INDIRA GANDHI INSTITUTE OF CHILD HEALTH
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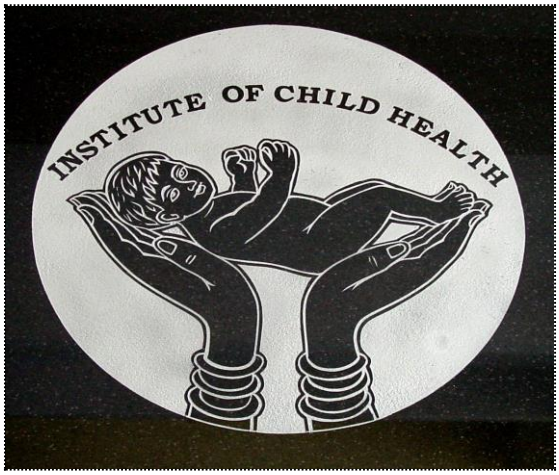
(AFFILIATED TO RAJIV GANDHI UNIVERSITY OF HEALTH SCIENCES, KARNATAKA)



PROSPECTUS
FOR
FELLOWSHIP IN PAEDIATRIC GENETICS

INDIRA GANDHI INSTITUTE OF CHILD HEALTH,
South Hospital Complex,
Dharmaram College Post, (Near Nimhans)
Bangalore-560 029.

FELLOWSHIP IN – **PAEDIATRIC GENETICS**

(Affiliated by Rajiv Gandhi University of Health Sciences)

FORMAT FOR DESIGNING CURRICULUM FOR FELLOWSHIP PROGRAMME IN PAEDIATRIC GENETICS

1) **Name of the subject: Paediatric Genetics**

2) **Please describe:**

a. Goals:

It is estimated that around 700, 000 births occur annually in India with a congenital anomaly or genetic disorder. As infectious disorders and infant mortality decrease, genetic disorders assume an increasingly important cause of neonatal and childhood mortality and morbidity. At present, only one centre in India (SGPGI, Lucknow) offers a DM programme in Medical Genetics. Understandably, there are few specialists with experience in clinical genetics for an increasing workload. Training pediatricians in the recognition and management of the commonest genetic disorders presenting in childhood is likely to be the way forward in addressing some of these needs.

There are no recognized training programmes in clinical genetics in Karnataka. The primary goal of this course will be to train interested and committed pediatricians in the sub-specialty of paediatric genetics to be able to:

- Construct and interpret a three generation pedigree in a family with suspected genetic disorder
- Evaluate an infant or a child with a common genetic or inherited metabolic disorder
- Arrange appropriate genetic tests with locally available resources
- Interpret the results of laboratory genetic investigations including chromosomes, FISH and DNA tests
- Counsel families with the commonest genetic disorders presenting in paediatric age group
- Organize family screening of other members at risk including appropriate prenatal diagnosis in a subsequent pregnancy
- Appropriately refer to senior colleagues where basic work-up has been completed

b. Statement of objectives of the Course:

The objectives of this course will to provide pediatrician trainees (Fellows) with

Knowledge –to equip pediatricians with ability to recognize, investigate, manage and counsel the commonest genetic disorders seen in paediatric practice. A mandatory rotation through the genetic laboratory to experience hands-on training in common cytogenetic and DNA techniques will familiarize them with the interpretation of these tests in the clinical situations encountered. The training will also enable them to focus on the family as a unit in genetic disorders rather than only on the index patient. It will also enable them to learn to serially test other family members at risk of a genetic disorder, the investigations available to a family in prenatal diagnosis and the interpretation of these prenatal diagnostic tests. All recent diagnostic concepts and therapeutic interventions available in genetic disorders will be taught in case based modules with information about availability and applicability of these technologies.

Skills – To enable pediatricians to acquire appropriate skills in the specialty for providing quality care to children and adolescents needing genetic evaluation and to provide counselling to their parents in order to help them attain the optimal results in the management of their child's disorder. The course will also provide trainees with skills of evaluation a pedigree, assessing genetic risk, interpreting genetic test results and the ability to professionally offer genetic counselling to these families

Communication abilities – Genetic counselling is an important and integral part of the management of a genetic disorder. In a country where majority of individuals are unfamiliar with the basic concepts of genetics and inheritance, the ability to make a family understand the basis of the genetic disorder in their family and its implications requires skill and good communication abilities.

c. **Course contents (syllabus):** Curriculum attached as Annexure A.

