

## CURRICULUM VITAE

1. Name : Dr. Shubha Rajendra Phadke  
Formerly Professor and Head  
Department of Medical Genetics,  
Sanjay Gandhi Postgraduate Institute of Medical  
Sciences, Lucknow

2. Date of birth : 9.12.1960

3. Sex : Female

5. Address : G 163, Kalpataru Jade Residences, Pancard club road,  
Baner, Pune, Maharashtra, India, 411045. INDIA

6. Phone: +91 9839220199 (M)  
Email: shubharaophadke@gmail.com

7. Nationality : Indian

8. Qualification : M.B.B.S. (1982)  
M.D. (Pediatrics) 1986  
DM (Medical Genetics) 1992

9. Maharashtra Medical Council Registration No. 76682  
UP Medical Council Registration No. 39855

10. Qualifications: Examinations:

<b>Examination &amp; Year Of passing</b>	<b>Institute</b>	<b>Percentage of marks (Aggregate)</b>	<b>No. of attempts</b>
H.S.C.E. (1977)	Nagpur Board	Grade I Distinction	One
M.B.B.S. I M.B.B.S. II M.B.B.S. III M.B.B.S	Govt.MedicalCollegeNagpur Final M.B.B.S (Nov'81)	60.6% 58.0% 62.25%	One
MD Pediatrics (April 1986)	Govt.MedicalCollege, Nagpur	Pass	One
DM Medical Genetics	Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow	Pass	One

## 11. 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'. Published by Prism books.**
- **Editor of GENETICS CLINICS since 2008, a three monthly publication of SIAMG<[http://iamg.in/genetic\\_clinics/index.php](http://iamg.in/genetic_clinics/index.php)>**
- **On the editorial board of American Journal of Medical Genetics**
- **Section editor for European Jornal of Human Genetics**
- **On the editorial board of Indian Pediatrics**
- **Founder member of Indian Society of Inborn Errors of Metabolism**
- **First DM in Medical Genetics from India**
- **Incharge of the first DM [medical genetics] program since 1996**
- **Trained 46 DM students who are heading various centres / departments of medical genetics in India**
- **Run yearly two week long training course in medical genetics since 2000. Twentyfirst ICMR Course in Medical Genetics & Genetic Counseling was held in July-August 2024. Trained more than 600 clinicians at various stages of careers**
- **More than 300 publications in National and International journals**
- **Reported 10 new malformation syndromes**
- **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 7 monogenic disorders**  
**OMIM 607539 (Complex campto-syn-polydactyly caused by BHLHA9],**  
**OMIM 617612 (Joubert syndrome 30 caused by ARMC9 gene),**  
**OMIM 610188 (Joubert syndrome 9 caused by CEP290)**  
**OMIM 615866 (Coffin Siris syndrome 9 caused by SOX 11),**  
**OMIM 271510 (Sponastrime type of spondyloepimetaphyseal dysplasia (SEMDSP) caused by TONSL gene),**  
**OMIM 617927 (Orofaciodigital syndrome 18 caused by IFT57 gene),**  
**OMIM 617983 (AR Primary microcephaly 21 caused by NCAPD2 gene)**
- **Member of Rare Disease Committee of Indian Council of Medical Research, New Delhi, since 2019**
- **Nodal officer for Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow for National Policy for Rare Diseases 2021 since June 2021 till Feb 2025 & Chair person, Rare Disease Committee for SGPGIMS, Lucknow**
- **Appointed as a Member [expert] of the National Assisted Reproductive Technology and Surrogacy Board for the purposes of the Assisted Reproductive Technology, (Regulation) Act, 2021 and the Surrogacy (Regulation) Act, 2021; since 4 Aug 2022 till date**
- **A member of ethics committee of Centre for Bio Medical Research, Lucknow since Sept 2021 to August 2023**
- **A member of Institutional Ethics Committee, King George Medical University, UP, Lucknow**
- **Delivered Tedx talk in Nagpur on 30<sup>th</sup> April 2023**  
<https://www.youtube.com/watch?v=MMEgBZcibUM>
- **Felicitation for establishing services for patients with thalassemia by Thalassemics India [patient organization] on the occasion of completion of 35 years of their journey on 25<sup>th</sup> March 2023.**

**tonsl**

- **'Dr IC Verma oration' in 3rd National Symposium on Genetic Diseases "RDIFCON2024" on 13th July 2024 at Ayurvigyan Auditorium, Army Hospital Research and Referral, New Delhi**
- **Honored as a legend, 'Fetal Medicine: Beyond USG' in the XVIII Clinical Ultrasonography in Practice (CUSP) conference, on 28<sup>th</sup> September 2024, at Chennai Trade Centre, Chennai.**

- Served as Chairman, Internal Complaint Committee of SGPGIMS from Jan 2023 to 31<sup>st</sup> March 2025
- **Chairman of the editorial committee for the Coffetable book of SGPGIMS, 2020-2024: IN PURSUIT OF EXCELLENCE**

## 12. EXPERIANCE

Institution	Position	Period	Duration
Department of Medical Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow	Professor & Head of the department	Jan 2006	To 31 <sup>st</sup> March 2025
Department of Medical Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow	Additional Professor	July 2004	Jan 2006
Department of Medical Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow	Associate Professor	July 2000 to June 2004	4 years
Department of Medical Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow	Assistant Professor	April 1996 to July 2000	4 years
Department of Pathology, University of Washington, Seattle, USA	Fellowship	April 1994 to July 1994	3 months
Department of Medical Genetics, SGPGIMS, Lucknow	Research associate	1 <sup>st</sup> Feb 1993 to 31 <sup>st</sup> Jan 1994	1 year
Department of Medical Genetics, SGPGIMS, Lucknow	Senior Resident (Medical Genetics)	Jan 1990 to Dec 1993	3 years
Department of Medical Genetics, SGPGIMS, Lucknow	Research officer (Medical Genetics)	Feb 1989 to Dec 1989	11 months

## 13. RESEARCH PROJECTS

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 trial 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 Investigator-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 Investigator. 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 4 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 Investigator], 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 Investigator](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 Investigator]
15. Centre for Molecular Medicine: Indian Council of Medical Research. Rs 499 lac, for 5 years. Sanctioned on 21-3-12. (Completed) [Principle Investigator]
16. Evaluation of utility of cytogenetic microarray in detection of etiology of Prenatally detected malformation Rs 3 lac. Intramural. 1<sup>st</sup> Nov 2012 to 31<sup>st</sup> oct 2014(Completed) [Principle Investigator]
17. Multicentric Collaborative Study of the Clinical, Biochemical and Molecular Characterization of Lysosomal Storage Disorders in India. Principle Investigator – Shubha Phadke. Indian Council of Medical Research (Rs 31.3 lac for 3 years- Sanctioned on 10 th Dec 2014 - Completed)
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 Investigator. (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 4 years in Oct 2018 – 20-9-2022 (Principle Investigator – 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 9<sup>th</sup> May 2019 to 8 May 2024 (Rs 1.23.64.000.00) [Principle Investigator]
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 Investigator - Completed]- 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 27<sup>th</sup> August 2019 - , Rs 21,57,100 for 3 and half years [Completed\[Principle Investigator]
23. National registry for rare and other inherited disorders – sanctioned by Indian Council of Medical Research in Sept, 2019 till 1<sup>st</sup> Feb 2025 - for Rs 39.6 lac [Principle Investigator]
24. A Post Marketing Surveillance (PMS) Study for VPRI (Velaglucerase alfa) in India. -Sponsorer: Takeda Biopharmaceuticals India Pvt. Ltd. Date of start: 08-Dec-2021 - Date of Completion: 21-06-2023. Rs 2.20,000-00

