CURRICULUM VITAE

- Name : Dr. Shubha Rajendra Phadke.

- . Qualification : M.B.B.S. (1982)

M.D. (Pediatrics) 1986
DM (Medical Genetics) 1992

- **IMPORTANT RESPONSIBILITIES / ACHIEVEMENTS**

- **Founder President of Society for Indian Academy of Medical Genetics [SIAMG] <http://iamg.in/>**
- **I C Verma Award for Excellence in Research [2016]**
- **Author of a book , 'Genetics for Clinicians'**
- **Editor of GENETICS CLINICS, a three monthly publication of SIAMG <http://iamg.in/genetic_clinics/index.php>**
- **On the editorial board of American Journal of Medical Genetics**
- **On the editorial board of Molecular Genetics & Genomic Medicine**
- **On the editorial board of Indian Pediatrics**
- **Founder member of Indian Society of Inborn Errors of Metabolism**
- **Founder member of Society of Fetal Medicine (India)**
- **First DM in Medical Genetics from India**
- **Reported 10 new malformation syndromes**
- **Three new syndrome entries in OMIM [**607539, 613343, 617927]
- **Recipient of Hargobind Foundation Fellowship for training in USA**
- **Recipient of International Scholarship of Clinical Genetics Society of UK**
- **On the advisory board of Journal of Fetal Medicine**
- **References cited in London Dysmorphology Database – 48**
- **References cited in OMIM -27**
- **Launched state government funded Newborn Screening Program for Uttar Pradesh since 2015 [more than 60000 babies screened]**

- **Running Thalassemia hypertransfusion program since 1989 [Funding from Uttar Pradesh government since 2016]**
- **Hemophilia management program funded by Uttar Pradesh government for last 10 years**
- **Contributed to identification of causative genes for 5 monogenic disorders [OMIM 607539, 615866, 617612 & IFT57, NCAPD2]**

- **RESEARCH PROJECTS**

[15 projects as Principle Invetigator, Ongoing -4]

1. Human Genetic Disorders (ICMR Project) (Complete) [worked as a research officer]
    a. Anthropometric measurements of complex area of development, which are often used in diagnosis of human genetic disorders.
    b. Double blind randomized trail of periconceptional vitamin supplementation for prevention of Neural Tube Defects
2. Radiological and Anthropometric study of Handigodu disease: A new variety of spondyloepimetaphyseal dysplasia (ICMR project) complete. [worked for data analysis]
3. Feasibility of introducing genetic screening in National Family Welfare Programme (ICMR Project). (Complete) [worked as a research officer]
4. Control of Thalassemia by Antenatal screening (ICMR Project). Complete [worked as a research officer]
5. Molecular Analysis of Premutation, Mutation and Mosaicism in Fragile X Mental Retardation Families (Department of Science & Technology, Project, Principle Iinvestigator - Dr. S.R. Phadke). Rs. 5,75,000.00/- from 5.9.1998(Completed)
6. Study of connexin 26 mutations in nonsyndromic deafness in Indian population. (Intramural project: Principle Iinvestigator. Dr. S.R. Phadke (Rs. 1,25,000/-) from 15.5.2001 (Completed)
7. Role of Biotechnology awareness program in prevention of genetic disorders among rural women. Phase I trial (Completed). Phase-II trial. Department of Biotechnology (Completed)
8. Point mutation/microdeletions and SMN (T/C) ratio in spinal muscular atrophy: Phenotype correlation and carrier analysis. (Intramural project) (Co-investigator) (completed)
9. Genetic Studies In Prader Willi & Angelman Syndrome. Sanctioned intramural project, 2005, (Co- Investigator) (Completed)
10. Creation of Genetic disease registry, DNA banking and EBV transformed cell lines from informative families of rare genetic disorders. ICMR, (Principle investigator) Rs 76 lacs. Started from July 2007 – for 3 years (Completed).

11. ICMR training course in Medical Genetics and Genetic counseling. Indian Council of Medical Research – Rs 3.45 lac (Completed)
12. 'Warfarin Dosing in Relation to CYP2C9 and VKORC Polymorphism in Indian Population' Intramural [Principle Iinvestigator], Rs 2.5 lac, Sanctioned on 4-5-10, period for 2 years. (Completed)
13. 'To create a newborn screening program for preventable causes of mental retardation, and create awareness about it among doctors and women of rural Uttar Pradesh.' A project by Department of Biotechnology. Sanctioned in 17-05-2010 for Rs 82.85 lakh for 3 years & 6 months [Principle Iinvestigator] (Completed)
14. Mutation analysis of COL1A1 and COL1A2 genes in Indian patients with osteogenesis imperfecta. Indian Council of Medical Research, sanctioned on 29-3-11, Rs 9,30,888/- (Completed) [Principle Iinvestigator]
15. Centre for Molecular Medicine: Indian Council of Medical Research. Rs 499 lac, for 5 years. Sanctioned on 21-3-12. (Completed) [Principle Iinvestigator]
16. Evaluation of utility of cytogenetic microarray in detection of etiology of Prenatally detected malformation Rs 3 lac. Intramural. 1st Nov 2012 to 31st oct 2014 (Completed) [Principle Iinvestigator]
17. Multicentric Collaborative Study of the Clinical, Biochemical and Molecular Characterization of Lysosomal Storage Disorders in India. Principle Iinvestigator – Shubha Phadke. Indian Council of Medical Research (Rs 31.3 lac for 3 years- Sanctioned on 10 th Dec 2014 - COMPLETE)
18. Genomic studies into limb malformations and related syndromes. Department of Biotechnology. Sanctioned on 03/03/2015 for 3 years. Rs 97,97,160. Principle Iinvestigator – Shubha Phadke. (Completed)
19. **The Indian Movement Disorder Registry and Biobank: Clinical and Genetic Evaluation of Movement Disorders in Indian Patients" Sanctioned by Department of Biotechnology for 3 years in Oct 2018 (Principle Iinvestigator – Shubha Phadke) (Rs. 4755520-00)**
20. **Training of in-service Clinicians from Government Hospitals and Outreach Program for Aspirational Districts",funded by Department of Biotechnology – 3 years from 9th May 2019 (Rs 12364000.00) [Principle Iinvestigator]**

21. **To study sequence variations in genes involved in chromosome / chromatid separation including cohesin – condensing complex, kinetocore complex and centromeric proteins in mothers of individuals with trisomy 21 to find out the genetic factors predisposing non-disjunction – Intramural project [Principle Iinvestigator]- Approved 2018 – Rs 5 lac**

22. **Study of genotypes and phenotypes of autosomal recessive osteogenesis imperfecta and search for new genes in patients osteogenesisimperfecta with no mutation in known genes – Sanctioned by Indian Council of Medical Research, sanctioned on 27th August 2019, Rs 21,57,100 for one year [Principle Iinvestigator]**

