



# International Conference on Human Genetics & 39<sup>th</sup> Annual Meeting of Indian Society of Human Genetics

*Healthy Genes - Healthy Life*

22<sup>nd</sup> - 25<sup>th</sup> January, 2014 | Ahmedabad

## Conference Program

**Organized by:**

Foundation for Research in Genetics & Endocrinology [FRIGE] - IHG &  
Institute of Life Sciences [ILS] - Ahmedabad University, Ahmedabad

[www.ishg2014.org](http://www.ishg2014.org)

# 23<sup>rd</sup> Jan 2014

## HALL A

- 8:30-10:10 AM **Session I-Human Genome/Variome Project**  
**CHAIRPERSONS:- Dr Rama Vaidya & Professor I C Verma**
- 8:30-8:55 AM Human Variome Project - Current Overview -**Dr Richard Cotton, Australia**
- 8:55-9:20 AM "3D Facial Phenotyping -  
**Professor Peter Hammond, United Kingdom**
- 9:20-9:45 AM Identification of Novel Genes for Human Disease - **Dr Radhakrishna Uppala, Switzerland**
- 9:45-10:10 AM Deciphering the Basis of Genomic Variability through phenotype - phenotype links: A novel Ayurgenomics Approach - **Dr Bhavana Prasher**

## HALL B

- 8:00-10:05 AM **Session II- Complex Diseases: Diabetes and Cardiovascular Disorders**  
**CHAIRPERSONS:- Dr Premal Thakore & Professor Rolf-Dieter Wegner**
- 8:00-8:25 AM Genetic of Type 2 Diabetes -  
**Dr Dwaipayan Bharadwaj**
- 8:25-8:50 AM Is it time to take Genomics to the Diabetic Clinics? -  
**Dr Radha Venkatesan**
- 8:50-9:15 AM Prakruti Genomics and Prameha-Proclivity: Relevance to Metabolic Syndrome -**Dr Ashok Vaidya**
- 9:15-9:40 AM Genetics of Cardiovascular Disorders: Influence of Maternal Nutrition -  
**Dr Sukhinder Kaur Cheema, Canada**

10:15-10:45 AM **Hall (A) Keynote lecture - Dr Vishwa Mohan Katoch, Director General, ICMR**  
**CHAIRPERSONS:- Dr Richard Cotton & Dr Partha Majumdar**

10:45-11:00 AM **Tea**

**HALL A**

- 11:00-1:05 AM **Session III-Cancer Genetics**  
**CHAIRPERSONS:- Dr Shilin Shukla & Professor Sanjeev Misra**
- 11:00-11:25 AM Affordable Diagnosis and Prevention of Genetic Disease - **Professor Sir John Burn, United Kingdom**
- 11:25-11:50 PM An Empirical Assay for Assessing Genomic Sensitivity: Implications for Improving Cancer Diagnostics - **Professor Diana Anderson, United Kingdom**
- 11:50-12:15 PM Mutational Landscape of Gingivo-Buccal Oral Cancer: New Cancer Genes and Molecular Subgroups Identified - **Professor Partha Majumdar**
- 12:15-12:40 PM Functional Genomics of Lung Cancer Progression Reveals Mechanism of Metastasis Suppressor Function - **Dr Shantanu Chowdhury**
- 12:40-1:05 PM Non-Coding RNA Regulation of Blood Vessel Development in Zebra Fish and Relevance to Humans - **Dr Sridhar Sivasubbu**

1:00-2:00 PM

**HALL B**

- 11:00-12:45 PM **Session IV - Molecular Cytogenetics in Disease Diagnosis**  
**CHAIRPERSONS:- Professor Syed Hashsham & Dr Ken McElreavey**
- 11:00-11:30 AM Small Supernumerary Marker Chromosomes - An Update - **Dr Thomas Liehr, Germany**
- 11:30-11:55 AM Molecular Cytogenetics in Intellectual Disabilities - **Dr Madhulika Kabra**
- 11:55-12:20 PM Molecular Cytogenetic Characterization of Chromosomal Rearrangements - Utility in Genetic Counseling and Research - **Dr Ashwin Dalal**
- 12:20-12:45 PM Micro-deletion Syndromes - **Dr Chaitanya Datar**