#### **14. 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 25<sup>th</sup> June to 28<sup>th</sup> 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 15<sup>th</sup> to Jan.22<sup>nd</sup>, 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, 8<sup>th</sup> to 11<sup>th</sup> Feb. 2001 at Patna.
6. International congress of Human Genetics, May 15<sup>th</sup>-19, 2001 at Vienna, Austria.
7. 5<sup>th</sup> National Conference of Indian Society of Prenatal Diagnosis and Therapy. 18<sup>th</sup>-20<sup>th</sup>, Jan. 2002 at PUNE.
8. 43<sup>rd</sup> Short Courses in Medical and Experimental Mammalian Genetics at Bar Harbor USA 15<sup>th</sup> to 26<sup>th</sup> July 2002.
9. WHO Intercountry consultation on identifying regional priorities in the Area of Human Genetics in SEAR at Bangkok, Thailand 23-25<sup>th</sup> September 2003 as an invited expert.

10. VIth National Conference of Indian Society of Prenatal Diagnosis andtherapy, 23<sup>rd</sup> Jan to 5<sup>TH</sup> Jan 2002 at Pune.
11. VIIth National Conference of Indian Society of Prenatal Diagnosis AND therapy, 23<sup>rd</sup> Jan to 5<sup>th</sup> Jan 2004 at Ahmadabad.
12. 41<sup>st</sup> annual National conference of Indian Academy of Pediatrics, 8<sup>th</sup> Jan to 11<sup>th</sup> Jan 2004 at Chennai.
13. National conference of Indian Academy of Pediatrics; 6<sup>th</sup> Jan TO 9<sup>th</sup> Jan 2005 at Kolkota.
14. International conference of Inborn Errors of Metabolism, 28<sup>th</sup> Sept to 2<sup>nd</sup> Oct 2005, at New Delhi. Multicentric Task Force Meeting on Inborn Metabolic Diseases , 5<sup>th</sup> May 2004 at Bangalore.
15. Clinical and Laboratory Approach to Inherited Metabolic Disorders, 21<sup>st</sup> to 23<sup>rd</sup> 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, 17<sup>th</sup> TO 20<sup>th</sup>Jan 2008
20. ISPAT 2007 Chennai , 23<sup>rd</sup> to 25<sup>th</sup> Nov 2007.
21. HUGO – 13<sup>TH</sup> Human Genome meeting at Hyderabad from 27<sup>th</sup> September to 30<sup>th</sup> September, 2008.
22. North India Conference on Fetal Medicine and Prenatal Diagnosis, at AIIMS, New Delhi, 16<sup>th</sup>nov 2008.
23. Spring Conference of Clinical Genetics Society, London, UK, 12<sup>th</sup> March 2009.
24. Fourth International Conference on Birth Defects and Disabilities in Developing World, New Delhi, 4 – 7<sup>th</sup> October 2009.
25. 12 th Annual Asia Symposium onLysosomal Storage Disorders, Taipei, 9<sup>th</sup> to 11<sup>th</sup> October 2009.
26. Annual conference of American College of Medical Genetics, at Vancouver, Canada from 15<sup>th</sup> March 2011 to 20<sup>th</sup> March 2011.
27. Twelfth ICHG and annual conference of American Society of Human Genetics at Montreal, Canada, 11<sup>th</sup> to 15<sup>th</sup> Oct 2011
28. Expert Group Meeting on Birth Defects in WHO SEAR ' from 13<sup>th</sup> to 15<sup>th</sup> Dec 2011 at AIIMS, New Delhi.
29. GENECON 2012 at Raipur, on 1<sup>st</sup> and 2<sup>nd</sup> Dec 12
30. Indo US Symposium on Disorders of Developing Brain' at Manipal, 27<sup>th</sup> -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" 31<sup>st</sup> 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 & 2<sup>nd</sup> national conference of ISIEN' at New Delhi on 5<sup>th</sup> -7<sup>th</sup> April 2013

36. 'Fabry disease: Diagnosis & ERT' in
37. 'Genetics of Epilepsy' in International conference on Cerebral Palsy and Developmental Medicine, 6<sup>th</sup> to 10<sup>th</sup> 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 6<sup>th</sup> Feb 13
39. 2<sup>nd</sup> International Congress of Society of Fetal Medicine, on 31<sup>st</sup> 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 – 29<sup>th</sup> 2013
41. MAHAPEDICON 2013, annual conference of Maharashtra API, on 19<sup>th</sup> Oct 13 at Nagpur
42. A workshop on "Maternal Fetal Medicine" from 25<sup>th</sup> – 27<sup>th</sup> November 2013, In the department of Obstetrics and Gynecology in King George's Medical College, Lucknow
43. 8<sup>TH</sup> Asia Pacific Regional Meeting of International Society of Newborn Screening, 27<sup>th</sup>, 28<sup>th</sup>, 29<sup>th</sup> Sept13, in New Delhi
44. MEDGENCON 2014, 1<sup>st</sup> March 2014 at Bangalore
45. One-Day state level symposium: "Exploring Statistics Applications in Diverse Fields" at Nagpur, 7<sup>th</sup> 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 5<sup>th</sup> Dec 2014
49. Annual Conference of Indian Society at Mumbai on 28- 29<sup>th</sup> Jan 2015
50. 11<sup>th</sup> ISUOG International Symposium, New Delhi, 1<sup>st</sup> to 3<sup>rd</sup> May 2015
51. Indo US Conference : Realizing the Potentials of Rare Disorders in India', at United Services Institutions, New Delhi on 7<sup>th</sup>& 8<sup>th</sup> Sept 2015
52. The UW Mendelian Data Analysis Workshop from 10<sup>th</sup> August 15 to 15<sup>th</sup> 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 27<sup>th</sup> Nov to 29<sup>th</sup> 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, 55<sup>th</sup> Annual Conference of Indian Academy of Pediatrics, from 4<sup>th</sup> to 7<sup>th</sup> January 2018, held at Nagpur
57. 2<sup>nd</sup> conference in the series Recent Advances in Rare Disease – RARD2018 (Frequently misdiagnosed hereditary disorders (FREMIDIS) – multidisciplinary

translational research affects global  
clinical impact ) May 3<sup>rd</sup>-5<sup>th</sup>, 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 16<sup>th</sup> Oct 2018 to 20<sup>th</sup> Oct 2018

60. ISPD 23rd International Conference athenon Prenatal Diagnosis Saturday, 7 September 2019 – Wednesday, 11 September 2019 MAX Atria At Singapore EXPO, Singapore

61. The 14<sup>th</sup> International Congress of Human Genetics, held from 22<sup>nd</sup> Feb to 26<sup>th</sup> Feb 2023, at Cape Town South Africa. presented my work as a poster titled “Exome Sequencing in Understanding the Etiologies of Ataxia in Children”

62. 19th Manchester Dysmorphology Conference 2023, held from 16<sup>th</sup> to 18<sup>th</sup> Oct 2023, at Hilton Hotel, Manchester, UK. Poster presentation poster titled “Polydactyly – A 20 Year Journey from Clinical to Molecular Diagnosis”

63. The International Genomics Education and Training Summit, held at Conference Centre on the Wellcome Genome Campus, Hinxton, Cambridge on 27<sup>th</sup> & 28<sup>th</sup> November, 2023 [Participation by invitation and funding by the organizers] Poster presentation on “Genomics Education for Clinicians at Any Stage of Career Using Diverse Approaches: *Our Experience of 3 Decades*”

64. **Felicitation for establishing services for patients with thalassemia by Thalassemics India** [patient organization] on the occasion of completion of 35 years of their journey on 25<sup>th</sup> March 2023.‘

65. Annual conference of Society of Indian Academy of Medical Genetics; SIAMGCON 2023 held from 30<sup>th</sup> nov 23 to 3<sup>rd</sup> dec 23 at New Delhi [Gurgaon].

