaptive benefits, natural and sexual selection do not always work in the same direction, resulting in adaptive molecular evolution that cannot always be interpreted as selecting for one type of fitness. For example, large ornamental structures to attract and compete for mates, may require a large energetic investment and make organisms more easily preyed upon. Therefore, coloration changes resulting from sexual selection may not have direct adaptive benefits.

The final and most interesting project in relation to humans, looks at the genetic basis of social communication. While non-primates lack the complex aspects of human language and communication, some similar
elements exist in non-human primates. They have been shown to use attention getting vocalizations and can recognize symbols.

Using the neurological and behavioral data of collaborators with over 100 captive subjects, Dr. Bradley decided to focus her labs attention on FOXP2, a candidate gene which plays a role in fine orofacial motor control\(^2\). Using a comparative genomic approach, the lab identified variation at FOXP2 and other language related genes across different primate species and at the intraspecies level. Phylogenetic analysis showed key amino acid changes at the human lineage not present in other primates. With these point mutations, structural and functional FOXP2 modeling between primates provides insight into language evolution. Interestingly, a small number of fixed nonsynonymous substitutions result in the unique human form of FOXP2. In addition, the small number of fixed and variable synonymous substitutions among non-human primates give rise to a large degree of variation in predicted protein structure and therefore function.\(^2\)

Primates, our closest evolutionary relative, have been studied in detail, however the genetic and molecular basis for much of the collected behavioral, geographic, ecological, and other data is not well understood. Using the wealth of genomic and demographic data sets available, Dr. Bradley can identify candidate genes associated with phenotypes of interest or selective pressures. Her lab has provided insight into the adaptive molecular evolution of color vision, body and hair coloration/camouflage, and communicative vocalizations, three adaptations of importance to non-human and human primates alike.

**References Cited**