Essential knowledge: The curriculum gives details of all the topics that will be covered during the training period.

Essential investigations and diagnostic procedures –

1. A two month rotation in the genetic laboratory at the Centre for Human Genetics (CHG) will be an essential part of this training. In the laboratory, they will gain experience in processing a blood sample for chromosome analysis and the reporting of a karyotype.
2. DNA extraction from a blood sample and its preservation and uses in genetic testing will be done by all candidates.
3. Observation of special molecular cytogenetic techniques used in clinical diagnosis of genetic disorders and its interpretation. These will include FISH testing for rapid diagnosis micro-deletions, spectral karyotyping multi-colour FISH and others.
4. Observation and methodology of special prenatal testing will be arranged. This will include:
    a) Nuchal translucency scans: in the diagnosis of chromosome aneuploidies and cardiac disorders
    b) Anomaly ultrasound scans: in the detection of congenital anomalies in a fetus in the second trimester.
    c) Maternal serum screening for genetic disorders, its applications, interpretations and limitations
    d) Amniocentesis: as a procedure for definitive diagnosis of a chrosmosomal disorder in pregnancy
    e) Chorion villus sampling (CVS): in the diagnosis of single gene and inherited metabolic disorders.
5. Skeletal survey and its interpretation in the diagnosis of skeletal dysplasias
6. Observation and recognition of special cytology and histopathology in microscopic sections of bone marrow and liver biopsy in inherited metabolic and haematological disorders
7. Observation of biochemical tests to estimate enzymes and tandem mass spectrometry in bold samples for the diagnosis of inherited metabolic disorders
8. Newborn screening tests by tandem mass spectrometry.

d. Teaching / Learning activities:

The faculty will provide intensive hands-on training for all the trainees in all outpatient clinics, in carrying out various genetic investigations. The trainee will also actively participate in discussion with other specialties in the holistic management of patients seen. The trainee will function as a consultant in training and hence will learn all aspects of management of genetic patients in order to be able to manage and counsel the commonest genetic disorders presenting in the paediatric age group.

The trainee will be given every opportunity to participate in all teaching / learning and research activities of the department to enrich his/her knowledge of the subject. He/she will be encouraged to present cases for clinical discussions daily.

Clinical lectures will be organized weekly to cover theoretical topics from the proposed curriculum. In addition, the trainee will be expected to attend any important and relevant lectures or training programmes in Bangalore, provided the regular clinical duties of the trainee are not disrupted.

The trainee will also attend clinical pathology discussions, mortality meetings, journal clubs, seminars, symposia and other teaching activities of both institutions.

He / she will be expected to present posters and oral presentations in national and international conferences and prepare and publish case reports in consultation with senior colleagues in the department

The trainee will be expected to undertake a short and focused clinical project during the course with the aim of publication of this work in an indexed journal.

e. Participation in departmental activities:

1. **Journal review meetings:** Once a week
2. **Clinical lectures and seminars:** Once a week
3. **Clinico Pathological Conferences:** once a month
4. **Inter-Departmental Meetings:** Radiology and Biochemistry once a week
5. **Community Work:** – Camps / field visits: for diabetes education, visits to special education schools and training units.
6. **Clinical rounds:** – daily
7. **Outpatient clinics:** twice weekly in the assessment of patients with genetic disorders
8. **Participation in Conferences / presentation of papers:** National and state courses and conferences once year. International conference if possible in training period
9. **Any other:** Participation in a genetic project of the department and submission of a short dissertation carried out by the candidate.

f. Monitoring of Teaching / Learning activities:

(a) Methods: Journal reviews, seminars, case presentations, laboratory training in cytogenetics and DNA testing with evaluation of results and maintenance of a log book.

(b) Frequency: 3 – 4 / week,

(c) Schedules or Checklists, log books, diary: The department will have a trainee handbook / manual outlining all genetic laboratory training and fetal medicine procedures. Log books will be maintained by the trainee. Diary of all clinical cases seen and handled will also be kept.

g. Scheme of Examination: (Total marks: 400).