66. **‘National Symposium on Rare Diseases: Challenges, Opportunities and Way Forward’ ORGANIZED BY** the Ministry of Health & Family Welfare (MoHFW), Government of India in collaboration with World Health Organization (WHO) **India on 29th February-1st March 2024 at Radisson Blu Palace, Udaipur, Presented report of Centre of Excellence for Rare Diseases at SGPGIMS**

67. National Conference on Rare Diseases (RarDisESICON) ORGANIZED ON 2<sup>ND</sup> March 2024 at ESIC Medical College, Faridabad

68. Annual conference of Society of Indian Academy of Medical Genetics; SIAMGCON 2024 held from at 5<sup>th</sup> Dec 2024 to 8<sup>th</sup> Dec 2024 at The Belvedere Golf and Country Club, **Ahmedabad**

69. “The Second Genomics Education and Training Summit” organized by held on 20<sup>th</sup> and 21<sup>st</sup> Feb 2025 at Divani Palace, Athens, Greece

70. The National Genetic Counseling Symposium, 2025; held on 7th–8th March, 2025, at the Centre for DNA Fingerprinting and Diagnostics (CDFD), Hyderabad.

## 15. Hospital visited, visiting scientist-fellowship:

- Service de Medicine et de Biologie foetales (Department of Fetal Medicine). Institute de Peuriculture de Paris. PARIS from 20<sup>th</sup>, April to 5<sup>th</sup> May, 1991.
- Visited University of Washington, Seattle USA from 12<sup>th</sup> April to 9<sup>th</sup> 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 2009
- Human Genetics, University of Michigan, Ann Arbor, MI, USA, 31<sup>ST</sup> March14 to 12<sup>th</sup> April 14

## 16. INVITED LECTURES INTERNATIONAL CONFERENCES

1. "Malformation Syndromes: Our experience" Spring Conference of Clinical Genetics Society, London, UK, 12<sup>th</sup> March 2009.
2. "Genetics of Short Stature" in Fourth International Conference on Birth Defects and Disabilities in Developing World, New Delhi, 4 – 7<sup>th</sup> October 2009.
3. "Genetic Screening" in Fourth International Conference on Birth Defects and Disabilities in Developing World, New Delhi, 4 – 7<sup>th</sup> October 2009.
4. 'Clinical Genetics: Indian Scenario' at Twelfth ICHG and annual conference of American Society of Human Genetics at Montreal, Canada, 11<sup>th</sup> to 15<sup>th</sup> Oct 2011
5. 'Genomic Techniques and Consanguinity in Rare Disorders' in MEDLAB [Arab Health Conference] on 26<sup>th</sup> Jan2016, at Convention centre, Dubai, UAE
6. Newborn screening in India: Experience from Pilot Initiatives (UP Experience-DBT project). in 8<sup>TH</sup> Asia Pacific Regional Meeting of International Society of Newborn Screening, 27<sup>th</sup>, 28<sup>th</sup>, 29<sup>th</sup> Sept13, in New Delhi

## 17. PUBLICATIONS:

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

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

@ - References cited in OMIM – 22

\$ - Papers related to prenatal diagnosis - 27

- 1) R V, **Phadke S R** (1990-1991) Spinal dysraphism in achondroplasia. Pediatr Neurosurg 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 ObstetGynaecol* 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\*#.

33) **Phadke S R**, Pandey A (1999) Genetic counseling in pediatric practice. Indian Pediatr 36: 789-797.

34) **Phadke S R** (1999) Iniencephaly in a live born and not Klippel Feil syndrome. Indian Pediatr 36: 1279.

35) Saxena A, **Phadke S R**, Agarwal S S (2000) Linear catch-up growth. Indian J Pediatr 67(3): 225-230.

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 supplementation 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

43) Chaturvedi L S, Shrivastav S, Mukherji M, Mittal R D, **Phadke S R**, **Pradhan S**, Mittal B (2001) Carrier detection of nondeletional Duchenne muscular dystrophy / Becker muscular dystrophy families using polymorphic dinucleotide repeat (CA) repeat loci of dystrophin gene. IJMR14(1):47-49 \$

44) Puri R D, **Phadke S R** (2002) Further delineation of mandibulofacial dysostosis: Toriello type. Clinical Dysmorphology 11(2)91-93\*@.

45) Chaddha V, **Phadke S R** (2002) Aarskog syndrome . Indian Pediatr 39:400

46) **Phadke S R** (2002) Down syndrome: A common genetic problem and the challenges ahead. Asian J of Pediatric Practice 5(4):10-14

47) Agarwal SS, **Phadke S R**, Agrawal S ( 2002) Primary prevention of Thalassemia major. Asian J of Pediatric Practice 5(4):23-29

48) Panigrahi I, **Phadke S R**, Agrawal A, Gambhir S, Agarwal S S (2002) Clinical profile of hereditary spherocytosis in North India .JAPI 50:1360-1367

49) Panigrahi I, Kesari A, **Phadke S R**, Mittal B (2002) Clinical and molecular diagnosis of spinal muscular atrophy. *Neurology India* 50:117-122

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\*#

51) Saxena A, **Phadke S R** (2002) Thalassemia control by carrier screening: The Indian scenario. *Current Science* 83(3):291-295

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.

53) Pandey G S, **Phadke S R**, Mittal B (2002) Carrier analysis and prenatal diagnosis of hemophilia in North India. *International J of Molecular Medicine* 10: \$

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.

57) **Phadke S R**, Agarwal S (2002) Prenatal screening for Down syndrome. *Perinatology* 4(4):198-206\$

58) **Phadke S R** (2003) Prevention of genetic disorders. *World Health Review*. April : 18-23.

59) **Phadke S R**, Agarwal S (2003) The phenotype score to grade the severity of Thalassemia Intermedia. *Indian J Pediatr* 70 (6): 477-481.

60) Gupta A, **Phadke S R**, Thakur S. (2003) Diagnosing Acrocallosal syndrome. *Indian J Pediatr* 70(2): 177-179.

61) **Phadke S R**, Puri R D, Agarwal S, Thakur S (2003) Counseling for prenatally detected malformation of uncertain prognosis. *Perinatology* 5(3): 132-137\$.

62) Agarwal S, Thakur S, Agarwal S, Khan F, **Phadke S R**, Pradhan M, Tripathi M (2003) Factor V Leiden mutation (C169S1A) in women with recurrent spontaneous abortion from North India. *Obs&Gynec VIII (II)*: 603-607.