- **WORKSHOPS AND CONFERENCES ATTENDED**

1. Workshop on Human cytogenetic at the Division of Human Genetics. Department of anatomy, St. John's Medical College, Bangalore from 25th June to 28th July 1990.
2. India-Japan International workshop on DNA diagnostics held at the Department of Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow from Jan 15th to Jan.22nd, 1994. Participated as technical organizer for the demonstration of Fluorescent in Situ Hybridization (FISH) technique.
3. Annual conference of Indian society of Human Genetics 9-10 Jan. 1997 at Bangalore.
4. Annual conference of Indian Society of Human Genetics, 9-10 Jan. 1998 at Nagpur.
5. XXXVII National conference of Indian Academy of Pediatrics, 8th to 11th Feb. 2001 at Patna.
6. International congress of Human Genetics, May 15th-19, 2001 at Vienna, Austria.
7. 5th National Conference of Indian Society of Prenatal Diagnosis and Therapy. 18th-20th, Jan. 2002 at PUNE.
8. 43rd Short Courses in Medical and Experimental Mammalian Genetics at Bar Harbor USA 15th to 26th July 2002.
9. WHO Intercountry consultation on identifying regional priorities in the Area of Human Genetics in SEAR at Bangkok, Thailand 23-25th September 2003 as an invited expert.
10. VIth National Conference of Indian Society of Prenatal Diagnosis and therapy, 23rd Jan to 5TH Jan 2002 at Pune.
11. VIIth National Conference of Indian Society of Prenatal Diagnosis AND therapy, 23rd Jan to 5th Jan 2004 at Ahmadabad.
12. 41st annual National conference of Indian Academy of Pediatrics, 8th Jan to 11th Jan 2004 at Chennai.
13. National conference of Indian Academy of Pediatrics; 6th Jan TO 9th Jan 2005 at Kolkota.
14. International conference of Inborn Errors of Metabolism, 28th Sept to 2nd Oct 2005, at New Delhi. Multicentric Task Force Meeting on Inborn Metabolic Diseases , 5th May 2004 at Banglore.
15. Clinical and Laboratory Approach to Inherited Metabolic Disorders, 21st to 23rd July 2005, at New Delhi.
16. ISLH 2007 [International symposium on technological innovations in laboratory hematology] at Miami, USA; May 8- 11, 2007-06-27.
17. Indo US conference of Genetics Chapter of IAP, dec 2007, at Sir Gangaram Hospital, New Delhi.
18. 'Trends in Human Genetics" Symposium organized by DST at Puri, Orissa on 20 -22 nd August , 2007.
19. PEDICON 2007, AT Bhuwaneshwar, Orissa, 17th TO 20thJan 2008
20. ISPAT 2007 Chennai , 23rd to 25th Nov 2007.
21. HUGO – 13TH Human Genome meeting at Hyderabad from 27th September to 30th September, 2008.
22. North India Conference on Fetal Medicine and Prenatal Diagnosis, at AIIMS, New Delhi, 16th nov 2008.

23. Spring Conference of Clinical Genetics Society, London, UK, 12th March 2009.
24. Fourth International Conference on Birth Defects and Disabilities in Developing World, New Delhi, 4 – 7th October 2009.
25. 12 th Annual Asia Symposium on Lysosomal Storage Disorders, Taipei, 9th to 11th October 2009.
26. Annual conference of American College of Medical Genetics, at Vancouver, Canada from 15th March 2011 to 20th March 2011.
27. Twelfth ICHG and annual conference of American Society of Human Genetics at Montreal, Canada, 11th to 15th Oct 2011
28. Expert Group Meeting on Birth Defects in WHO SEAR ' from 13th to 15th Dec 2011 at AIIMS, New Delhi.
29. GENECON 2012 at Raipur, on 1st and 2nd Dec 12
30. Indo US Symposium on Disorders of Developing Brain' at Manipal, 27th - 28th Oct 12
32. International conference on 'Next revolutions in genetics and genomics: Applications in health and diseases' organized by Indo-UK genetic education forum, Sir Ganga Ram Hospital and Emory University, USA at New Delhi (January 2013).
33. "Current Trends in Genetic and Genomic Medicine" 31st January 2013, At Dr.RMLIMS, Lucknow.
34. 9th Annual Symposium of Ranbaxy Foundation on "Gains of Genomic Research in Biology and Medicine" on February 4, 2013
35. 'International conference on inborn errors of metabolism & 2nd national conference of ISIEN' at New Delhi on 5th -7th April 2013
36. 'Fabry disease: Diagnosis & ERT' in
37. 'Genetics of Epilepsy' in International conference on Cerebral Palsy and Developmental Medicine, 6th to 10th March 13 at Lucknow.
38. 'Newborn Screening in India: Why and How' in the North Zone conference of Indian Society of Perinatology and Reproductive Biology' in Lucknow on 6th Feb 13
39. 2nd International Congress of Society of Fetal Medicine, on 31st August 2013 at Hyderabad
40. **The** 8th Asia Pacific **-** Regional Meeting of the International Society for Neonatal Screening, at Jawaharlal Auditorium, All India Institute of Medical Sciences **(**AIIMS**),** New Delhi, INDIA; September 27th – 29th 2013
41. MAHAPEDICON 2013, annual conference of Maharashtra API, on 19th Oct 13 at Nagpur
42. A workshop on "Maternal Fetal Medicine" from 25th – 27th November 2013, In the department of Obstetrics and Gynecology in King George's Medical College, Lucknow
43. 8TH Asia Pacific Regional Meeting of International Society of Newborn Screening, 27th, 28th, 29th Sept 13, in New Delhi
44. MEDGENCON 2014, 1st March 2014 at Banglore

45. One-Day state level symposium: "Exploring Statistics Applications in Diverse Fields" at Nagpur, 7th Dec 13
46. International conference 2014 ACMG Annual Clinical Genetics Meeting, March 25 to 29, 2014 at Nashville, Tennessee, USA
47. INDO-US Symposium on Genomic insights into Human Morphogenesis: Prenatal, Postnatal and Molecular Dysmorphology and First Annual Meeting of Society for Indian Academy of Medical Genetics in Hyderabad, India from November 7th to 9th 2014.
48. 'Genetics for Clinicians' at Kasturba Medical College, Manipal on 5th Dec 2014
49. Annual Conference of Indian Society at Mumbai on 28- 29th Jan 2015
50. 11th ISUOG Intenational Symposium, New Delhi, 1st to 3rd May 2015
51. Indo US Conference : Realizing the Potentials of Rare Disorders in India', at United Services Institutions, New Delhi on 7th & 8th Sept 2015
52. The UW Mendelian Data Analysis Workshop from 10th August 15 to 15th August 15 at University of Washington, Seattle, USA
53. "Recent Advances in Rare Disease: Gaucher disease as a model **(**RARD **2017)"** May 18-20, 2017 in Moscow, Russia.
54. An international conference on RASopathies, Crowne Plaza, Kochi from 27th Nov to 29th Nov, 2017
55. Manipal Genetics Update on Genomics of Neurodevelopmental Disorders,from 9th and 10th February 2018 at Kasturba Medical College, Manipal
56. PEDICON 2018, 55th Annual Conference of Indian Academy of Pediatrics, from 4th to 7th January 2018, held at Nagpur
57. 2nd conference in the series Recent Advances in Rare Disease – RARD2018 (Frequently misdiagnosed hereditary disorders (FREMIDIS) – multidisciplinary translational research affects global clinical impact) May 3rd-5th, 2018: New Delhi, India
58. Rare Disease Update 2018-UP, organized by Indian Society of Inborn Errors of Metabolism, held on 28 July 2018, at Hotel Fairfield by Marriott, Lucknow
59. ASHG 2018, Annual Conference of American Society of Human Genetics, at San Diego, USA, from 16th Oct 2018 to 20th Oct 2018
60. ISPD 23rd International Conference on Prenatal Diagnosis *Saturday, 7 September 2019 – Wednesday, 11 September 2019* MAX Atria At Singapore EXPO, Singapore
61. Indo-US Symposium on Genetic Neuromuscular Disorders & Sixth Annual Conference of the Society for Indian Academy of Medical Genetics (SIAMGCON 2019) on 21st to 23rd Nov 2019
62. Manipal Genetics Update VI on Marfan syndrome and other aortopathies held on 14th-15th February, 2020 at Kasturba Medical College, Manipal.