(a) Written: Two theory papers – 100 marks each, two essays 20 marks each, six short notes questions 10 marks each

(b) Clinical: Number & Type of cases: 2 to 4 clinical cases (100 marks) and "spotters" (50 marks)

(c) Viva Voce (50 marks)

h. Recommended Books and Journals:

1. Emery's Elements of Medical Genetics, Turnpenny and Ellard,14th edition
2. New Clinical Genetics, Read and Donnai, 2nd Edition
3. Oxford Desk reference: Clinical Genetics, Firth, Hurst & Hall
4. Practical Genetic Counselling, Ed: Peter Harper, OUP, 7th Edition
5. Introduction to Risk Calculation in Genetic Counseling, ID Young, 3rd Edition
6. Metabolic and molecular basis of inherited disease, Ed: Valle, Beaudet,Vogelstein,
7. Kinzler, Antonarkis, Ballabio, 8th Edition
8. Chromosome abnormalities and genetic counseling, Ed: Gardner and Sutherland, 3rd edition
9. Journal of Medical Genetics, BMJ publications
10. Journal of Genetics, Indian Academy of Science
11. Genetic databases:
12. OMIM (Online Mendelian Inheritance in Man)
13. LMD (London Dysmorphology Database)

(2). Rotation and posting in other departments

Two month laboratory rotation in genetics laboratory to have hands-on training in common diagnostic procedures.

One week in fetal medicine unit to observe common prenatal screening and diagnostic procedures

(3). Orientation Programme:

(a) Use of Library: regular use will be encouraged.

(b) Laboratory procedures: 2mths posting per year. Will need to become familiar with the laboratory procedures in genetic diagnosis and clinical interpretation of reports

(c) National Programmes: World Rare diseases day, World diabetes day

(d) Any other: visits to special needs schools and evaluation of children with physical and intellectual disability

(4). Training in Teaching Skills and Research Methodology:

The candidate will be expected to present cases and clinical signs in outpatient clinics and to postgraduates (DCH, MD, DNB courses) by way of case presentations, seminars and bedside teaching etc in the wards.

Patient and parent education and of other paramedical health professionals regarding implications and management of genetic disorder involved in the treatment will be undertaken by the trainee under supervision.

A research methodology course for 3-5 days will be arranged every year.

Concepts to be covered during the one year programme in Paediatric genetics:
Genetic and molecular epidemiology
Clinical evaluation and genetic counseling
Chromosomal disorders
Inheritance patterns- single gene mendelian inheritance pattern
Unusual modes of inheritance- Uniparental disomy, imprinting, triplet repeat disorders, mitochondrial inheritance
Pedigree taking and analysis
Phenotypes/genotype correlations
Testing protocols
Laboratory techniques in genetics - chromosomes, DNA and molecular cytogenetics (FISH)
Prenatal interventions for genetic conditions- prenatal diagnosis, pre-implantation diagnosis
Ethical issues in management of genetic disorders
Newer concepts: Cloning, human genome project, stem cell applications, gene therapy, pharmacogenetics and personalized medicine
Associated topics: clinical photography in genetic medicine, record keeping and retrieval, research methodology, public education and social impact of genetic disorders, developmental assessment and special education

These concepts will be covered in clinical modules starting with a patient evaluation and moving on to genetic investigations and counselling with introduction of ethical and social aspects as applicable to the subject. The commonest disorders and disorder groups seen in the Paediatric genetic clinics are listed below. The trainee will be expected to see affected individuals in all these categories and will learn management under the sub-headings listed under each topic.
A detailed clinical diary will be maintained by each candidate listing patients seen, investigations observed or done personally, courses and conferences attended, case reports prepared and published and lectures and other teaching programmes attended.

1. Down's syndrome
Clinical evaluation and genetic counselling
Chromosome studies and FISH
Maternal age & recurrence risks,
Prenatal diagnosis: nuchal translucency scans, serum screening, amniocentesis and rapid Trisomy screening-FISH & PCR based techniques
Population based maternal serum screening programs
Early intervention and Special education

2. **Other Chromosome imbalances**
Translocations, inversions, ring deletions additions in relation to clinical phenotypes
Chromosome analysis and clinical interpretation of these alterations
Family work up and genetic counselling
Implications for offspring risks and prenatal diagnosis
Special diagnostic techniques: spectral karyotyping and multi-colour FISH

3. Approach to ambiguous genitalia
Approach to sex chromosome disorders
Rapid FISH diagnosis and laboratory FISH session
Biochemical and endocrine evaluation in congenital adrenal hyperplasia
Management of ambiguous genitalia and counselling issues
Surgical interventions in ambiguous genitalia
Multidisciplinary approach to treatment and long term issues
Ethics of sex of rearing
Psychological issues in family counselling