63) Chaddha V, Agarwal S, **Phadke S R**, Mittal B (2003) Low level mosaicism in atypical Prader Willi syndrome: Detection using fluorescent in situ hybridization. *Indian Pediatr* 40:166-168.

64) **Phadke SR**, Agrawal S, Puri Dua R (2003) Recurrence of complex camptopolydactyly in a sibling suggestive of autosomal recessive inheritance. *Am J Med Genet* 116A:94-96\*@\$

65) Singh H, Pradhan M, Singh R L, **Phadke S**, Naik S R, Aggarwal R, Naik S (2003) High frequency of hepatitis B virus infection in patients with beta thalassemia receiving multiple transfusions. *Vox Sanguinis* 84: 292-299.

66) Pandey G S, Panigrahi I, **Phadke S R**, Mittal B (2003) Knowledge and attitude towards hemophilia: The family side and role of hemophilia societies. *Community Genetics* 6(2):120-122

67) Mukherjee M, **Phadke S R**, Mittal B (2003) Connexin 26 and autosomal recessive nonsyndromic hearing loss. *Indian J Human Genet* 9(2): 40-50.

68) Puri R D, **Phadke S R** (2003) Catel-Manzke syndrome without cleft palate – a case report. *Clinical Dysmorphology*. 12(4): 279-281@.\*

69) **Phadke S R** (2004) Genetic counseling. *Indian J Pediatr* 71:151-158.

70) Pandey U B, **Phadke S R**, Mittal B (2004) Molecular diagnosis and genetic counseling for fragile X mental retardation. *Neurology India* 52: 36-42.

71) Thakur S, Paul L, **Phadke S R** (2004) Lethal arthrogryposis with ichthiosis : overlap with Neu Laxova syndrome, restrictive dermopathy and Harlequin fetus. *Clinical Dysmorphology* 13 (2): 117-119\*\$.

72) Pandey A, **Phadke S R**, Gupta N, Phadke R V (2004) Neuroimaging in mental retardation. Indian J Pediatr 71: 203-209.

73) Gupta N, Girisha K M, **Phadke S R** (2004) Prenatally diagnosed omphalocele – what more to look for. Perinatology 6(2):87-95\$.

74) Thakur S, Singh R, Pradhan M, **Phadke SR** (2004) Spectrum of holoprosencephaly. Indian J Pediatr 71:593- 597\$

75) **Phadke S R**, Patil S J (2004) Partial trisomy 13 with features similar to ‘C’ syndrome. Indian Pediatr 41:614-617

76) **Phadke S R**, Pandey A, DuaPuri R, Patil S J (2004) Genetic counseling: The impact in Indian milie. Indian J of Pediatrics 71:1079-1082 \$

77) Shreeram K, Kumar S, Baijal SS, Handique A, Agarwal SK, **Phadke S R** (2004) Endovascular management of a posterior tibial artery aneurism in Ehlers Danlos syndrome type VI. EJVES Extra (e- pub) 7(6): 74-75

78) Khan F, **Phadke S**, Nityanand S, Talwar S, Agrawal S (2004) Use of ApoB3' hypervariable region in studying mixed chimerism and maternal contamination in North Indian populations. J of Clinical Forensic Medicine 11:183-188

79) **Phadke SR** (2004) Perinatal pathology: A prerequisite for genetic counseling. Asian J of Pediatric Practice 8(1):33-38

80) Mukherjee M, Pandey G S, Kesari A, **Phadke S R**, Mittal B (2004) Prenatal diagnosis of genetic disorders by molecular methods. Perinatology 6(5): 221- 230 \$

81) Singh R, **Phadke SR**, Pradhan M. Dalal A (2004) Unexpected chromosomal abnormalities at prenatal diagnosis: A counseling dilemma. Perinatology 6(6): 305-309 \$

82) Gupta N, Pradhan M, Singh R, **Phadke S R** (2004) Prenatally diagnosed trisomy 6 mosaicism. Prenatal Diagnosis 24:841-844 \$

83) **Phadke S R** (Feb2005) Fragile X syndrome. Orphanet Encyclopedia. <http://www.orpha.net/data/patho/GB/uk-Fragile-X.pdf>

84) Kesari A, Misra U K, Kalita J, Mishra V N, Pradhan S, Patil S J, **Phadke S R**, Mittal B (2005) Study of survival motor neuron(SMN) and neuronal apoptosis inhibitory protein (NAIP) gene deletions in SMA patients. J of Neurology

85) Girisha K M, **Phadke S R** (2005) Anotia and facial palsy: Unusual features of cardiofacial syndrome. Indian J of Pediatrics 72:525 -526 \*

86) Dalal A, **Phadke S R** (2005) Hemihyperplasia with Ehlers Danlos syndrome like skin changes. Clinical Dysmorphology

87) Kesari A, Rennert H, Leonard D G B, **Phadke S R**, Mittal B (2005) Prenatal diagnosed of spinal muscular atrophy: Indian experience. Prenatal Diagnosis 25:641-644 \$

88) **Phadke S R**, Thakur S, Sankar VH (2005) Utility of information brochure as a part of pretest counseling for Down syndrome screening: Indian scenario. Perinatology 7(4):178- 182 \$

89) Thakur S, **Phadke S R** (2005) familial breast cancer: Genetics and counseling. Indian J of Surgery 67(6):297-301

90) Patil SJ, **Phadke S R** (2006) Delleman Syndrome. Indian Pediatrics 43; 173-174

91) Dalal A, **Phadke S R** (2006) MTHFR polymorphism in neural tube defects. J OF Metabolism and Genetics. 1(2): 109-114

92) Sankar V H, **Phadke S R** (2006) Ring chromosome 13 in an infant with ambiguous genitalia. Indian Pediatrics 43:258-260

93) **Phadke SR**, Manisha (2006) Further delineation of acro-renal-mandibular syndrome. Clin Dysmorphology 15:119-120 \*

94) **Phadke SR**, Girisha KM, Dalal A (2006) Handless footless fetus. Clin Dysmorphology15:233-234 \*

95) Sankar V H, **Phadke S R** (2006) Clinical utility of autopsy and comparison with prenatal ultrasound findings. *J of Perinatology* 26:224-229 \$

96) Gupta N, **Phadke S R** (2006) Cutis Laxa Type II and Wrinkly Skin Syndrome: Distinct phenotypes. *Pediatr Dermatol*;23(3):225-30.

97) Julka S, Bhatia V, Singh U, Northam E, Dabadghao P, **Phadke S**, Wakhlu A, Warne GL (2006) Quality of life and gender role behavior in disorders of sexual differentiation in India. *J of Pediatric Endocrinology & Metabolism* 19:879-888

98) **Phadke SR**, Girisha KM (2006) Basal ganglia changes: A diagnostic clue to Sandhoff disease. *Indian Pediatrics* 43:919-910

99) Gupta N, Goel H, **Phadke SR**. Unbalanced X; autosome translocation. *Indian J Pediatr.* 2006 Sep;73(9):840-2.

