Announcement

Fifteenth ICMR Course in Medical Genetics & Genetic Counseling

Pedigree to Genome

Useful for clinicians from all branches of medicine and medical students

From 25th July 2016 to 6th August 2016

For details, see: http://www.sgpgi.ac.in/conf/icmr_course16.pdf

Contact

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Workshop

"To Develop a Scientific Program for Research on Rare Diseases"

April 22-23, 2016

Indian National Science Academy, New Delhi

Organizers: Dr V M Katoch, Dr P P Majumder and Dr A Bhattacharya

National Task Force Multicentric Collaborative Study of the Clinical Biochemical and Molecular Characterization of Lysosomal Storage Disorders in India

The Department of Health Research and Indian Council of Medical Research have constituted a National Task Force to perform a multicentric collaborative study at different research institutes across the country. The goal is to establish a smooth network of referral and counselling facilities for affected families. Accurate data management is planned to understand the overall impact of lysosomal disorders on patients, families and the communities. Nine different centres conduct mutation screening for about 30 different lysosomal storage disorders. Interested physicians can contact the principal investigators directly for mutation analysis. Further information can be found at,

http://www.icmrmetbionetindia.org/task-force-disoredrs.php

Lysosomal Storage Disease	Genes Tested	Principal Investigator (PI) and Co-PI	Centre
MPSI Hurler Syndrome	IDUA	Dr Madhulika Kabra madhulikakabra@hotmail.com Dr Neerja Gupta neerja17aiims@gmail.com	**AIIMS** All India Institute of Medical Sciences, New Delhi
Sanfilippo syndrome Type A/MPSIII A	SGSH		
Sanfilippo syndrome Type B/MPSIII B	NAGLU		
Sanfilippo syndrome Type C/MPSIII C	HGSNAT		
Sanfilippo syndrome Type D/MPSIII D	GNS		
Fucosidosis	FUCA1		
Gaucher Disease	GBA	Dr Jayesh Sheth jshethad1@gmail.com	**FRIGE** Foundation For Research in Genetics and Endocrinology, Ahmedabad
Juvenile Onset NCL	PPT1 TPP1		
Tay-Sachs/GM2 gangliosidosis	GM2A HEXA		
Niemann-Pick Disease Type A	SMPD1	Dr Ashwin Dalal ashwindalal@gmail.com Dr Prajnya Ranganath prajnyaranganath@gmail.com	**CDFD** Centre for DNA Fingerprinting and Diagnostics, Hyderabad
Niemann-Pick Disease Type C 1	NPC1		
Niemann-Pick Disease Type C 2	NPC2		
Mucolipidosis I/Sialidosis	NEU1		
Mucolipidosis II (I Cell Disease) & Mucolipidosis III A/B (Pseudo-Hurler polydystrophy)	GNPTAB /GNPTG		
Mucolipidosis type IV	MCOLN1		
Farber disease	ASAH		
MPS VI Maroteaux-Lamy	ARSB		
MPS VII Sly Syndrome	GUSB		

Sandhoff disease/GM2 gangliosidosis-Infantile	HEXB	Dr Parag Tamhankar paragtmd@gmail.com Dr Susan Thomas thomass@nirrh.res.in	**NIRRH** National Institute for Research in Reproductive Health, Mumbai
Krabbe disease	GALC		
Activator Deficiency/GM2 Gangliosidosis	GM2A		
Alpha-mannosidosis	MAN2B1		
Beta Mannosidosis	MANBA		
Morquio Type A/MPS IVA	GALNS	Dr Girisha KM girishkatta@gmail.com	**KMC** Kasturba Medical College, Manipal
GM1 gangliosidosis	GLB1		
Metachromatic Leukodystrophy	ARSA	Dr Seema Kapoor drseemakapoor@gmail.com	**MAMC** Maulana Azad Medical College, New Delhi
Sphingolipid activator protein deficiencies	PSAP		
Infantile Free Sialic Acid Storage Disease /ISSD	SLC17A5		
Gaucher Disease	GBA	Dr Sankar VH sankarvh@gmail.com	**SATH** SAT Hospital, Trivandrum
Galactosialidosis	PPGB		
Fabry disease	GLA	Dr Shubha Phadke shubharaophadke@gmail.com	**SGPGIMS** Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow
MPS II Hunter syndrome	IDS		
Schindler disease	α-NAGA		
Pycnodysostosis	CTSK		
Wolman disease	LIPA		
Pompe Disease/Glycogen storage disease type II	GAA	Dr Ratna Dua Puri ratnadpuri@yahoo.com	**SGRH** Sri Ganga Ram Hospital, New Delhi
Multiple sulfatase deficiency	SUMF1		
Aspartylglucosaminuria	AGA		