SYLLABUS FOR GENZYME-SIAMG FELLOWSHIP IN CLINICAL GENETICS

Introduction

There have been significant advances in the field of Medical Genetics in recent years, which are of direct relevance to patient care. With completion of mapping & sequencing of the Human Genome it is expected that all the genes causing genetic disorders will be identified soon. This will include not only the genes causing monogenic disorders, but also those which predispose to complex multifactiorial disorders and those which may be responsible for causing cancer, autoimmunity & aging. It is possible to offer DNA diagnostic tests for screening of carriers and antenatal diagnosis, as well as premorbid diagnosis which would open avenues for primary prevention (predictive medicine). This has already led to radical changes in the concepts of genetic counseling & practice of clinical genetics. Access to these benefits that aim at birth of healthy children could be of immense importance to our National Family Welfare Program. Besides prevention, better understanding of pathophysiology of genetic diseases shall open new avenues of pharmaco-therapy, and even cure by gene therapy, of these disorders. This calls for availability of appropriate clinical services for counselling & management of patients with genetic disorders, backed up with adequate specialized laboratory support, which hardly exists in the country at present. At present there are very few clinical geneticists in India and there is only one training program in genetics for clinicians. There is an urgent need to create manpower trained in clinical genetics. The proposed course aims to fill this void.

General Objectives

The practice of clinical genetics is based on in-depth knowledge of basic genetic principles, a broad range of knowledge of genetic disease as it affects all body systems, and a clear understanding of the principles of genetic counseling. **The fellow in Clinical Genetics is to be trained in all aspects of genetics and**

medicine relevant to the practice of medical genetics, and should be capable of using this knowledge in the diagnosis and management of patients with gene related conditions. After successful completion of the program, the fellow shall be competent to diagnose genetic disorders, interpret genetic laboratory data, and have excellent communication and counseling skills.

Course curriculum

The training will be provided in the following areas:

- Eliciting of medical history of the index patient, including developmental and reproductive history;
- Eliciting of the family history, including drawing of detailed pedigree chart;
- Conduct of physical examination of affected and related individuals with special emphasis on morphological features and anthropometric measurements, and proper documentation of the findings, including photographs;
- Recognizing variations in human form (taking into account the features of the parents), and identification of *formae frustae*;
- Following a logical approach in syndrome identification (Dysmorphology) including the use of diagnostic aids e.g. computer assisted diagnosis, literature search;
- Recognizing the psycho-social and economic implications of the genetic problem in the family;
- Formulation of an appropriate differential diagnosis and a plan of appropriate medical consultations and investigations, and its discussion with the family (pre-test counseling);
- Ordering tests & other medical consultations, and performing current techniques of obtaining samples for genetic study e.g. skin biopsy etc., after obtaining the informed consent;

- Interpreting and explaining the results of genetic tests and other diagnostic studies, especially in areas of chromosomal, biochemical and molecular diagnosis (post-test counseling);
- Explaining the diagnosis, etiology, natural history, and management of the condition to the patient and the family;
- Providing general, supportive & specific medical care to the affected individuals, including appropriate interventions where necessary;
- Providing client-centered counseling and anticipatory guidance;
- Providing psychosocial support related to emotional, social, educational and cultural issues;
- Identifying and using community resources that provide medical, educational, financial and psychosocial support and advocacy;
- Determining the mode of inheritance and risk of occurrence and recurrence of the genetic condition/birth defect, and appropriate communication of the same to the patient and family, including availability of antenatal diagnosis and other reproductive options;
- Evaluating the client's and/or family's responses to the risk of occurrence;
- Promoting informed decision-making about further testing and management of the risk of occurrence/recurrence, including provision of antenatal diagnosis, if possible;
- Providing written documentation of medical, genetic and counseling information for families and other health professionals.
- Treatment of rare genetic disorders mainly inborn errors of metabolism
- Coordinate interdisciplinary management of genetic disorders
- Organize supportive care of genetic disorders
- Collaborate and communicate with genetic laboratories and act as a link between genetic laboratory and clinical specialties

All the above activities shall follow the ethical, social, & legal guidelines laid down for the purpose.

The student should be able to deal with clinical and applied laboratory aspects of the following clinical presentations of genetic disorders, including the exclusion of non-genetic causes: -

- Mental retardation/Developmental delay/Regression of milestones
- Congenital malformations, dysmorphism, multiple malformation syndromes
- Genetic hemolytic anemias
- Genetic disorders of blood coagulation
- Genetic neurological and muscle disorders, including disorders of senses such as deafness
- Disorders of male and female sexual differentiation and development, such as ambiguous genitalia, hypogonadism, primary amenorrhea and infertility
- Neonatal hyperbilirubinemia
- Short stature , including genetic disorders of skeleton
- Overgrowth syndromes
- Genetic disorders of eye and skin
- Recurrent spontaneous abortions, stillbirths and perinatal deaths
- Inborn errors of metabolism presenting as acute, intermittent or slowly progressive illnesses presenting during neonatal period, infancy childhood or at any age
- Any familial disorder
- Any congenital disorder
- Teratogenic exposure during pregnancy including drugs, infections and radiation

Eligibility for the course

The applicant must have a basic medical qualification recognized by the Medical Council of India (i.e. MBBS or an equivalent degree) and a postgraduate medical degree in Pediatrics/ Internal Medicine/ Obstetrics and Gynaecology recognized by the Medical Council of India (i.e. MD/MS/DNB or an equivalent degree).

Duration of Training/Rotation of training and Exit Evaluation under the Genzyme-SIAMG fellowship

The duration of training in the fellowship shall be of 3 months. The trainee should personally participate in the care of at least 50 patients with various genetic disorders, in outpatient and inpatient settings and the record should be maintained in a Log Book of clinical Training.

Provision shall be made for up to 3 months rotation in genetic laboratories to observe various diagnostic procedures. This includes one month each in Cytogenetics, biochemical genetics, and molecular genetics. Laboratory training will be concurrent with clinical training in OPD. The laboratory rotations are aimed at understanding principles of testing, interpreting results and understanding limitations and sources of errors in the tests.

The student will be required to attend academic activities of the department and will be required to present at least 2 seminars, 2 journal clubs and 3 case presentations. The student will be required to attend various additional courses held in the department.

Exit Evaluation:

The exit exam will be held at the end of 3 months. It will consist of Theory evaluation and Clinical/Practical/Oral evaluation.