100) Patil S J, **Phadke S R** (2006) Urorectal septum malformation sequence: Ultrasound correlation with fetal examination. *Indian J Pediatrics* 73(4):287 -293 \$

101) Girisha K M, **Phadke S R**, Khan F, Agrawal S (2006) S252W mutation in Indian patients with Apert syndrome. *Indian pediatrics* 43(80):733-735

102) Dalal AB, **Phadke SR**, Pradhan M, Sharda S. (2006) Hemihyperplasia syndromes. *Indian J Pediatrics* 73(3):609-615

103) Dalal A, **Phadke S R** (2006) Twin pregnancy with Roberts syndrome in one fetus and trisomy 18 in the other. *J Clin Ultrasound.* 2006 Mar-Apr;34(3):146-9.

104) **Phadke SR**, Patil SJ (2006) Pericentric inversion giving rise to duplication and deletion of chromosome 13q22→qter in the offspring. *Am J Med Genet* 143(8):899-902 \$

105) Dalal A B, **Phadke SR**.(2007) Morphometric analysis of face in dysmorphology. *Comput Methods Programs Biomed.* Feb;85(2):165-72

106) **Phadke S R**, Dua Puri Ratna, Phadke R V (2007) Severe Form of Congenital Cerebral and Cerebellar Atrophy : A Neurodegenerative Disorder of Fetal Onset. *J of Clinical Ultrasound Jul-Aug;35(6):347-50.* \$

107) **Shubha R Phadke**, Girisha KM, Rajendra V Phadke (2007) A New autosomal recessive disorder of bilateral frontotemporal pachygyria without microcephaly: Report of a case and review of literature. *Neurology India* 55(1):57-60

108) **PhadkeS R**, PatilSJ, Niraj Kumari, KrishnaniN. (2007) Spondylothoracic dysplasia: prenatal diagnosis and the problems of nosologic overlap. *Am J Med GenetA.* Apr 15;143(8):899-902. \$

109) **Phadke S R**, Girisha K M (2007) Issues in counseling for Down syndrome: Indian scenario. *Indian Pediatrics* 44:131-133.

110) **Phadke S R**, Dalal A (2007) Short stature, ulnar deviation of hands with absent carpal and joint contractures: a new syndrome. *Clin Dysmorphol* 16(1):55-7.

111) Girisha K M, Mittal B, **Phadke S R** (2007) Prevention of common genetic neuromuscular disorders: Duchenne muscular dystrophy and spinal muscular atrophy. *Pediatric Clinics of India.* jan;22-38

112) Brancati F, Barrano G, Silhavy JL, Marsh SE, Travaglini L, Bielas SL, Amorini M, Zablocka D, Kayserili H, Al-Gazali L, Bertini E, Boltshauser E, D'Hooghe M, Fazzi E, Fenerci EY, Hennekam RC, Kiss A, Lees MM, Marco E, **Phadke SR**, Rigoli L, Romano S, Salpietro CD, Sherr EH, Signorini S, Stromme P, Stuart B, Sztriha L, Viskochil DH, Yuksel A, Dallapiccola B; The International JSRD Study Group, Valente EM, Gleeson JG. CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndrome-Related Disorders. *Am J Hum Genet.* 2007 Jul;81(1):104-13. @

113) Sharada S , **Phadke S R** (2007) Uptake of invasive prenatal diagnostic tests in women after detection of soft markers for chromosomal abnormality on ultrasonographic evaluation. **J Perinatol.** 2007 Jul 5; \$

114) **Phadke SR**, Ramirez M, Difeo A, Martignetti JA, Girisha KM.(2007) Torg-Winchester syndrome: lack of efficacy of pamidronate therapy. **Clin Dysmorphol.** 16(2):95-100.

115) Malik S, Girisha KM, Wajid M, Roy AK, **Phadke SR**, Lerche D, Richardt T, Haque S, Ahmad W, Koch MC, Grzeschik K. Identical HOXD 13 polyalanine expansion mutations in syndactyly type II families of Pakistani and Indian origin. **BMC Med Genet** 8(1):78.

116) Girisha KM, Cormier-Daire V, Heuertz S, Phadke RV, **Phadke SR**. (2008) Novel mutation and atlantoaxial dislocation in two siblings from India with Dyggve-Melchior-Claussen syndrome. **Eur J Med Genet** 51(3):251-6.

117) **Phadke S R** (2007) Hemihyperplasia, Isolated. In *Atlas of Genetics and Cytogenetics in Oncology and Haematology*. <<http://atlasgeneticsoncology.org/Kprones/HemihyperplasiaID10046.html>>

118) **Phadke SR**. (2008) Challenges in identifying genetic risk factors for common multifactorial disorders. **Indian J Med Res.** 2008 Feb;127(2):106-9.

119) Mandal K, **Phadke SR**, Kalita J. Congenital swan neck deformity of fingers with syndactyly. **Clin Dysmorphol.** 2008;17(2):109-11.

120) Sahu R, Boddula R, Sharma P, Bhatia V, Greaves R, Rao S, Desai M, Wakhlu A, **Phadke S**, Shukla M, Dabadghao P (2008) Genetic analysis of SRD5A2 gene in Indian patients with 5 alpha reductase deficiency. **Journal of Pediatric Endocrinology and Metabolism**

121) **Phadke S R**, Sharda S (2008) A report of a patient with interstitial deletion of 15q22: further delineation of a new micro deletion syndrome. **Am J Med Genet A.** 146A(15):1999-2000

122) Goel H, Girisha KM, **Phadke SR**. (2008) Long-term efficacy of oral deferiprone in management of iron overload in beta thalassemia major. **Hematology.** 13(2):77-82.

123) Kumari N, Pradhan M, Shankar VH, Krishnani N, **Phadke SR**. (2008) Post-mortem examination of prenatally diagnosed fetal renal malformation **J Perinatol.** 2008 Jul 3.

124) Gupta A, **Phadke S R** (2008) Chromosomal anomalies in couples with recurrent spontaneous abortions: Prevalence and counseling. **Perinatology** 10(3): 69-74

125) Ranganath P, Agrawal M, **Phadke SR** (2008) Second trimester screening for fetal aneuploidy through triple marker test: the two year experience of genetics unit at a referral institute. **Perinatology** 10(6): 149-154. \$

126) RPGRIP1L mutations are mainly associated with the cerebello-renal phenotype of Joubert syndrome-related disorders. Brancati F, Travaglini L, Zablocka D, Boltshauser E, Accorsi P, Montagna G, Silhavy JL, Barrano G, Bertini E, Emma F, Rigoli L, **Phadke S**; International JSRD Study Group, Dallapiccola B, Gleeson JG, Valente EM. **Clin Genet.** 2008 Aug;74(2):164-70.

127) Mandal K, Boggula V R, Borkar M, Agarwal S, **Phadke S R** (2009) Use of multiplex ligation probe amplification (MLPA) in screening of subtelomeric regions in children with idiopathic mental retardation. **Indian J Pediatrics** 76(10):1027-31. Epub 2009 Nov 12.

128) **Phadke S R**, Mandal K, Girisha K M (2009) Fabry disease: A treatable lysosomal storage disorder. **National Med J of India** 22(1): 20-22

129) Expanding CEP290 mutational spectrum in ciliopathies. Travaglini L, Brancati F, Attie-Bitach T, Audollent S, Bertini E, ...., **Phadke SR**, et al. Am J Med Genet A. 2009 Oct;149A(10):2173-80. @

130) Patil SJ, Banerjee M, **Phadke SR**, Mittal B (2009) Mutation analysis in Indian children with achondroplasia - utility of molecular diagnosis. Indian J Pediatr. 76(2):147-9. Epub 2009 Mar 28..

