REGENERON™
GENETICS CENTER

The DRIFT Consortium:
Discovery Research Investigating Founder Population Traits

A wholly-owned subsidiary of Regeneron Pharmaceuticals, Inc., the Regeneron Genetics Center (RGC) is a large-scale, fully integrated genomics program that builds on Regeneron's well-established expertise in genetics and related technologies. The objective of the RGC is to use DNA sequencing to gain new insights into disease mechanisms, identify novel therapeutic targets and ultimately speed drug discovery and development to help patients in need. We work collaboratively with academic colleagues worldwide and use a number of study designs and experimental approaches. One approach is to identify and study rare individuals with loss of function mutations and their associated phenotypes. Founder populations offer a unique opportunity to identify these informative alleles since they may be enriched in the population through drift and also may be more often found in their homozygous state through endogamy.

We have founded the DRIFT Consortium with the goals of (i) cataloging population-specific allelic architecture; (ii) understanding the biological and functional consequences of specific mutations identified; and (iii) sharing and establishing best practice approaches to relieve disease burden in these populations.

DRIFT is planning two tiers of collaboration models (see details below). For both models, we intend to broadly share data and results with the research community. If exciting new results are generated from a Tier 1 or Tier 2 collaboration, there will be potential for the design and funding of follow-up "genotype-first call-back" studies for additional collaborative research to delve more deeply into biological mechanisms and pathways.

Tier 1

GOAL: To canvas the allelic architecture of the population by exome sequencing and GWAS chips from relatively unrelated individuals

- Collaborator will provide the RGC de-identified DNA samples (300-400)
- RGC will provide high-depth exome sequence and GWAS chip data back to collaborator free of charge
- No exchange of phenotype information is necessary for Tier 1
- If the joint sequence data is used for any genotype-phenotype analyses, we ask that results be shared with Regeneron; collaborator is free to publish results
- A short form material transfer agreement will be used to govern the collaboration

Tier 2

GOAL: To establish a partnership/ collaborative effort focused on novel gene discovery for phenotypes of mutual interest

- An academic collaboration model where Collaborator and the RGC will jointly develop the research plan
- A larger number of DNA samples (100s – 10,000s) will be provided by Collaborator; the exact number will be determined jointly
- RGC will provide all exome sequence data to Collaborator free of charge
- As part of the collaboration, deidentified phenotype data will be shared
- Data analyses of the combined sequence and phenotype data set will be performed collaboratively and each party is free to use the data set for its internal research
- Collaborators will be encouraged to publish results and each party is free to use published results for any and all purposes
- Where justified, the RGC can provide financial support to Collaborator to assist with sample preparation/shipment, or towards other activities related to the collaboration

Program Leader & Contact:
Alan R. Shuldiner, M.D.
Vice President & Co-Head, RGC
Alan.Shuldiner@regeneron.com
914-847-1081