4. Beta thalassemia
Case study and population genetics
Autosomal recessive inheritance
Haematological and genetic testing
Family screening and counselling
Prenatal diagnosis in beta thalassemia,
Treatment options including transfusion and chelation therapy, bone marrow and stem cell therapy
Long term issues and reproduction in patients with beta thalassemia

5. Duchenne muscular dystrophy
Clinical case history
X-linked inheritance and pedigree analysis,
Asymptomatic and manifesting carriers, gonadal mosaicism
Deletion and point mutation screening, newer diagnostic investigations including MLPA
Bayes clinical risk assessment and newborn screening
Occupational therapy, schooling and other interventional strategies
Newer therapies including exon skipping and gene therapy
Parent support groups and implications

6. Organic acidemias
Inborn errors of metabolism,
Autosomal recessive inheritance,
Newborn screening program and its interpretation
Presymptomatic diagnosis and treatment strategies,
Treatment in pregnancy and prenatal diagnosis
Dietary management in inherited metabolic disorders
Long term outcome

7. Lysosomal storage disorders
Chronic storage disorders including Gaucher, Mucopolysaccharidoses and Niemann-Pick disease
Pathogenesis and pathology
Enzyme based biochemical diagnosis
Mutation testing in LSDs
Inheritance patterns and counselling
Prenatal diagnosis in LSD
New therapies including bone marrow transplant, enzyme replacement and substrate reduction
Patient and parent support groups and range of activities covered

8. Ophthalmic genetics
Evaluation of a child with an inherited condition in infancy
Genetic testing and counselling
Evaluation of a newborn with congenital cataracts
Retinitis pigmentosa and genetic syndromes associated with RP
Eye manifestations in inherited storage disorders
Genetic heterogeneity and issues with genetic testing
Schooling in a child with visual handicap
Prenatal diagnosis of an inherited ophthalmic disorder
New therapies: stem cell therapy, gene therapy and bionic eye

9. Skeletal dysplasia:
Approach to short stature and evaluation of genetic aetiology
Achondroplasia: diagnosis, management, genetic testing and long term issues
Skeletal survey and its evaluation
Inheritance pattern and genetic counseling
Genetic testing and prenatal diagnosis in skeletal dysplasias
Skeletal involvement in lysosomal storage disorders
Therapeutic interventions: growth hormone therapy, surgical management and newer therapies
Schooling in a child with skeletal dysplasias

10. Obesity disorders:
Commonly seen obesity syndromes: Down's syndrome Bardet Biedl syndrome, Prader Willi syndrome, Beckwith Wiedemann syndrome, Albright's syndrome, congenital hypothyroidism and others
Evaluation of growth parameters and nutrition in childhood obesity
Genetic investigations and counselling in imprinted disorders
Genetic imprinting and parent of origin effects
FISH and special tests for genetic imprinting
Long term issues in genetic obesity

11. Diabetes mellitus:
Infantile and juvenile onset diabetes mellitus
Case study and population studies,
Multi-factorial inheritance and genetic counselling
Treatment strategies - newer insulins and pumps
Parent and patient education programmes
Genetic studies in a multi-factorial condition

12. Hearing disorders:
Pedigree analysis
Communication with deaf patients,
Investigation of hearing loss at appropriate ages
Autosomal recessive inheritance in Connexin 26 mutations,
Connexin 26 mutation testing and testing of other genes
Genotype-phenotype correlations
Ethical issues in prenatal diagnosis
Other hearing loss syndromes- Long QT, Brancio-oto-renal, Waardenburg and Usher syndromes
Speech and hearing intervention and techniques

Course design (Fellowship Course):

The trainee will spend 10 months in clinical rotation and two months completely in the laboratory. He / she will participate in the teaching programmes in the department.

Clinical:

The trainee will attend outpatient genetics clinic twice weekly. (On each of these days Dr Meenakshi Bhat will be present in OPD). The trainee will be also be responsible for evaluating and completing a genetic work-up proforma in all inpatient pediatric genetic and inherited metabolic disorder admissions, as well as interdepartmental consultations and emergencies.

A log book of all cases seen will be maintained. The log book must be submitted every month to a consultant and signed by him/her. The trainee is expected to become familiar with the patho-physiology and genetics, presentation, diagnosis and management, counselling of disorders of chromosome aberrations, common single gene disorders including Duchenne muscular dystrophy, beta thalassemia, achondroplasia, other skeletal dysplasias, genetic disorders with early onset vision and hearing loss, inherited metabolic disorders, genetic disorders with complex inheritance patterns including imprinted genetic disorders, mitochondrial inheritance and triplet repeat disorders, prenatal diagnosis in these disorders and long term management.

Laboratory:

1. A two month rotation in the genetic laboratory at the Centre for Human Genetics (CHG) will be an essential part of this training. In the laboratory, they will gain experience in processing a blood sample for chromosome analysis and the reporting of a karyotype.
2. DNA extraction from a blood sample and its preservation and uses in genetic testing will be done by all candidates.
3. Observation of special molecular cytogenetic techniques used in clinical diagnosis of genetic disorders and its interpretation. These will include FISH testing for rapid diagnosis micro-deletions, spectral karyotyping multi-colour FISH and others.
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7. Observation of biochemical tests to estimate enzymes and tandem mass spectrometry in bold samples for the diagnosis of inherited metabolic disorders
8. Newborn screening tests by tandem mass spectrometry.

During the laboratory rotation, the trainee will not have any ward or out-patient duties. Logbook will be maintained to demonstrate laboratory procedures done and observed including clinical interpretation.

Teaching programme:

The department already has case presentations 2 per week, seminar once a week, journal club and multi-disciplinary meetings once in 2 weeks, and pathology/mortality/research project presentation/combined endocrinology surgery meeting once a month. The trainee would present and participate in all of these along with the other postgraduates in the Institute. In addition the trainee will attend all teaching courses in both institutions that are relevant as well as other specialty lectures and programmes in Bangalore as long as these do not interfere with the formal schedule in the institute.

Evaluation:

Day to day evaluation of performance during clinical and laboratory rotations will be made. Clinical case presentations, seminars and other presentations will also be evaluated. Six monthly written and practical evaluations will be held. The result of these internal evaluations will be communicated to the trainee, and will be viewed as an exercise for self-improvement.

The exit examination will be held at the end of one year, as theory (200 marks) plus practical/viva (200 marks). Rajiv Gandhi University guidelines* will apply for dates of examinations. The numbers of theory papers, proportion of marks for theory and practical examinations, etc will be according to general University guidelines. Theory papers will examine the areas of basic genetics, recent advances and clinical pediatric genetic cases. Practical examination will consist of 2 to 4 cases and “spotters” (150 marks), and viva voce: X rays and genetic investigation interpretation (50 marks).

Evaluation of the programme is also envisaged. This will be done not only by a future board of studies, but also by the graduates of the course.

* (There will be 2 theory papers, and for the practical examinations, 1 external examiner and 2 internal examiners in addition to the Convener)

Duration of Course:

12 Months (full time work as per RGUHS guidelines and not permitted to work elsewhere)

Eligibility:

1. MD (Pediatrics), DNB or its Equivalent / DCH with Three Years Clinical Experience.

Selection:

1) Candidates will be selected from 4 member panel by interview (Approved by RGUHS)
2) If necessary entrance test will be conducted.

Fees & Stipend:

Fees for the course	:As per IGICH norms
Monthly stipend	:Rs.60000/- Per Month

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*In the event of the candidate leaving the course by discountenance or otherwise and thus failing to complete course;

1. The fee paid by candidate will not be refunded.
2. The stipend drawn by the candidate from the Institute during the period of the Fellowship programme to be paid to Institute.

Experience:-

Preference will be given to candidates having three years experience after post graduation in the concerned specialty.

Attendance & Leave:

As per University Guidelines

Faculty:

Staff from Indira Gandhi Institute of Child Health, Bangalore.

SCHEME OF EXAMINATION

THEORY EXAMINATION – includes 2 theory papers 100 marks each.

- Two long essays(20 marks each)
- Remaining six short essays (10 marks each)

PRACTICAL EXAMNATION – 2 Cases – 75 marks each
VIVA VOCE – 50 marks
Total – 400 marks

Copies of certificates to be enclosed with application (Originals at the time of Interview)

1. Photos –2
2. SSLC Marks card.
3. MBBS Degree Certificate and marks cards for all the four years.
4. MD(Paediatrics)/DCH Certificates / Marks card / Convocation Certificates
5. KMC Registration Certificate(updated qualification)
6. Application of in-service candidates should be routed through proper channel only.
7. Experience Certificate if any.

*** This Fellowship Programme is not recognized by Medical Council of India.**