131) **Phadke S R**, Gupta N, Girish K M, Kabra M, MaedaM, Vidal E, MoserA, Steinberg S, Puri R D, Verma I C, Braverman. (2010) Rhizomelic Chondrodyplasia Punctata Type 1- Report of 3 cases from India. Journal of Applied Genetics ;51(1):107-110

132) Aggarwal S, Gupta G, **Phadke S R** (2009) Good outcome in two fetuses with echogenic lungs. Perinatology 11(2):82-85. \$

133) **Phadke S R** and Sankar V H (2009) Polydactyly and genes. Indian J of Pediatrics

134) **Phadke S R**, FischerB, GuptaN, Prajnya R, Kabra M, Uwe Kornak (2010) Mutation spectrum in Indian patients with autosomal recessive infantile malignant osteopetrosis: Report of novel mutations. Indian Journal of Medical Research 131:508-514

135) Tamhankar PM, **Phadke SR** (2010) Clinical profile and molecular diagnosis in patients of facioscapulohumeral dystrophy from Indian subcontinent. Neurol India. 2010 May-Jun;58(3):436-40

136) **Phadke SR**, FischerB, GuptaN, Prajnya R, Kabra M, Kornak U (2010) Mutation spectrum in Indian patients with autosomal recessive infantile malignant osteopetrosis: Report of novel mutations. Indian Journal of Medical Genetics 131:508-514

137) Girisha K M, Mandal M, **Phadke S R** (2009) milder form of pachydermoperiostosis: a report of four cases. Clinical Dysmorphology 18:85-89

138) Ranganath P, Laine CM, Gupta D, Mäkitie O, **Phadke SR**. COL1A1 Mutation in an Indian Child with Caffey Disease. Indian J Pediatr. 2011 Jan 20.

139) Ahmad Z, **Phadke S**, Arch E, Glass J, Agarwal A, Garg A. Homozygous null mutations in ZMPSTE24 in restrictive dermopathy: evidence of genetic heterogeneity. Clin Genet. 2010 Oct 23. doi: 10.1111/j.1399-0004.2010.01580.x.

140) Girisha KM, Lewis LE, **Phadke SR**, Kutsche K Costello syndrome with severe cutis laxa and mosaic HRAS G12S mutation. Am J Med Genet A. 2010 Nov;152A(11):2861-4.

141) Agarwal M, Parveen F, Faridi RM, **Phadke SR**, Das V, Agrawal S Recurrent pregnancy loss and apolipoprotein E gene polymorphisms: a case-control study from north India. Am J Reprod Immunol. 2010 Sep;64(3):172-8.

142) Run-on mutation in the PAX6 gene and chorioretinal degeneration in autosomal dominant aniridia. (2011) Shagun Aggarwal, Worapoj Jinda, Chanin Limwongse, LaongsriAtchaneeyasakul, **Shubha R. Phadke**. Molecular Vision. 2011; 17:1305-1309.

143) Prajnya R, Rehder C, Phadke SR, Bali D. Prenatal diagnosis of Pompe disease: enzyme assay or molecular testing? Indian Pediatr. 2011 Nov 11;48(11):901-2. doi: 10.1007/s13312-011-0130-x. PMID: 22711147. \$

144) Ranganath P, Agarwal M, **Phadke SR**. (2011) Angelman syndrome and prenatally diagnosed Prader-Willi syndrome in first cousins. American Journal of Medical Genetics. 155A(11):2788-90. \$

145) **Phadke SR**, Agarwal M, Aggarwal S. Late termination of pregnancy for fetal abnormalities: The perspective of Indian lay persons and medical practitioners. Prenat Diagn. 2011 Dec;31(13):1286-91. \$

146) Shagun Aggarwal, Farah Parveen, Rehan Mujeeb Faridi, **Shubha Phadke**, Minal Borkar and Suraksha Agrawal. Vascular Endothelial Growth Factor gene polymorphisms in Indian patients with recurrent miscarriages. ReprodBiomedOnline. 2011 Jan;22(1):59-64.

147) **Phadke S R**, Sharda S, Urquhart J, Jenkinson E, Chawala S, Trump D (2011) Report of two brothers with short stature, microcephaly, mental retardation, and retinoschisis-A new mental retardation syndrome? *Am J Med Genet A*. 155A(1):9-13.

148) **Phadke SR**, Aggarwal S, Kumari N. (2011) The expanding spectrum of Elejalde syndrome: overlap with other disorders of overgrowth. *Clin Dysmorphol*.20(2):98-101.

149) Goel H, **Phadke SR** (2011) Reciprocal balanced translocation: infertility and recurrent spontaneous abortions in a family. (2011) *Andrologia*. Feb;43(1):75-7

150) Verma PK, Dalal A, Mittal B, **Phadke SR**. (2012) Utility of MLPA in mutation analysis and carrier detection for Duchenne muscular dystrophy. *Indian J Hum Genet*.18(1):91-4.

151) **Phadke SR**, Ranganath P, Boggula VR, Gupta D, Phadke RV, Sloman M, Turnpenny PD. Brothers with hypospadias, vertebral segmentation defects, and intellectual disability: new syndrome? *Am J Med Genet A*. 2012 Dec;158A(12):3065-70.

152) Singh K, Kumar R, Shukla A, **Phadke SR**, Agarwal S. (2012) Status of 25-hydroxyvitamin D deficiency and effect of vitamin D receptor gene polymorphisms on bone mineral density in thalassemia patients of North India. *Hematology*. 2012 Sep;17(5):291-6

153) Dalal A, Bhavani G SL, Togarrati PP, Bierhals T, Nandineni MR, Danda S, Danda D, Shah H, Vijayan S, Gowrishankar K, **Phadke SR**, Bidchol AM, Rao AP, Nampoothiri S, Kutsche K, Girisha KM. (2012) Analysis of the WISP3 gene in Indian families with progressive pseudorheumatoid dysplasia. *Am J Med Genet A*. 2012 Nov;158A(11):2820-8.

154) Bashyam MD, Chaudhary AK, Reddy EC, Reddy V, Acharya V, Nagarajaram HA, Devi AR, Bashyam L, Dalal AB, Gupta N, Kabra M, Agarwal M, Phadke SR, Tainwala R, Kumar R, Hariharan SV. A founder ectodysplasin A receptor (EDAR) mutation results in a high frequency of the autosomal recessive form of hypohidrotic ectodermal dysplasia in India. *Br J Dermatol*. 2012 Apr;166(4):819-29. doi: 10.1111/j.1365-2133.2011.10707.x. Epub 2012 Mar 5. PMID: 22032522.

155) Aggarwal S, Bogula VR, Mandal K, Kumar R, **Phadke SR**. (2012) Aetiological spectrum of mental retardation & developmental delay in India. *Indian J Med Res*. 2012 Sep;136(3):436-44.

156) **Phadke S**, Agarwal M. (2012) Neural tube defects: A need for population-based prevention program. *Indian J Hum Genet*. 2012 May;18(2):145-7.