Lunch

## HALL A

2:00-3:00 PM **Free Paper Session (HALL A) / Poster Presentation (HALL C)**

**CHAIRPERSONS:- Dr Shalmi Mehta & Dr Prakash Gambhir**

2:00-2:10 PM A Poisson Regression Model For Association Mapping Of Count Phenotypes - **Saurabh Ghosh, India**

2:15-2:25 PM Therapeutic potential of Histone Deacetylase Inhibitor for treatment of Niemann - Pick type C1 Disease - **Nina H. Pipalia, USA**

2:30-2:40 PM Genetics of Autism Spectrum Disorder & BDNF Gene - **Dr Usha P. Dave, India**

2:45-2:55 PM Identification and Clinical Evaluation of Segments of Homozygosity, Uniparental Disomy and Complex Chromosomal Abnormalities Revealed by Copy-Number SNP Arrays - **Trilochan Sahoo, USA**

3:00-4:45 PM **Session V - Current Trends in Prenatal Screening of Genetic Disorders**

**CHAIRPERSONS:- Dr Jigish Trivedi & Dr Mayank Chowdhury**

3:00-3:30 PM Non-invasive Prenatal Diagnosis using Massively Parallel Sequencing - First Experience in Germany - **Professor Rolf-Dieter Wegner, Germany**

3:30-3:50 PM Basic Principles of Prenatal Screening for Aneuploidies - **Dr Prakash Gambhir**

3:50-4:10 PM Fetal Medicine: Multispeciality approach - **Dr Sharad Gogate**

4:10-4:30 PM Prenatal Screening for Mendelian Disorders in Antenatal Care - **Dr Amar Verma**

## HALL B

3:00-4:40PM **Session VI - Single Gene Disorders**

**CHAIRPERSONS:- Dr Harshuti Shah & Dr Raju C Shah**

3:00-3:25PM Single Gene Disorders: Indian Scenario - **Professor I C Verma**

3:25-3:50PM DMD - Diagnosis and Management Challenges - **Dr Swati Shah**

3:50-4:15PM Mutation Spectrum of Cystic Fibrosis in India - **Dr Renu Saxena**

4:15-4:40PM Molecular Diagnosis for Genodermatosis - **Dr Parag Tamhankar**

**HALL A**

4:30-4:45PM Non-invasive prenatal testing, a better option for patients - **Dr Ashish Fauzdar**

4:45-5:00PM

5:00-6:15 PM **Session VII - Genetic Basis of Reproductive Disorders**

**CHAIRPERSONS:- Dr Manish Banker & Dr Prashant Acharya**

5:00-5:25PM Exome Sequencing Reveals Novel Genetic Causes of Human Infertility - **Professor Kenneth McElreavy, France**

5:25-5:50PM Genetics of Male Infertility : Indian Scenario - **Dr K Thangaraj**

5:50-6:15PM Fertility Options for Women with Cytogenetic Infertility - **Dr Rama Vaidya**

**HALL B**

Tea

5:00-6:40 PM **Session VIII - Epigenomics**  
**CHAIRPERSONS:- Dr G R Chandak & Dr Mitali Mukherji**

5:00-5:20 PM Uniparental Disomy - Clinical Consequences due to Imprinting and Activation of Recessive Genes - **Dr Thomas Liehr, Germany**

5:20-5:40 PM Epigenetic Alterations during Cervical Cancer Progression - **Dr K Satyamoorthy**

5:40-6:00 PM In search of Epi-driver Genes in Head and Neck Cancer - **Dr Susanta Roychoudhury**

6:00-6:20 PM Birth Defects: Etiology to Prevention - **Dr Koumudi Godbole**

6:20-6:40 PM Genomic packaging and epigenetic regulation of genes - **Dr Rakesh Mishra**