- **Hospital visited, visiting scientist-fellowship:**

- Service de Medicine et de Biologie foetales (Department of Fetal Medicine). Institute de Peuriculture de Paris. PARIS from 20th, April to 5th May, 1991.
- Visited University of Washington, Seattle USA from 12th April to 9th July 1994 on Hargobind foundation Fellowship.
- Departments of Clinical Genetics, cytogenetics, St Mary's children's Hospital, Manchester. June, 2006
- Wilinks Metabolic lab, Manchester, June 2006
- May -June 2007, Pediatric Genetics Department, Shands Medical Centre, Gainesville, USA
- Guy's Hospital, London, as a recipient of International Scholarship of Clinical Genetics Society of UK
- Human Genetics, University of Michigan, Ann Arbor, MI, USA, 31ST March14 to 12th April 14

- . INVITED LECTURES INTERNATIONAL CONFERENCES

1. "Malformation Syndromes: Our experience**"** Spring Conference of Clinical Genetics Society, London, UK, 12th March 2009.
2. "Genetics of Short Stature" in Fourth International Conference on Birth Defects and Disabilities in Developing World, New Delhi, 4 – 7th October 2009.
3. "Genetic Screening" in Fourth International Conference on Birth Defects and Disabilities in Developing World, New Delhi, 4 – 7th October 2009.
4. 'Clinical Genetics: Indian Scenario' at Twelfth ICHG and annual conference of American Society of Human Genetics at Montreal, Canada, 11th to 15th Oct 2011
5. 'Genomic Techniques and Consanguinity in Rare Disorders' in MEDLAB [Arab Health Conference] on 26th Jan 2016, at Convention centre, Dubai, UAE
6. Newborn screening in India: Experience from Pilot Initiatives (UP Experience-DBT project). in 8TH Asia Pacific Regional Meeting of International Society of Newborn Screening, 27th, 28th, 29th Sept 13, in New Delhi

- PUBLICATIONS:

(Note: * - References cited in London Medical Databases -48,

 # - Figures contributed to London Medical Databases -10 cases,

@ - References cited in OMIM – 22

$ - <u>Papers related to prenatal diagnosis - 27</u>

1) R V, **Phadke S R** (1990-1991) Spinal dysraphism in achondroplasia. <u>Pediatr</u> Phadke <u>Neurosurg</u> 16: 32-34.
2) Sharma A K, **Phadke S R** (1991) Porencephaly : A possible complication of chorionic villus sampling. Indian Pediatr 28: 1061-1063.
3) Naveed M, **Phadke S R**, Agarwal S S (1992) Sociocultural problems in Genetic counseling. J Med Genet 29: 140 (letter).

4) Sharma A K, **Phadke S R**, Chandra K, Upreti M, Khan E M, Naveed M, Agarwal S S (1992) Overlap between Majewski and Hydrolethalus syndromes : A report of two cases. Am J Med Genet 43: 949-953.*
5) **Phadke S R**, Sharma A K, Agarwal S S (1993) A report of Freeman Sheldon syndrome with bilateral simian crease and malpositioned second toes. Indian Pediatr 30: 91-93.
6) Sharma A K, **Phadke S R**, Agarwal S S (1993) Beemer syndrome. Am J Med Genet 46: 345.(letter)*#
7) **Phadke S R**, Sharma A K, Agarwal S S (1993) A new syndrome of multiple joint dislocations with metaphyseal dysplasia, natal tooth and lymphoedema. Clinical Dysmorphology 2: 264-268.*
8) Sharma A K, **Phadke S R** (1993) CVS and porencephaly. Prenatal diagnosis. 13: 1077 $.
9) Sharma A K, Halder A, **Phadke S R**, Agarwal S S (1994) Preaxial brachydactyly with abduction of thumbs and hallux varus – A distinct entity. Am J Med Genet 49: 274-277.*@
10) **Phadke S R**, Sharma A K, Halder A, Pandey R, Bhatia V L, Agarwal S S, (1994) GAPO syndrome in a child without dermal hyaline deposits. Am J Med Genet 51: 191-193.*
11) Sharma A K, **Phadke S R** (1994) Midline malformation syndrome. Am J Med Genet 50: 304.
12) Sharma A K, **Phadke S R** (1994) Another case of spondylocostal dysplasia and severe anomalies: A diagnostic counseling dilemma. Am J Med Genet 50: 383-384.*
13) Sharma A K, **Phadke S R**, Agarwal S S (1994) The clinical value of a limited fetal autopsy. Australian and New Zealand J of Obstet Gynaecol 34: 1-3 $.
14) **Phadke S R**, Sharma A K, Agarwal S S (1994) Anophthalmia with cleft palate and micrognathia: a new syndrome. J Med Genet 31: 960-961.*
15) Agarwal S S, **Phadke S R**, Phadke R V, Das S K, Singh G K, Sharma J P, Teotia S P S, Saxena B N (1994) Handigodu disease : A radiological study. A new variety of spondyloepi (meta) physeal dysplasia of the autosomal dominant type. Skeletal Radiol. 23: 611-619.
16) Halder A, Sharma A K, **Phadke S R**, Jain A, Agarwal S S (1994) OEIS complex with craniofacial anomalies defect of blastogenesis ? Am J Med Genet 53: 21-23.*
17) Sharma A K, Jain A, **Phadke S R**, Srivastava S (1994) Prenatal diagnosis of Robert syndrome. Indian Pediatr. 31: 1261-1264.$
18) Sharma A K, Halder A, **Phadke S R**, Agarwal S S (1994) Marshall Smith syndrome – A distinct entity. Indian Pediatr 31 (8) : 1098-1100.
19) Sharma A K, **Phadke S R** (1994) Jarcho Levine syndrome. A case report. Indian Pediatr. 31 (6) : 707-708.
20) Sharma A K, Halder A, **Phadke S R** (1994) Postmortem radiography of perinatal deaths: an aid to genetic counseling. Indian Pediatr 31(6): 702-706 $.
21) Dhanda S, **Phadke S R**, Agarwal S S (1996) Lessons from fibroblast growth factor receptor mutations in craniosynostosis syndromes. The J of Clin Genet and Tribal Research. 1(3) : 176-180.
22) **Phadke S R**, Pahi J, Phadke R V, Pradhan S, Agarwal S S (1997) Importance of etiologic diagnosis of hydrocephalus as illustrated by a case of Walker Warburg syndrome. Indian Pediatr 34(4): 1037-1038.
23) Gulati R, **Phadke S R**, Agarwal S S (1997) Associated malformations in the family of a patient with Meckel syndrome: Heterozygote expression? J Med Genet 34(11): 937-938.*@