157) Tamhankar PM, Boggula V, Girisha KM, **Phadke SR**. (2012) Profile of patients with Von Gierke disease from India. *Indian Pediatr*. Mar;49(3):228-30

158) Jain S, Panigrahi I, Gupta R, **Phadke SR**, Agarwal S. (2012) Multiplex quantitative fluorescent polymerase chain reaction for detection of aneuploidies. *Test Mol Biomarkers*. Jun;16(6):624-7

159) **Phadke S R (2012)** Arthralgia and limb pains: Pseudo-rheumatoid genetic disorders. *Indian Journal of Rheumatology* Vol. 7(1), Supplement: 57-68

160) Kaur A, **Phadke SR**. (2012) Analysis of short stature cases referred for genetic evaluation. *Indian J Pediatr*. Dec;79(12):1597-600.

161) Sardhara J, Behari S, Jaiswal AK, Srivastava A, Sahu R N, Mehrotra A, **Phadke S**, Singh U (2013) Syndromic versus nonsyndromic atlantoaxial dislocation: do clinico-radiological differences have a bearing on management? Accepted in *Acta Neurochirurgica*

162) Aggarwal S, **Phadke SR**. Recurrence of urorectal septum malformation sequence spectrum anomalies in siblings: Time to explore the genetics. *Am J Med Genet A*. 2013 Jul;161(7):1718-21. \$

163) Kumar A, Agarwal S, Agarwal D, **Phadke SR**. Myotonic dystrophy type 1 (DM1): A triplet repeat expansion disorder. *Gene*. 2013 Jun 15;522(2):226-30.

164) Kaur A, Khan F, Agrawal SS, Kapoor A, Agarwal SK, **Phadke SR**. Cytochrome P450 (CYP2C9\*2,\*3) & vitamin-K epoxide reductase complex (VKORC1 -1639G<A) gene

polymorphisms & their effect on acenocoumarol dose in patients with mechanical heart valve replacement. Indian J Med Res. 2013 Jan;137(1):203-9.

165) Agarwal M, Gupta R, Boggula V, **Phadke S.** Utility of chromosomal microarray in five cases with cytogenetic abnormalities detected by traditional karyotype. Clin Genet. 2013 Feb 21.

166) Agarwal M, **Phadke SR.** Atlantoaxial dislocation in a child affected by warfarin embryopathy: a case report. Clin Dysmorphol. 2013 Jul;22(3):124-6..

167) Dimopoulos A, Fischer B, Gardeitchik T, Schröter P, Kayserili H, Schlack C, Li Y, Brum JM, Barisic I, Castori M, Spaich C, Fletcher E, Mahayri Z, Bhat M, Girisha KM, Lachlan K, Johnson D, **Phadke S**, Gupta N, Simandlova M, Kabra M, David A, Nijtmans L, Chitayat D, Tuysuz B, Brancati F, Mundlos S, Van Maldergem L, Morava E, Wollnik B, Kornak U. (2013) Genotype-phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Mol Genet Metab. 2013 Nov;110(3):352-61

168) Shukla A, Taywade O, Stephen J, Gupta D, **Phadke SR.**(2013) Fibrodysplasia Ossificans Progressiva: Three Indian Patients with Mutation in the ACVR1 Gene. Indian J Pediatr. 2013

169) **PhadkeS**, Gowda M (2013) Genetic testing in children. Indian Pediatr. 50(9):823-7.

170) Sharma N, Dixit P, Awasthi S, **Phadke SR.** (2013) Genetic variations of the FCER2 gene and asthma susceptibility in north Indian children: a case-control study. Biomarkers. 2013 Dec;18(8):660-7

171) Gupta R, Agarwal M, Boqqua VR, Phadke RV, **Phadke SR.** (2014) Hemiconvulsion-hemiplegia-epilepsy syndrome with 1q44 microdeletion: causal or chance association. Am J Med Genet A. Jan;164A(1):186-9.

172) Singh S, Kumar A, Agarwal S, **Phadke SR**, Jaiswal Y.(2014) Genetic insight of schizophrenia: past and future perspectives. Gene. Feb 10;535(2):97-100.

173) Tsurusaki Y, Koshimizu E, Ohashi H, **Phadke S**, Kou I, Shiina M, Suzuki T, Okamoto N, Imamura S, Yamashita M, Watanabe S, Yoshiura K, Kodera H, Miyatake S, Nakashima M, Saitsu H, Ogata K, Ikegawa S, Miyake N, Matsumoto N (2014) De novo SOX11 mutations cause Coffin-Siris syndrome. Nat Commun. 2014 Jun 2;5:4011. @

174) Bidhol AM, Dalal A, Shah H, S S, Nampoothiri S, Kabra M, Gupta N, Danda S, Gowrishankar K, **Phadke SR**, Kapoor S, Kamate M, Verma IC, Puri RD, Sankar VH, Devi AR, Patil SJ, Ranganath P, Jain SJ, Agarwal M, Singh A, Mishra P, Tamhankar PM, Gopinath PM, Nagarajaram HA, Satyamoorthy K, Girisha KM. GALNS mutations in Indian patients with mucopolysaccharidosis IVA. Am J Med Genet A. 2014 Nov;164A(11):2793-801

175) Stephen J, Shukla A, Dalal A, Girisha KM, Shah H, Gupta N, Kabra M, Dabaghao P, **Phadke SR.** Mutation spectrum of COL1A1 and COL1A2 genes in Indian patients with osteogenesis imperfecta. Am J Med Genet A. 2014 Jun;164A(6):1482-

176) Agrawal D, Prakash S, Misra MK, **Phadke SR**, Agrawal S. Implication of HLA-G 5' upstream regulatory region polymorphisms in idiopathic recurrent spontaneous abortions. Reprod Biomed Online. 2014 Oct 16. doi: 10.1016/j.rbmo.2014.09.015. [Epub ahead of print]

177) Agarwal M, Joshi K, Bhatia V, Gopalakrishnan V, Dabaghao P, Das V, Pandey A, Kumar M, **Phadke SR.** Feasibility Study of an Outreach Program of Newborn Screening in Uttar Pradesh. Indian J Pediatr. 2014 Nov 1

178) Dalal AB, Ranganath P, **Phadke SR**, Kabra M, Danda S, Puri RD, V H S, Gupta N, Patil SJ, Mandal K, Tamhankar P, Aggarwal S, Agarwal M. Prenatal diagnosis in India is not limited to sex selection. Genet Med. 2014 Oct 16. \$

179) Nandagopalan RS, **Phadke SR**, Dalal AB, Ranganath P. Novel mutations in PRG4 gene in two Indian families with camptodactyly-arthropathy-coxa vara-pericarditis (CACP) syndrome. *Indian J Med Res.* 2014 Aug;140(2):221-6.

180) Patil SJ, Rai GK, Bhat V, Ramesh VA, Nagarajaram HA, Matalia J, **Phadke SR**. Distal arthrogryposis type 5D with a novel ECEL1 gene mutation. *Am J Med Genet A.* 2014 Nov;164A(11):2857-62. doi: 10.1002/ajmg.a.36702. Epub 2014 Aug 5.

181) Agarwal D, Stephen J, Phadke RV, **Phadke SR**. Severe short stature, profound microcephaly, developmental brain abnormality, agenesis of optic disc and retinal vessels, and bilateral cryptorchidism in two male siblings: a new lethal recessively inherited syndrome. *Clin Dysmorphol.* 2014 Oct;23(4):117-20.

