Indian Council of Medical Research

Division of Reproductive Biology Maternal Health & Child Health

Call for letter of intent (LOI) for participation in National Registry for Rare Diseases

More than 70 million people of India suffer from the plethora of Rare Diseases, manifesting in childhood which remain with them throughout their lives. Registry is important for determining the precise number of patients, age of onset of symptoms, age of diagnosis (to determine for delay for making the diagnosis), the course of disease / natural history of disease. In this backdrop the ICMR proposes to set up national registry for rare diseases. Letter of intent is invited from interested investigators to take part in the national registry. To begin with following few disorders will be considered.

Broad outline of the LOI:

- Name of the institution
- Name of the investigator, coinvestigators (Paediatrician, geneticist,.etc)
- Approximate number of cases diagnosed per year **for the disorder of interest**

Disorders:

I. Lysosomal Storage Diseases – Diagnosis confirmed by Enzyme assay +/- Molecular Studies
   - Gaucher Disease
   - MPS I
   - MPS II
   - MPS III
   - MPS IV
   - MPS VI
   - Pompe disease
   - Fabry Disease

II. Inborn Errors of Metabolism (Small Molecule)- Diagnosis Confirmed by TMS & GCMS+/- Molecular studies
   - Maple Syrup Urine Disease
   - Phenylketonuria
   - Methylmalonic aciduria
   - Propionic academia
   - Urea cycle disorders
   - Glutaric Aciduria
   - Homocystinuria
   - Galactosemia

III. Skeletal Dysplasias – Diagnosis based on Phenotype & radiology +/- Molecular studies
   - Osteogenesis Imperfecta

- Achondroplasia+ others

IV. Hematological Disorders-
a)Hemophilia A &B
i. laboratory diagnosis of haemophilia with/without treatment facilities
ii. At least 50-100 diagnosed cases annually
b) Hemoglobinopathy:
i) Lab diagnosis of hemoglobinopathy with or without treatment facility
ii) At least 50-100 cases diagnosed annually

V. Primary Immune Deficiency
i) Immunodeficiencies affecting cellular and humoral immunity
ii) Combined immunodeficiencies with associated or syndromic features
iii) Predominantly antibody deficiencies
iv) Diseases of immune dysregulation
v) Congenital defects of phagocyte number, function, or both
vi) Defects in Intrinsic and Innate Immunity
vii) Autoinflammatory disorders
viii) Complement deficiencies
ix) Unclassified immunodeficiencies

VI. Neuromuscular Disorders
- Duchenne Muscular Dystrophy
- Becker Muscular Dystrophy
- Limb Girdle Muscular Dystrophy – and its type
- Spinal muscular Atrophy – Type 1 / 2 / 3
- Myotonic dystrophy

- Whether basic diagnostic and counseling facilities available in the institute
- Whether facilities for confirming diagnosis is available in the institute

Who can apply:
Research institutions and teams within the country who are involved / interested in research on rare diseases

How to apply?
The LOIs has to be submitted online following ICMR guidelines: www.icmr.nic.in . To submit a proposal online, please follow the following steps:

1. Go to ICMR website www.icmr.nic.in
2. Go to online submission of extramural research proposal
3. Click on new user for registering yourself as project investigator
4. Using the user ID and password begin submission of proposal by clicking on advertisement
5. Enter title of the call
6. Click on broad area as RCH
7. Call end date: 31st May 2017
8. Major discipline: Maternal and Child Health
9. Select programme officer : Dr. Reeta Rasaily

Review and selection process
Letters of intents will be reviewed and selection of sites will be done based on availability of infrastructure and cases. The short listed research teams will be notified and invited to participate to a proposal finalization workshop.

The deadline for the submission of letters of intent is 31 May 2017.

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