24) Dhanda S, **Phadke S R**, Agarwal S S (1997) Acromesomelic dwarfism: Report of a family with two affected siblings. Indian Pediatr 34(4): 1127-1130.
25) Agarwal S S, **Phadke S R**, Fredlund V, Viljoen D, Beighton P (1997) Mselini and Handigodu Familial Osteoarthropathies: syndromic identity. Am J Med Genet 72: 435-439.*
26) **Phadke S R**, Gulati R, Agarwal S S (1998) Further delineation of a new (Van Den Ende- Gupta) syndrome of blepherophimosis, contractual arachnodactyly and characteristic face. Am J Med Genet 77: 16-18 *#@
27) Pahi J, **Phadke S R**, Halder A, Gupta A, Pandey R, Agarwal S S (1998) Does autopsy of antenatally diagnosed malformed fetus aid in genetic counseling. National Med J India 11: 169-170 $
28) **Phadke S R**, Gupta A, Pahi J, Pandey A, Gautam P, Agarwal S S, (1999) Malignant recessive osteopetrosis. Indian Pediatr 36: 69-74.
29) Singh K, **Phadke S R**, Agarwal S S (1999) Mandibuloacral dysplasia: Indian patient with severe bony changes. J Association Physn India 47: 833-834.
30) **Phadke S R**, Gautam P (1999) Complex camptopolydactyly : an unusual hand malformation. Am J Med Genet 83: 191-192.*@
31) Pradhan M, **Phadke S R**, Jain S, Agarwal S S (1999) Pachygyria / Hypogenitalism: A monogenic syndrome. Am J Med Genet 87: 254-257.*#
32) **Phadke S R**, Pahi J, Pandey A, Agarwal S S (1999) Oral- facial – digital syndrome with acromelic short stature : a new variant overlap with Ellis Van Creveld syndrome. Clinical Dysmorphology 8:185-188*#.
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36) Gautam P, **Phadke S R** (2000) Fetal brain disruption sequence. Indian Pediatr 37: 662- 664#.
37) **ICMR** Collaborating center & central technical co-ordinating unit, ICMR, New Delhi(2000) Multicentric study of efficacy of periconceptional folic acid containing vitamin supplimentation in prevention of neural tube defects from India. Indian J Med Res 112: 206-211
38) **Phadke S R** , Halder A(2000) Fluorescence In Situ Hybridization: A novel method to study chromosomes and genes. Perinatology 2(4): 203-210
39) Gupta A, **Phadke S R**(2001) Bowen Conradi syndrome in an Indian infant: A first nonHutterite case. (Letter) Clinical Dysmorphology 10:1-2*@
40) **Phadke S R**, Agarwal S S (2001) Adverse effects of genetic counseling on women carriers of disease : The Indian perspective. National Med J of India 14(1): 47-49
41) Choudhury N, **Phadke S R** (2001) Transfusion transmitted diseases. Indian J Pediatrics 68:957-958
42) Puri R D, **Phadke S R** (2001) Molecular diagnosis of monogenic disorders. I of Internal Medicine of India 4(4):174- 180
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50) Panigrahi I, **Phadke S R**, Agarwal S S (2002) Mental retardation, ptosis and polydactyly: A new autosomal recessive syndrome? Clinical Dysmorphology 11:289-292*#
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52) Saxena A, Phadke SR. Feasibility of thalassaemia control by extended family screening in Indian context. J Health Popul Nutr. 2002 Mar;20(1):31-5.

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54) Pandey U B, **Phadke S R**, Mittal B (2002) Molecular screening of FRAXA and FRAXE in Indian patients with unexplained mental retardation. Genetic Testing 6(4): 335-339
55) **Phadke S R**, Thakur S (2002) Prenatal diagnosis of iniencephaly and alobar holoprosencephaly with trisomy 13 mosaicism: a case report. Prenatal Diagnosis 22: 1240-1241 $.
56) **Phadke S R**, Agarwal S, Agarwal S S (2002) Medical genetics education in India (letter) . National Medical J of India 15(6): 363.
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- BOOK AND CHAPTERS IN BOOK

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- PAPERS/POSTERS PRESENTED IN THE CONFERENCE

1. Role of fetal autopsy in counseling for prenatally diagnosed malformed fetuses. In Annual conference of Indian Society of Human Genetics at Bangalore.
2. Asphyxiating thoracic dysplasia. In Annual Conference of Indian Society of Human Genetics at Nagpur – 1998
3. Genetic Counseling in Indian Milieu. International Conference of Human Genetics at Vienna, Austria-2001.
4. Counseling of prenatally diagnosed malformation of uncertain prognosis: A dilemma. Annual conference of Indian Society of Prenatal Diagnosis & Therapy at Pune-2002.
5. "C" Syndrome phenotype in a child with partial trisomy of 13q. At 43rd Short course in Medical & Experimental Mammalian Genetics at Bar Harbor, USA 15th July to 26th July, 2002
6. " Cytogenetic Microarray in the evaluation in evaluation of Intellectual disability' in Annual conference of American College of Medical Genetics, at Vancouver, Canada from 15th March 2011 to 20th March 2011
7 'Clinical genetics: Indian Scenario" in Twelfth ICHG and annual conference of American Society of Human Genetics at Montreal, Canada, 11th to 15th Oct 2011
8. 'Homozygosity Mapping and Gene Sequencing Identifies a Novel Mutation in an Indian patient with Warburg Micro Syndrome in 'International conference 2014 ACMG Annual Clinical Genetics Meeting, March 25 to 29, 2014 at Nashville, Tennessee, USA'
9. Genomic techniques and consanguinity in rare disorders ' in Molecular Diagnostics Conference [26th Jan 16 to 27th Jan 16] at MEDLAB Congress in Dubai
8. "Recent advances in rare disease: Gaucher disease as a model (RARD 2017)"

May 18-20, 2017 in Moscow, Russia. Presented a poster on 'Molecular Diagnosis of Mucopolysacchardosis II (Hunter Syndrome) by Sequencing of IDS gene' and got award

9. 'Homozygosity Stretches Around Homozygous mutations for Autosomal Recessive Disorders in patients from Non –Consanguineous Families from India: Inheritance by Descent is common' in ASHG 2018, at San Diego, USA, from 16th Oct to 20th Oct 2018
10. 'Rare neurodegenerative disease of prenatal onset: Fourth case with PNPLA8 related autosomal recessive disorder,' in ISPD 23rd International Conference on Prenatal Diagnosis Saturday, 7 September 2019 – Wednesday, 11 September 2019 MAX Atria At Singapore EXPO, Singapore
11. 'Spectrum of malformations detected before 16 weeks by ultrasonography' in ISPD 23rd International Conference on Prenatal Diagnosis Saturday, 7 September 2019 – Wednesday, 11 September 2019 MAX Atria At Singapore EXPO, Singapore

• ORGANIZATION OF CONFERENCES/ WORKSHOP

1. National Workshop on Carrier detection & antenatal diagnosis of Thalassemia (14/15 Nov. 1997) (Treasurer).
2. International CME & update on prenatal diagnosis and management of genetic disorders. (10.02.1998) (Organizing Secretary)
3. Summer Camp for Hemophilia children at Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow 10.03.99 to 13.03.99.
4. Workshop on Down Syndrome- 24.09.99 (Organizing Secretary)
5. Indo-European Seminar cum Workshop in Molecular Cytogentics (06-to 09.12.99) (Treasurer)
6. ICMR Course on Genetic Counseling (18th to 30th, Sept. 2000) (Course Convenor)
7. Camp for Hemophilia children 9th to 30th Dec. 2000 (Organizer) at SGPGIMS.
8. Foundation Day Cultural program on 14th December 2000 at SGPGIMS.
9. 2nd ICMR course on Genetics Counseling (10th to 21st, April 2001) (Course coordinator)
10. 3rd ICMR course on Medical Genetics & Genetic Counseling (11th to 22nd Dec. 2001) (Course Convenor)
11. 4th ICMR course on Medical Genetics & Genetic Counseling (9th to 20th, April 2002) (Course Convenor)
12. Camp for Hemophilia Children at Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow 25th –28th, Dec. 2002.
13. 5th ICMR course on Medical Genetics and Genetic Counseling. 9th Feb. to 21stFeb. 2004.
14. Sixth ICMR course on Medical Genetics and Genetic Counseling . 11th April to 23rd April 2005
15. Seventh ICMR course on Medical Genetics and Genetic Counseling July 17th-July 22nd, 2006
16. 'GENETICS FOR PAEDIATRICIANS' A CME organized on behalf of Genetics Chapter of Indian Academy of Paediatrics, at Sir Gangaram Hospital, New Delhi, 15th and 16th Dec 2007.
17. Eighth ICMR course on Medical Genetics and Genetic Counseling. 20th July 2008 to 2nd August 2008
18. 'Ninth ICMR Course on Medical Genetics and Genetic Counseling' (2nd August 2010 to 14th August 2010)