182) Siddesh A, Parveen F, Misra MK, **Phadke SR**, Agrawal S. Platelet-specific collagen receptor glycoprotein VI gene variants affect recurrent pregnancy loss. *Fertil Steril.* 2014 Oct;102(4):1078-1084

183) Sharma N, Jaiswal I, Mandal RK, **Phadke SR**, Awasthi S. Genetic variation of TBX21 gene increases risk of asthma and its severity in Indian children. *J Hum Genet.* 2014 Aug;59(8):437-43

184) Gupta D, Gupta V, Singh V, Chawla S, Parveen F, Agrawal S, **Phadke SR**. Study of Polymorphisms in CX3CR1, PLEKHA1 and VEGF genes as risk factors for age-related macular degeneration in Indian patients. *Arch Med Res.* 2014 Aug;45(6):489-94.

185) Kandasamy S, Saxena D, Kishore Y, **Phadke SR**. Williams syndrome: a case series. *Indian Pediatr.* 2014 May;51(5):411-2.

186) Boggula VR, Shukla A, Danda S, Hariharan SV, Nampoothiri S, Kumar R, **Phadke SR**. Clinical utility of multiplex ligation-dependent probe amplification technique in identification of aetiology of unexplained mental retardation: a study in 203 Indian patients. *Indian J Med Res.* 2014 Jan;139(1):66-75.

187) Girisha KM, Abdollahpour H, Shah H, Bhavani GS, Graham JM Jr, Boggula VR, **Phadke SR**, Kutsche K. A syndrome of facial dysmorphism, cubital pterygium, short distal phalanges, swan neck deformity of fingers, and scoliosis. *Am J Med Genet A.* 2014 Apr;164A(4):1035-40.

188) Sharma N, Awasthi S, **Phadke SR**. A novel genotyping method for detection of the CRHR1 (rs1396862: C>T) gene variation among North Indian population. *Mol Biol Rep.* 2014;41(5):2809-13

189) Narayanan DL, **Phadke SR**. Infantile Systemic Hyalinosis with Mutation in ANTXR2. *Indian J Pediatr.* 2016 Jan 25.

190) **Phadke SR** Lysosomal Storage Disorders: Present and Future. *Indian Pediatr.* 2015 ;52(12):1025-6.

191) Uttarill A, Ranganath P, Jain SJ, Prasad CK, Sinha A, Verma IC, **Phadke SR**, Puri RD, Danda S, Muranjan MN, Jevalikar G, Nagarajaram HA, Dalal AB. Novel mutations of the arylsulphatase B (ARSB) gene in Indian patients with mucopolysaccharidosis type VI. *Indian J Med Res.* 2015 Oct;142(4):414-25.

192) Tuteja M, Agarwal M, **Phadke SR**. Knowledge of Cord Blood Banking in General Population and Doctors: A Questionnaire Based Survey. *Indian J Pediatr.* 2015 Nov 21.

193) Dalal A, Bhowmik AD, Agarwal D, **Phadke SR**. Exome sequencing and homozygosity mapping for identification of genetic aetiology for spastic ataxia in a consanguineous family. *Indian J Med Res.* 2015 Aug;142(2):220-4.

194) Bhavani GS, Shah H, Dalal AB, Shukla A, Danda S, Aggarwal S, **Phadke SR**, Gupta N, Kabra M, Gowrishankar K, Gupta A, Bhat M, Puri RD, Bijarnia-Mahay S, Nampoothiri S, Mohanasundaram KM, Rajeswari S, Kulkarni AM, Kulkarni ML, Ranganath P, Ramadevi AR, Hariharan SV, Girisha KM. Novel and recurrent mutations in WISP3 and an atypical phenotype. *Am J Med Genet A.* 2015 Oct;167A(10):2481-4.

195) Bidchol AM, Dalal A, Trivedi R, Shukla A, Nampoothiri S, Sankar VH, Danda S, Gupta N, Kabra M, Hebbar SA, Bhat RY, Matta D, Ekbote AV, Puri RD, **Phadke SR**, Gowrishankar K, Aggarwal S, Ranganath P, Sharda S, Kamate M, Datar CA, Bhat K, Kamath N, Shah H, Krishna S, Gopinath PM, Verma IC, Nagarajaram HA, Satyamoorthy K, Girisha KM. Recurrent and novel GLB1 mutations in India. *Gene*. 2015 Aug 10;567(2):173-81. doi: 10.1016/j.gene.2015.04.078. Epub 2015 Apr 30.

196) Shukla A, Mandal K, Patil SJ, Kishore Y, **Phadke SR**, Girisha KM. Co-occurrence of a de novo Williams and 22q11.2 microdeletion syndromes. *Am J Med Genet A*. 2015 Aug;167A(8):1927-31. doi: 10.1002/ajmg.a.37116. Epub 2015 Apr 21.

197) Muthuswamy S, Bhalla P, Agarwal S, **Phadke SR**. Performance of QF-PCR in targeted prenatal aneuploidy diagnosis: Indian scenario. *Gene*. 2015 May 10;562(1):55-61. doi: 10.1016/j.gene.2015.02.028. Epub 2015 Feb 25.

198) Tuteja M, Bidchol AM, Girisha KM, **Phadke SR**. White matter changes in GM1 gangliosidosis. *Indian Pediatr*. 2015 Feb;52(2):155-6.

199) Kumar A, Agarwal S, **Phadke SR**, Pradhan S. Application of a reliable and rapid polymerase chain reaction based method in the diagnosis of myotonic dystrophy type 1 (DM1) in India. *Meta Gene*. 2014 Jan 15;2:106-13.

200) Shukla A, **Phadke SR**. Chondrodysplasia punctata tibia metacarpal type: report of a 1.5 year old child with severe short stature and extensive calcific stippling. *Clin Dysmorphol*. 2015 Jul;24(3):118-21.

201) Sharma N, Awasthi S, **Phadke SR**. A Mutagenic Primer Assay for Genotyping of the CRHR1 Gene Rare Variant rs1876828 (A/G) in Asians: A Cost-Effective SNP Typing. *J Clin Lab Anal*. 2014 Dec 26.

202) Agrawal D, Prakash S, Misra MK, **Phadke SR**, Agrawal S. Implication of HLA-G 5' upstream regulatory region polymorphisms in idiopathic recurrent spontaneous abortions. *Reprod Biomed Online*. 2015 Jan;30(1):82-91.

203) Stephen J, Girisha KM, Dalal A, Shukla A, Shah H, Srivastava P, Kornak U, **Phadke SR**. Mutations in patients with osteogenesis imperfecta from consanguineous Indian families. *Eur J Med Genet*. 2015 Jan;58(1):21-7.

204) **Phadke S**. S.S. Agarwal. *Natl Med J India*. 2014 Jan-Feb;27(1):44-5. No abstract available.

205) Agarwal D, Nayak SS, Adiga PK, **Phadke SR**, Girisha KM. Symmetrical terminal transverse limb deficiencies. *Indian J Pediatr*. 2015 May;82(5):478-9.

206) Agarwal M, Joshi K, Bhatia V, Gopalakrishnan V, Dabadghao P, Das V, Pandey A, Kumar M, **Phadke SR**. Feasibility study of an outreach program of newborn screening in Uttar Pradesh. *Indian J Pediatr*. 2015 May;82(5):427-32. doi: 10.1007/s12098-014-1557-6. Epub 2014 Nov 1.

207) Dalal AB, Ranganath P, **Phadke SR**, Kabra M, Danda S, Puri RD, