With the exception of injuries, all diseases have a genetic component. The goal of this workshop on human genetics and epidemiology is to familiarize participants on the genetic etiology of diseases, and the methods and techniques for conducting gene-disease association studies. Gene disease association studies are aimed at identifying whether a mutation or a polymorphism is associated with a disease/disorder, or susceptibility to the disease or disorder. Association studies require a combination of skills in both human genetics as well as epidemiology.

Theoretical:
- Genetic etiology of diseases
- Epidemiological study designs for gene disease association studies
- Government of India regulations and guidelines for conducting human genetic studies

Practical component:
- Designing a gene association study
- Writing an informed consent
- Laboratory techniques for studying mutations and polymorphisms
- Calculating genotypic and allele frequencies
- Measuring Hardy Weinberg Equilibrium
- Calculation and interpretation of Odds Ratios and Confidence Intervals

Contact: For further details about the workshop, please contact dranitakar@gmail.com or phone 020 25691758
Goal of the Workshop
What happens after candidate genes for a particular disease are identified? The goal of this workshop is to familiarize participants on the epidemiological methods used in deciphering whether a candidate gene is associated with a clinical condition or not.

Expected competencies from this Workshop
Participants undertaking this course will develop the skills of
- Laboratory methods for studying, interpreting and reporting mutations and polymorphisms from clinical samples,
- Learning basic epidemiological research designs,
- Learning the design and conduct of case-control studies, methods of drawing appropriate sample, power of studies, understanding the importance of HWE calculations, method of measuring genotypic and allelic frequencies, understanding and interpreting Odds Ratios and other statistical methods,
- understand the ethical and regulatory needs prior to conducting genetic studies,
- learn how to develop an information brochure and write informed consent,
- learn the use of online tools for measurement of various genetic measures

Each participant in the course will receive a protocol/study manual.

Lecture 1: Genes and disease
This lecture will describe the genetic contribution to disease aetiology. It will highlight the difference between mutations and polymorphisms, underlining the difference between mutations in disease causation, and association of polymorphisms with diseases and disorders. This lecture will highlight the polygenic and multifactorial nature of most diseases.

Lecture 2: The purpose of gene-disease association studies
Why are gene disease association studies needed? This lecture will discuss how a candidate gene is tested to identify whether it is associated with predisposition to a disease and disorder. This lecture will also explain why there are clinical guidelines for genetic testing in Europe and the US.

Lecture 3 Single gene disorders
Using the molecular pathology of thalassemia, this lecture will underline the causative role of mutations in the HBB gene for beta-thalassemia. The lecture will describe the most prevalent types of thalassemia mutations in India, and the role of secondary modifiers in influencing the clinical presentation.

Lecture 4: Genetic counselling
This lecture will underline the similarities and differences in genetic counselling for a monogenic condition (thalassemia), and a multifactorial condition (neural tube defects).

Lecture 5 and 6 Ethical and regulatory guidelines for conducting human genetic studies and Hands-on exercise I:
This lecture will deal with the ethical and regulatory guidelines for conducting human genetic studies. This will include a description of the PCPNDT Act, ICMR Ethics guidelines, and have a hands-on session on developing information brochure and informed consent forms for genetic analysis, using selected case studies.

Lecture 7-9
This session will briefly describe the principle of PCR, and describe two specific types of PCR: ARMS PCR for mutation analysis, and RFLP-PCR for analysis of the MTHFRC677T analysis.

Hands-on exercise II – III:
DNA extraction from whole blood and from buccal swabs; Group I: ARMS PCR for thalassemia, Group II: PCR-RFLP for MTHFRC677T polymorphism

Lecture 10:
This lecture will introduce participants to the science of epidemiology, the need for epidemiological study designs for association (case control) studies. The use of other types of epidemiological study designs (cross-sectional, cohort and randomized controlled trials) for understanding the role of genes in disease aetiology will also be discussed.

Hands on exercise IV
Reading and analysis of a gene disease association paper so as to understand the role of the lab and the necessity to design a study with appropriate samples and sampling.

Lecture 11 and Hands on exercise V: Statistical analysis of genetic data,
Theoretical discussion on estimating Hardy Weinberg Equilibrium; measuring genotypic and allele frequencies, statistical analysis of association of polymorphisms using different genetic models of inheritance. This session will go hand in hand estimating HWE, calculation of allele and genotypic frequencies, and Odds Ratios using established datasets and online calculators.
REGISTRATION FORM

(Registration limited to 12 participants on first come basis)

NAME:

DESIGNATION:

NAME OF THE INSTITUTE:

MAILING ADDRESS:

EMAIL ADDRESS:

PHONE NUMBER (PREFERABLY MOBILE):

DETAILS OF DD:

DD NUMBER:

ISSUING BANK:

AMOUNT:

Applicant Signature

Place:

Please submit registration form with DD attached to School of Health Sciences, SPPU, Pune-411007