19. **'First Indo US Symposium on Skeletal Dysplasia' at Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow on 12th and 13th Feb 2011 [In collaboration with Dr Rimoin]**
20. Workshop on DNA diagnostics 2011, in Medical Genetics, SGPGIMS, on 24th to 26th Nov 2011
21. 'Tenth ICMR Course on Medical Genetics and Genetic Counseling' from 9th to 20th August 2011
22. 'Eleventh ICMR Course on Medical Genetics and Genetic Counseling' from 29th July 12 to 11th August 2012
23. 'Twelth ICMR Course on Medical Genetics and Genetic Counseling' from 29th July 2013 to 13th Aug 2013
24. 'Thirteenth ICMR Course on Medical Genetics and Genetic Counseling' from 28th July 2014 to 9th August 2014
25. Fourteenth ICMR Course on Medical Genetics and Genetic Counseling' from 20th July 2015 to 1st August 2015
26. Fifteenth ICMR Course on Medical Genetics and Genetic Counseling' from 25th July 2016 to 6th August 2016
27. Sixteenth ICMR Course on Medical Genetics and Genetic Counseling' from 24th July 2017 to 5th August 2017
28. Seventeenth ICMR Course on Medical Genetics and Genetic Counseling' from 20th August 2018 to 1st September 2018
29. Eighteenth ICMR Course on Medical Genetics and Genetic Counseling' from 26th August 2019 to 7st September 2019
30. Indo-US Symposium on Genetic Neuromuscular Disorders & Sixth Annual Conference of the Society for Indian Academy of Medical Genetics (SIAMGCON 2019) on 21st to 23rd Nov 2019, as organizing chairperson

- Online Mendelian Inheritance in Man (OMIM) Entries of New Syndromes
  1. Camptosyndactyly, complex (607539)
  2. Handigodu disease (613343)
  3. Oro facio digital syndrome 18 [# 617927]
  4. Coffin Siris syndrome 9 [# 615866]

- REVIEWER FOR THE FOLLOWING JOURNALS
  1. Indian Pediatrics
  2. British Journal of Hematology
  3. Indian Journal of Medical Research
  4. Indian Journal of Pediatrics
  5. Am J Medical Genetics
  6. Clinical Genetics
  7. Clinical Dysmorphology
  8. Prenatal diagnosis [official journal of International Society of Prenatal Diagnosis]
  9. Indian Journal of Human Genetics
  10. Gene
  11. BMC journal
  12. Molecular Genetics and Genomic Medicine

- COMMITTEES

1. Member of Department of Biotechnology, New Delhi Task Force on Genetic Disorders, 2013, 2014
2. Alternate Chairperson of 'National Task Force on Newborn Screening for congenital hypothyroidism and Congenital adrenal hyperplasia: A Multicentric Study'- 2014
3. Member of project review committee of BMS, ICMR, New Delhi, 2013, 2014
4. Chairperson of ICMR Task Force on Rare Diseases [2018]

- INVITED LECTURES

1. "Prenatal sex determination" In workshop on "Female Feticide" 6th Sept. 1997. Organized by 'Watsalya', Lucknow
2. "Approach to a case with mental retardation" Lucknow Branch of Indian Association of Pediatrics. 1995.
3. Genetics": Indian Medical Association, Lucknow 1998
4. "Genetics in Pediatrics Practice": Lucknow Branch of Indian Association of Pediatrics. October'99
5. "Genetic Counseling for deafness", CME at Annual conference of ISHA, Post Graduate Institute of Medical Sciences, Chandigarh, 12Feb. 1999.
6. "Hemophilia common problems and management": Annual function of Hemophilia Society Lucknow 1999
7. "Hemophilia-Struggle with a lifelong problem" Keynote address at engineering college. Sultanpur on the occasion of concluding function of the Health week. April 1999.
8. "Clinical Genetics-Indian perspective: at 25th Annual conference of Indian Society of Human Genetics 9th-10th, 2000 Nagpur
9. "Hemophilia-A lifelong struggle" A lecture for school children in St. Paul's School on the occasion of Hemophilia Day Celebration 6th April 2000.
10. "Genetics in Clinical Practice" A lecture at Balrampur Hospital on their Foundation day 2001.
11. "Preconceptional community Training-Genetic aspects" Guest Lectures at XXXVIII National Conference of Indian Academy of Pediatrics at Patna, 7th-10th Feb. 2001.
12. "DNA Diagnosis of Genetic Disorders" Guest Lectures at XXXVIII National Conference of Indian Academy of Pediatrics at Patna, 7th-10th Feb. 2001.
13. "Dysmorphology Diagnosis in fetus & a neonate" Guest Lectures at XXXVIII National Conference of Indian Academy of Pediatrics at Patna, 7th-10th Feb. 2001
14. Primary Prevention of Thalassemia major. The need & the Feasibility" At a Seminar "Thalassemia Control A Challenge organized by Thalassemia society, Lucknow Obstetrics & Gynecology Society of Lucknow & SEARCH on 6.5.2001.
15. "Genetic Counseling in Prenatal Diagnosis" Guest Lecture at VI National Conference of ISPAT on 18th Jan 2002 at Pune

16. "Prenatal diagnosis of chromosomal disorders" Lecture during workshop on "Prenatal & Postnatal Diagnosis of Genetic Disorders using molecular methods" at All India Institute of Medical Sciences, New Delhi Jan. 2002.
17. "Molecular techniques for prenatal diagnosis of monogenic disorders" in Xith All India Congress of Cytology & Genetics at Mahatma Gandhi Institute of Medical Sciences, Sewagram on 28th Oct 2002
18. "Genetics in clinical practice" at CME, Indian Association of Pediatrics, Gorakhpur branch on 25th Dec 2002
19. "Prenatal diagnosis of hemophilia" at Hemophilia Workshop, by Hemophilia Society, Nagpur, on 19th Jan 2003
20. "Genetics in clinical practice" at Postgraduate students association, Government Medical College, Nagpur
21. "Congenital malformations: before and after birth" GENTICON by Indian Association Medicine, Rajkot on 23rd Feb 2003
22. "Cytogenetic techniques & applications" GENTICON by Indian Association Medicine, Rajkot on 23rd Feb 2003
23. "Prenatal cytogenetic diagnosis" workshop on Genetics at departments of Anatomy & Pediatrics, All India Institute of Medical Sciences, New Delhi, March 2003
24. "Congenital malformations" in Pediatrics Update 2003 at Manipal College of Medical Sciences on 18th Oct 2003.
25. "Teaching and training in human genetics" in WHO Intercountry consultation on identifying regional priorities in the area of Human Genetics in SEAR, at Bangkok, Thailand 23rd -25th Sept 2003
26. "Congenital malformations & genetic metabolic disorders: Indian scenario" in WHO Intercountry consultation on identifying regional priorities in the area of Human Genetics in SEAR, at Bangkok, Thailand 23rd -25th Sept 2003
27. "Prenatal diagnosis of CNS malformations" in Neuropedicon 200314th Annual conference of Indian Society of Pediatric Neurosurgery at Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, on 6th Nov 2003
28. "Genetic counseling – Illustrative cases" in Pediatric Multispeciality Update at Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, on 15th nov 2003
29. "Molecular diagnosis in clinical genetics: scope and limitations" lecture in Pediatric subspeciality symposium in PEDICON 2004, on 10th jan 2004
30. "Counseling after diagnosis of genetic metabolic liver disease" in Annual conference of study of liver diseases at Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, on 14th march 2004
31. "Prenatal diagnosis of cytogenetic disorder" lecture in Workshop on genetic techniques at the Departments of Anatomy & Pediatrics, All India Institute of Medical Sciences, New Delhi, on 7th march 2004
32. "Molecular medicine: Diagnostic and therapeutic potentials" Lecture in National Workshop on Molecular Techniques in Transplant Biology, Department of Medical

Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, 30th sept 2004

33. "Imprinting and DNA methylation" Lecture in National Workshop on Molecular Techniques in Transplant Biology, Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, 1st oct 2004
34. "Ethical issues in fetal medicine and Perinatology" Lecture in 01 course of Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow , 8th dec 2004
35. "Clinical cytogenetics and role of FISH" in Workshop on techniques in genetic toxicology at Indian Institute of Toxicological Research , Lucknow on 12th dec 2004.
36. "Congenital Malformations :Diagnosis & counseling" Annual conference of Indian Association o Pediatrics – PEDICON 2005, at Kolkota,8th jan 2005.
37. "Diagnosis and prevention of congenital malformations" R N Ganguli public forum in 21st conference of Indian society of perinatology and reproductive biology, in Lucknow , 6th feb 2005
38. "Genetic counseling and prenatal diagnosis for inborn errors of metabolism" in First Workshop on Clinical and Laboratory Approach to Inborn Metabolic Disorders' AIIMS, New Delhi, 21st july 2005
39. "Pedigree drawing" in S R Naik memorial symposium on Computers in biomedical Science, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow. 11 TH Sept 2005.
40. "Molecular techniques in cancer: Research and management" in Department of Radiotherapy, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow. 23rd sept 2005.
41. "Congenital malformations: A gateway to developmental genetics" In 74th Annual meeting of Society of biological chemists, India. Central Drug Research Institute, Lucknow. 8th nov 2005.
42. "Congenital malformations: USG and autopsy correlation" in 8th Conference of Indian Society of Prenatal Diagnosis and Fetal Therapy at New Delhi. 19th February 2006
43. "Malformation Syndromes: Indian scenario" in Institute of Medical Genetics, University of Wales, College of Medicine, Heath Park**,** Cardiff, UK
44. "Genetics in clinical practice" in students' workshop on Human genetics in G R Medical college, Gwalior on 1st August 2006
45. "USG in prenatal diagnosis of malformations" in Conference on antenatal diagnosis, at Vivekanand polyclinic, Lucknow on 26th August 2006
46. "Role of perinatal autopsy in genetic counseling" in Workshop on Prenatal Medicine in Deenanath Mangeshkar Hospital and Research Centre, Pune on 6th October 2006.
47. "DNA diagnosis of monogenic disorders" in Workshop on Prenatal Medicine in Deenanath Mangeshkar Hospital and Research Centre, Pune on 6th October 2006.
48. "Common presentations or rare genetic disorders" in PEDICON 2007, Mumbai on 14th jan 2007
49. "Skeletal Dysplasia" in PEDICON 2007, Mumbai on 12th jan 2007

50. "Five lectures in workshop" of CLINICAL GENETICS at Rainbow hospital, Hyderabad.
51. ''Malformation Syndromes: Indian scenario' on 5th June 2007, in Pediatric Genetics Department, Shands Medical Centre, Gainesville, USA
52. **"**Importance of Family history" 'GENETICS FOR PAEDIATRICIANS' A CME organized on behalf of Genetics Chapter of Indian Academy of Paediatrics, at Sir Gangaram Hospital, New Delhi, 15th and 16th Dec 2007
53. "Enzyme replacement therapy for Fabry disease" in 'GENETICS FOR PAEDIATRICIANS' A CME organized on behalf of Genetics Chapter of Indian Academy of Paediatrics, at Sir Gangaram Hospital, New Delhi, 15th and 16th Dec 2007
54. "DNA diagnosis in clinical practice" in ISPAT 2007 Chennai , 23rd to 25th Nov 2007
55. "Genetics of artificial reproductive techniques" in ISPAT 2007 Chennai , 23rd to 25th Nov 2007
56. "Chromosomal abnormalities: How to choose the best test" in ISPAT 2007 Chennai , 23rd to 25th Nov 2007
57. "Dysmorphology diagnosis" in Indo Europian CME on Clinical Genetics and Dysmorphology, at CMC, Vellore on 2nd October 2008
58. "Medical Genetics: Indian scenario" in PERICON, at Ernakulum, Kerala on 5th October 2008
59. "Fetal autopsy: Indications & utility" in North India conference on fetal medicine and prenatal diagnosis , on 16th Nov 2008, at AIIMS, New Delhi
60. "Malformation Syndromes: Our experience' Spring Conference of Clinical Genetics Society, London, UK, 12th March 2009
61. 'Genetics of Short Stature' in Fourth International Conference on Birth Defects and Disabilities in Developing World, New Delhi, 4 – 7th October 2009
62. 'Genetic Screening' in Fourth International Conference on Birth Defects and Disabilities in Developing World, New Delhi, 4 – 7th October 2009
63. 'Ethical issues in in Genetics and stored tissue research' in 'ICMR-FERCAP Human subject protection course' held on 7th August 2010 in SGPGIMS, Lucknow.
64. 'Prenatal diagnosis of Thalassemia' in the monthly meeting of Hematology society of Lucknow, held at Command Hospital, Lucknow on 5th No 2010
65. 'Cytogenetic Microarray: A new tool to solve the puzzle of mental retardation' in The Molecular Cytogenetic Workshop held at Institute of Genomics and Integrated Biology, New Delhi on 23rd Nov 2010
66. 'New tools for evaluation of mental retardation' at Annual conference of ISHG at Manipal on 16th Feb 2011
67. 'Mental Retardation: An unsolved Mystery' in 'The current practice of genetics and genomic medicine' held at Apollo Hospital , New Delhi on 26th and 27th March 2011
68. 'Fetal autopsy: a must' in 'The current practice of genetics and genomic medicine' held at Apollo Hospital , New Delhi on 26th and 27th March 201

69. " Molecular Medicine in resource poor countries' in 'Synergy and diversity of Molecular Medicine' at PSG Centre for Molecular Medicine, Coimbtoor on 27th and 28th June 2011.

70. "Genetic disorders with joint problems" in National Conference of Pediatric Rheumatology on 3rd Sept 2011, at SGPGIMS, Lucknow

71. " Approach to unknown dysmorphic syndromes" in workshop "Dysmorphology: Unmystified" organized by Genetic Chapter of IAP in AIIMS, New Delhi on 4th Sept 2011

72. "Skeletal Dysplasia: Clinical Approach" in workshop "Dysmorphology: Unmystified" organized by Genetic Chapter of IAP in AIIMS, New Delhi on 4th Sept 2011

73. "DNA to disease: Genetic counseling for retinal disorders" in Retinaware 2011, on 24th Sept 2011, In Vigyan Bhawan, New Delhi

74. "Malformation syndromes in fetus" in First international Congress of Society of Fetal Medicine, at New Delhi, 1st and 2nd Oct 2011

75. "Medical Genetics Services: Need for National & Regional Networks for Genetic Services" in Expert Group Meeting on Birth Defects in WHO SEAR ' from 13th to 15th Dec 2011 at AIIMS, New Delhi

76. 'Molecular diagnostics in Clinical Practice" in Workshop on DNA diagnostics 2011, in Medical Genetics, SGPGIMS, on 24th to 26th Nov 2011

77. "Fetal Malformations:Illustrative cases" in Workshop on fetal medicine and genetics at New Delhi, by NARCHI on 14th Sept 12

78. 'Clinical approach in mental retardation' in Indo US Symposium on Disorders of Developing Brain' at Manipal, 27th -28th Oct 12

79. 'Prenatal diagnosis of Malformation syndromes' in Lucknow Ultrasound Course 2012 in Lucknow, 3rd Nov 12

80. 'Genetics of Endocrine Disorders' I 42nd Annual Conference of Endocrine Society of India, in Kolkota, 13th Dec 2012

81. "Pearls of Dysmorphology' in GENECON 2012 at Raipur, on 1st and 2nd Dec 12

82. 'Molecular cytogenetics" in GENECON 2012 at Raipur, on 1st and 2nd Dec 12

83. 'Cytogenetic microarray in evaluation of Intellectual Disability' in International conference on 'Next revolutions in genetics and genomics: Applications in health and diseases' organized by Indo-UK genetic education forum, Sir Ganga Ram Hospital and Emory University, USA at New Delhi (January 2013).

84. 'Medical Genetics: Indian Scenario' in "Current Trends in Genetic and Genomic Medicine" 31st January 2013, At Dr.RMLIMS, Lucknow.

85. 'Cytogenetic microarray in evaluation of Intellectual Disability' in 9th Annual Symposium of Ranbaxy Foundation on "Gains of Genomic Research in Biology and Medicine" on February 4, 2013

86. 'Fabry Disease' in 'International conference on inborn errors of metabolism & 2nd national conference of ISIEN' at New Delhi on 5th -7th April 2013

87. 'Fabry disease: Diagnosis & ERT' in

88. 'Genetics of Epilepsy' in International conference on Cerebral Palsy and Developmental Medicine, 6th to 10th March 13 at Lucknow.

89. 'Newborn Screening in India: Why and How' in the North Zone conference of Indian Society of Perinatology and Reproductive Biology' in Lucknow on 6th Feb 13

90. 'aCGH, NIPD, FISH, QF PCR Demystifying the New Genetic Alphabets' in 2nd International Congress of Society of Fetal Medicine, on 31st August 2013 at Hyderabad

91. 'Genetic approach to short stature' in Growth hormone symposium organized by IAP, Mumbai branch on 1st Sept 13 at B Y L Nair Children's hospital, Mumbai.

92. 'Newborn screening in Uttar Pradesh: An outreach program' in **the** 8th Asia Pacific - Regional Meeting of the International Society for Neonatal**,** at Jawaharlal Auditorium, All India Institute of Medical Sciences (AIIMS)**,** New Delhi, INDIA; September 27th – 29th, 2013

93. 'Dysmorphology diagnosis: Face speaks' in MAHAPEDICON 2013, annual conference of Maharashtra API, on 19th Oct 13 at Nagpur

94. 'Genetics: A Science of Probabilities' in State level symposium 'Exploring statistics applications in Diverse Fields' organized by Department of Statistics, Hislop college, Nagpur on 7th Dec 2013.

95. 'Sonological evaluation of fetal intracranial anatomy' in a workshop on "Maternal Fetal Medicine" from 25th – 27th November 2013, In the department of Obstetrics and Gynecology in King George's Medical College, Lucknow.

96. ': Next Generation Sequencing In The Clinic: Enabling Genomic Medicine' in Workshop on DNA Diagnostics, 'Frontiers in Medical Genetics' organized by the Department of Medical Genetics, SGPGIMS, Lucknow on 28th , 29th and 30th No 2013.

97. "Statistics in Genetics' in "Exploring Statistics Applications in Diverse Fields" at Nagpur, 7th Dec 13

98. "Malformation Syndromes in India' in INDO-US Symposium on Genomic insights into Human Morphogenesis: Prenatal, Postnatal and Molecular Dysmorphology and First Annual Meeting of Society for Indian Academy of Medical Genetics in Hyderabad, India from November 7th to 9th 2014.

99. 'Next Generation sequencing in Clinical Practice' in 'Genetics for Clinicians' at Kasturba Medical College, Manipal on 5th Dec 2014

100. 'Genomic testing in Clinical Practice'

101. 'Genetic Disorders and Consanguinity: Indian Scenario' at Annual Conference of Indian Society of Human Genetics, at Mumbai on 29th Jan 2015

102. 'Newborn Screening' in International Symposium on 'Genetics and Genomics in Modern Clinical Medicine' at King George's Medical University, Lucknow on 9th Feb 2015

103. 'When to order a new generation test' in "Genetics for the Practicing Pediatrician " 21st January 2015, Sir Ganga Ram hospital, New Delhi

104. 'Nonsyndromic Mental Retardation' in PediGen2015, at Deenanath Mangeshkar Hospital, Pune ON 15TH Feb 15

105. 'Genomic techniques in diagnostics' in National Conference on " Biotechnology and Human Welfare: New Vista' in VBS Purvanchal University, Jaunpur on 22nd March 2015

106. "Genetics: Bench to bedside' in CME on Molecular Genetics for Practicing Clinicians on 19 April 2015, Mumbai

107. "Treatment of Gentic Disorders' in CME on Molecular Genetics for Practicing Clinicians on 19 April 2015, Mumbai

108. 'Genetics in Clinical Practice' at Armed Forces Medical College, Pune, on 14th May 2015

109. 'Training in Medical Genetics in India' in Indo US Conference : Realizing the Potentials of Rare Disorders in India', at United Services Institutions, New Delhi on 7th Sept 2015

110. 'USG and Genetics' in Sixth Ultrasonography Course in Lucknow Under Aegis of Ultrasound Education & Research Foundation, 1st Nov 2015

111. 'Clinical Genomics' in Second Annual conference of Society for Indian Academy of Medical Genetics, at Jodhpur on 12th and 13th Dec 2015

112. 'Genomic Techniques and Consanguinity in Rare Disorders' in MEDLAB [Arab Health Conference] on 26th Jan2016, at Convention centre, Dubai, UAE

113. 'Hemophilia Program in UP:A Prototype of Government Supported Initiative' in Workshop to Develop Scientific Program for Research In Rare Diseases, organized by Indian National Science Academy, New Delhi on 22nd April 16

114. 'Screening for chromosomal disorders in the era of genomics' in Workshop of International Congress on Advances in ObGyn Ultrasound 2016 in Hyderabad, 6th May 16

115. 'Genetics for Obstetricians' in International Congress on Advances in ObGyn Ultrasound 2016 in Hyderabad, 7th , 8th May 16

116. 'Syndromes and Congenital Heart Disease' in International Congress on Advances in ObGyn Ultrasound 2016 in Hyderabad, 7th , 8th May 16

117. ' ' In ILLUMNATI at Armed Forces Medical College, Pune on 5th Aug 2016

118. ' In ILLUMNATI at Armed Forces Medical College, Pune on 5th Aug 2016

119. ' In ILLUMNATI at Armed Forces Medical College, Pune on 5th Aug 2016

120. 'Technological Milestones in (*Applications in*) Fetal Genetics' in FetalMed2016 on 10th September 2016, in Kolkota

121. 'Genetic Factors in Fetal Growth Restriction' in in FetalMed2016 on 10th September 2016, in Kolkota

122. "Exome sequencing in clinical practice' in Workshop on NGS in SGPGIMS Bioinformatics Centre on

123. 'Prenatal diagnosis dilemmas for mothers and Doctors ' in the National Conference of Association of Medical Women in India **(AMWI 2016**) to be held on 15th , 2016th October 2016 at Hotel Centre Point, Ramdaspeth, Nagpur.

124. 'Skeletal Dysplasias in India' in 'Update in Genetic Disorders of Bone' in Kasturba Medical College, Manipal on 1st and 2nd Dec 16

125. ‘Molecular Diagnostics in Clinical Dysmorphology: Indian Experience’ in third Annual Conference of Society for Indan Academy of Medical Genetics, at All India Instittue of Medical Sciences, New DELHI on 8th and 9th Dec 16
126. ‘Approach to neurometabolic disorders’ in IV National Conference of Indian Society of Inborn Errors of Metabolism, in Chennai, 11th to 12th Feb 17
127. ‘Genomic Diagnosis : Journey with Patients’ in Manipal Genetics Update IV on Genetic Counseling at Kasturba Medical College, Manipal [3rd and 4th March 2017
128. ‘Genetics for Clinicians- Pedigree to Genome’ in Faculty Development Program, School of Medicine, KIIT University, Bhubaneshwar, on 29th June 17
129. ‘Genetics of Multifactorial Disorders: Learning from Age Related Multifactorial Disorders’ in Faculty Development Program, School of Medicine, KIIT University, Bhubaneshwar, on 29th June 17
130. What aneuploidy screening can be offered to a woman who presents after 14 weeks’ in FetalMed 2017[13th Asia Pacific Congress of Maternal Fetal Medicine] on 2nd & 3rd sept2017 at Leela Ambiance Hotel, Gurugram, New Delhi
131. ‘Aneuploidy screening: Indian scenario’ in FetalMed 2017[13th Asia Pacific Congress of Maternal Fetal Medicine] on 2nd 3rd sept2017 at Leela Ambiance Hotel, Gurugram, New Delhi
132. ‘Learning Dysmorphology: Noonan Syndrome’ in An international conference on RASopathies, Crowne Plaza, Kochi from 27th Nov to 29th Nov, 2017
133. ‘Prenatal Growth Retardation: Diagnostic Challenges’ in Annual conference of Society for Indian Academy of Medical Genetics, on 8th and 9th Dec 2017 at Trivendrum.
134. ‘Genomic alterations in microcephaly’ in ‘Manipal Genetics Update on Genomics of Neurodevelopmental Disorders’ to be held on 9th and 10th February 2018 at Manipal.
135. ‘Genomic Diagnosis: Journey with the patients’ in PEDICON 2018, AT Nagpur, 5th, 6th and 7th Jan 2018
136. ‘Medical Genetics: Indian Scenario’ in PEDICON 2018, AT Nagpur, 5th, 6th and 7th Jan 2018
137. ‘Prenatal Diagnosis’ in PEDICON 2018, AT Nagpur, 5th, 6th and 7th Jan 2018
138. ‘Down Syndrome & More’ in Mayo Medical College, Barabanki on 21st March 2018
139. ‘Genes for Generalists’ in ACP India Chapter Annual Internal Medicine Congress 2018 at Ramada Convention Centre, Lucknow
140. ‘Prenatal Diagnosis: What is New’ in ISOPARB, Lucknow [The Lucknow Chapter of Indian Society of Perinatology & Reproductive Biology] Prenatal screening & diagnosis of Genetic disorders on 25th April 2018
141. ‘Rare Disease Scenario in Uttar Pradesh’ in Rare Disease Update 2018-UP, organized by Indian Society of Inborn Errors’ of Metabolism, held on 28 July 2018, at Hotel Fairfield by Marriott, Lucknow
142. ‘Genetics for Clinicians’ in Rare Disease Update 2018-UP, organized by Indian Society of Inborn Errors of Metabolism, held on 28 July 2018, at Hotel Fairfield by Marriott, Lucknow

143. 'Genetics for Obstetricians' Key Note address in GYNECON 2018 at Command Hospital, Lucknow on 20th September 2018

144. 'Disorders of Genomic Architecture –Microdeletion & Microduplication Syndromes' in 'Current Trends in Genomic and Molecular Medicine' The first Indo- UK Training Workshop, in KGMU, 19th to 21st Nov 2018.

145. 'Genetics of facial clefts' in APSI Accredation Course on Cleft Lip Palate & Cranio Facial Anomalies – 2018 , 19th Nov 201

146. "Future of Genetics in Neuro-disability" in Child Neurodisability Conference- 2018 on 3rd Dec 2018

147. 'Roadmap for Management of Rare Disorders in India', 13th April 2018, 7th International conference on Rare and Undiagnosed diseases. Addressing patient needs for Rare Disorders in India, 13-15th April 2019, New Delhi, India, Hotel Leela Ambience

148. 'Current Consensus And Practical Tips To Apply Newborn Screening In Clinical Practice' in Annual conference of ISIEM, at Pune 19th Jan 2019

149. 'Panels, panels everywhere; which test to choose - Genetic testing' in Genetics Workshop, PEDICON 2019, at KEM Hospital, Mumbai on 6th Feb 2019

150. 'Charting and interpreting pedigrees' in Genetics Workshop, PEDICON 2019, at KEM Hospital, Mumbai on 6th Feb 2019

151. 'Genetic diagnosis: Selecting the right tests ' in PEDICON 2019, at Mumbai, 9th March 2019

152. 'Genetics in Medicine' in "Latest Trends in Biotechnology for the benefit of the People of Uttar Pradesh" on 12th March, 2019.

153. 'Recurrent Congenital Anomalies: How to Approach?' in AMMC-CON [AN UPDATE ON MEDICAL DISORDERS IN OBSTETRICS] organized in association with Kanpur Obstetrics & Gynaecology Society and Kanpur Physicians Association at Kanpur on 17th March 2019

154. 'Genetics: You Need to Know for PCPNDT Implementation: Our Mission' on 23rd March 2019 for medical officers at CMO office Lucknow

155. 'Genetics in Autism' on World Autism Day – Keynote address o 2nd April 2019 at program organized by Association of Child Brain Research in M B Club, LucknowLucknow

156. ' Genetics - the Present and The Future of Medicine" on 7th April 2019 , at Genetics for Pediatricians in Nanavati Hospital, Mumbai

157. 'Practical demonstration of fetal growth charts and their application in day to day practice' in GESTOSIS 2019 organized at Scientific Convention Center, Lucknow on 23rd March 2019 under the aegis of India Gestosis Chapter, hosted by Lucknow Obstetric & Gynaecological Society (LOGS)

158. "Treatable Lysosomal Storage Disorders-Diagnosis and Management " on 27th July 2019, on line CME telecasted to pediatricians in India

159. "Prenatal Diagnosis: Current Scenario" in ISAR-ISPAT Conclave in Varanasi from 21st to 22nd September 2019

160. PR Dange Oration –'Utility of Genomic Tests in Clinical Practice' in 15th Annual Conference of Academy of Pediatrics **(NAPCON 2019**) & National Mid-Term CME of Infectious Diseases Chapter of IAP held on 29th September 2019 at Hotel Centre Point, Nagpur.
161. 'Diagnosis of genetic neuromuscular disorders - the Indian scenario' in Indo-US Symposium on Genetic Neuromuscular Disorders & Sixth Annual Conference of the Society for Indian Academy of Medical Genetics (SIAMGCON 2019) on 22nd Nov 2019
162. 'Autosomal recessive inheritance' in "Decoding Genetic Investigations: Interpretation of NGS and other Genetic Tests in the Clinical Setting" on 24th Nov 2019 in Centre for DNA Diagnostics and Fingerprinting, Hyderabad
163. 'Cytgenetic microarray' on 24th Nov 2019 in Centre for DNA Diagnostics and Fingerprinting, Hyderabad
164. 'Genetic Diagnosis in India: *Are we on right track?' Keynote address in* **MPAICON 2020 (8th Annual Conference of Molecular Pathology Association of India)** Held on 10th to 12th Jan 2020, a at Sri Ram Cancer Center, Mahatma Gandhi Medical College & Hospital, Sitapura, Jaipur.