7:00PM Onwards

**Conference Dinner at AMA**

## HALL C

2.00-3.00 PM

POSTER PRESENTATION

## HALL D

5:00-6:30 PM

**WORKSHOP - PCR Based Strategies  
for Genetic Testing**

**Workshop Faculty:**

**Dr Parag Tamhankar (India)**

**Coordinator :** Mehul Mistri, Harsh  
Sheth, Avisek Mazumdar & Chitra  
Ankleshwaria



# 24<sup>th</sup> Jan 2014

## HALL A

- 8:00-9:50 AM **Session IX - Next Generation Sequencing**  
**CHAIRPERSONS:- Dr Shubha Phadke & Professor Virginia Kimonis**
- 8:00-8:30 AM Non Syndromic Sporadic Intellectual Disability: An Exome Sequencing Study Our experience -  
**Professor Anita Rauch, Switzerland**
- 8:30-9:00 AM New Paradigms in Whole Genome Sequencing: from Lab Bench to Cell Phone - **Jonathan O'Halloran, United Kingdom**
- 9:00-9:25 AM From Personal Genomes to Precision Medicine - **Dr Vinod Scaria**
- 9:25-9:50 AM Exome Sequencing Applied to Disorders of Sex Development (DSD) - **Professor Kenneth McElreavey, France**

## HALL B

- 8:00-9:45 AM **Session X - Diagnosis and Therapeutic Approach of Lysosomal Storage Disorders**  
**CHAIRPERSONS:- Dr Usha Dave & Dr Inusha Panigrahi**
- 8:00-8:30 AM Diagnosis and Management of Lysosomal Storage Disorders: Gaucher Experience in Israel -  
**Professor Hanna Rosenbaum, Israel**
- 8:30-8:55 AM ERT for Lysosomal storage disorders: Our Experience in India -  
**Dr Mamta Muranjan**
- 8:55-9:20 AM Mucopolysaccharidosis: Diagnosis to Therapeutics - **Dr Sunita Bijarnia**
- 9:20-9:45 AM Molecular Study of Lysosomal Storage Disorders in India - **Dr Jayesh Sheth**

9:55-10:15 AM

Tea

**HALL A**

- 10:15-12:15 PM **Session XI - Array-CGH**  
**CHAIRPERSONS:- Dr Anaita Hegde & Dr Suresh Amin**
- 10:15-10:40 AM The Advantages of SNP Arrays over CGH Arrays - **Dr Boris Keren, France**
- 10:45-11:05 AM Cytogenetic Microarray in Prenatal and Postnatal Diagnosis -  
**Dr Shubha Phadke**
- 11:05-11:25 AM Genomic Copy Number Variations in Glaucomatous Neurodegeneration -  
**Dr Arijit Mukhopadhyay**
- 11:25-11:55 AM Clinical Utility and Dilemmas of SNP Microarray Testing - **Professor Virginia Kimonis, USA**
- 11:55-12:15 PM Detection and Inheritance Pattern of CNVs in MCA - **Dr Frenny Sheth**

**HALL B**

- 10:15-11:35 AM **Session XII - Genetics of Neurobiology**  
**CHAIRPERSONS:- Dr Sanjiv Mehta & Dr Sudhir Shah**
- 10:15-10:45 AM Neuroferritinopathy: Iron in the Brain -  
**Professor Sir John Burn, United Kingdom**
- 10:45-11:10 AM Clinical Aspects of Neuroregression: Our Experience on Batten disease -  
**Dr Mahesh Kamate**
- 11:10-11:35 AM Early Stress Evokes Age-Dependent Biphasic Changes in Hippocampal Neurogenesis, Epigenetic Regulation of the Bdnf Gene, and Cognitive Behaviour -  
**Dr Ankit Sood**

**12:00-1:00 PM Free Paper Session (HALL B)**

- CHAIRPERSONS:- Dr Bipin Shah & Dr Darshna Naik**
- 12:00-12:15 PM Utility of CD-138 Negative Fraction for Chromosome Analysis in Plasma Cell Dyscrasias (PCD): A Novel Approach -**Dr Gopalrao Velagaleti**
- 12:15-12:30 PM Prenatal Screening above aneuploidies : Pre-Eclampsia - **Dr Gurjit Kaur**
- 12:30-12:45 PM Suspected Microdeletion Syndromes and Molecular Cytogenetic Techniques: An Experience with 330 Cases - **Dr Asutosh Halder**
- 12:45-1:00 PM Molecular Characterization of Mutations in Galactosemia Genes: Structural and Functional Implications - **Dr Rejendra Prasad**



## HALL A

1:00-2:00 PM

1:00-2:30 PM

2:30-4:20 PM

**Session XIII: Inborn Errors of Metabolic Disorders**

**CHAIRPERSONS:- Dr Parag Dagli & Dr Radha Rama Devi**

2:30-3:00 PM

Dysmorphology of Inborn Errors of Metabolism -

**Professor Virginia Kimonis, USA**

3:00-3:20 PM

New Born Screening Program in India: ICMR Multicentric Experience -  
**Dr Roli Mathur**

3:20-3:40 PM

Application of Tandem Mass Screening in IEM - **Dr Rohit Cariappa**

3:40-4:00 PM

National Newborn Screening the Road Map - **Dr Seema Kapoor**

4:00-4:20 PM

Treatment of Inborn Error of Metabolism - **Dr Anil Jalan**

4:20-5:00 PM

**Hall (A) Keynote lecture -System Biology Platform for Genome Sequence to Novel Drug Target Identification - Professor Samir Brahmachari, Director General, CSIR**

**CHAIRPERSONS:- Professor Sir John Burn & Dr Ashok Vaidya**

## HALL B

Lunch

Poster Presentation (Hall C)

2:30-4:20 PM

**Session XIV: Inherited Blood Disorders: Hemoglobinopathies**

**CHAIRPERSONS:- Dr Annie Hasan & Dr Nidhish Nanavaty**

2:30-3:00 PM

New Genes and Mechanism Regulating Blood Formation - **Dr Dhvanit Shah, USA**

3:00-3:20 PM

Thalassemia-Can We Reduce the National Burden? - **Dr Roshan Colah**  
Hemophilia - Diagnosis and Management Challenges -

3:20-3:40 PM

**Dr Shrimati Shetty**

3:40-4:00 PM

Current Status of Sickle Cell Disease in India - How Can You Attenuate? -

**Dr Dipika Mohanthy**

4:00-4:20 PM

Community Genetic Approaches in the Prevention of  $\beta$ -Thalassemia -

**Dr V R Rao**

**HALL A**

5:00-5:15 PM

5:15-6:30 PM **Session XV - Human Chromosomal Disorders**  
**CHAIRPERSONS:- Dr Balraj Mittal & Dr Thomas Liehr**

5:15-5:40 PM Chromosomal Instability and Molecular Mutations in Multi Spectrum Disease of Fanconi Anemia - **Dr V Babu Rao**

5:40-6:05 PM Newer Methodologies of Chromosomal Diagnosis and Role of Existing Ones - **Dr Hema Purandare**

6:05-6:30 PM Illumina : Integrating Technologies to Provide Clinically Meaningful Solutions - **Dr Debjani Saha**

**HALL B****Tea**

5:15-6:30 PM **Session XVI: Nanotechnology in Biomedical Research**

**CHAIRPERSONS:-Professor Alok Dhawan & Professor Diana Anderson**

5:15-5:45 PM Nano Mission Programmes - **Dr Praveer Asthana**

5:45-6:15 PM Self-Therapeutic Nanoparticle - **Dr Priyabrata Mukherjee, USA**

6:15-6:45 PM Unlocking the Chemistry of Bile Acids for Cancer Therapeutics - **Dr Avinash Bajaj**

## HALL C

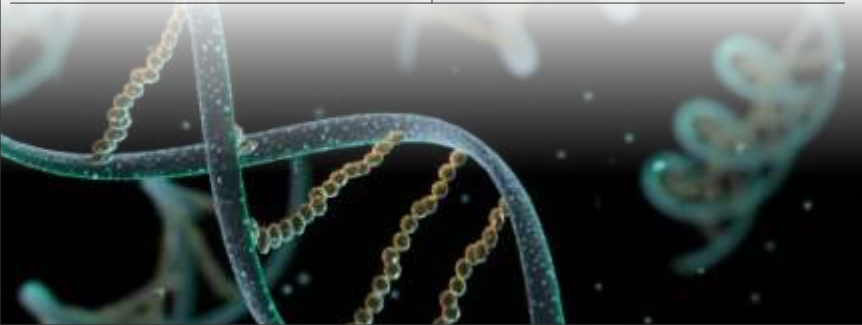
1.00-2.30 PM POSTER PRESENTATION

## HALL D

3.00-4.00 PM ISHG Society General Body Meeting  
5:00-6:30 PM WORKSHOP - NASI Student - Teacher interaction program

**Workshop Coordinator** - Dr G R Chandak, Dr Mitali Mukerji & Professor Alok Dhawan

**Workshop Faculty** - Professor Sir John Burn, Dr Thomas Liehr, Professor Harish Padh, Dr Kenneth McElreavey, Mr Jonathan O'Halloran, Dr Dhvanit Shah & Professor I C Verma



25<sup>th</sup> Jan 2014

HALL A

HALL B

8:30-10:30 AM ISHG Young Scientist Awards & Lecture (Hall A)

**CHAIRPERSONS:- Dr K Thangaraj & Dr Q Annie Hasan**

**Invited Guest:- Mr Pankaj Patel (Chairman and Managing Director -ZYDUS CADILA & Patron - ISHG2014 Conference)**

11:00-11:15 AM

Tea

11:15-1:00 PM **Session XVII: Pre-implantation and Prenatal Diagnosis of Genetic Disorders: Indian Scenario**

**CHAIRPERSONS:- Dr Atul Munshi & Dr Bindu Shah**

11:15-11:40 AM Pre-implantation Diagnosis in Cases of Parental Chromosomal Translocations Applying Array-CGH -  
**Professor Rolf Dieter Wegner, Germany**

11:40-12:05 PM Challenges of Prenatal and Pre-implantation Genetic Diagnosis Studies -  
**Dr Prochi Madon**

12:05-12:30 PM Role of USG in the Prenatal Diagnosis of Genetic Disorders -  
**Professor Rolf Becker, Germany**

11:15-12:30 PM **Session XVIII: Significance of Pharmacogenomics in Clinical Research**

**CHAIRPERSONS:- Professor R K Goyal & Dr Navneet Shah**

11:15-11:40 AM Newer Vistas of Pharmacogenomics -  
**Professor Harish Padh**

11:40-12:05 AM Pharmacogenomics of Cardiovascular Drugs -  
**Dr C Adithan**

12:05-12:30 AM Point of Care Testing for Improving Risk-Benefit Ratio of Aspirin and Warfarin -  
**Mr Harsh Sheth, United Kingdom**

**HALL A**

12:30-12:55 PM Consanguinity and perinatal medicine -  
the Berlin perspective -  
**Professor Rolf Becker, Germany**

1:00-2:00 PM

2:00-3:20 PM **XIX: Gene Discovery - Translational  
Research from Bench to Bedside**  
**CHAIRPERSONS:- Dr Amit  
Chakravarty & Dr Madhulika Kabra**

2:00-2:25 PM Digital Health, Micro Fluidics and  
Genetic Testing -  
**Professor Syed Hashsham, USA**

2:25-2:50 PM G protein Signaling in Tumor Cell  
growth and Metastasis - **Dr Danny N.  
Dhanasekaran, USA**

2:50-3:10 PM Vitiligo: A complex Disease and a  
Complex Approach -  
**Professor Rasheedunnisa Begum**

3:10-3:30 PM STR Markers in Clinics : A Rapid Prenatal  
Diagnosis by QF-PCR for Aneuploidies -  
**Dr Sarita Agarwal**

3:30 PM Onwards

4:30 PM Onwards

**HALL B**

**Lunch**

2:00-3:15 PM **Session XX: Stem Cell and ERT  
Therapeutics**

**CHAIRPERSONS:- Professor AJS  
Bhanwer & Dr Vineet Mishra**

2:00-2:25 PM Mitochondrial Donation by Stem Cells:  
Potential for Novel Therapeutics -  
**Dr Anurag Agrawal**

2:25-2:50 PM Stem Cell Transplantation: Our  
Experience at Apollo Group of hospitals  
- **Dr Chirag Shah**

2:50-3:15 PM Perspective of Stem Cell Research and  
Therapy in Diabetes - **Dr Sarita Gupta**

**Valedictory Function**

Tea

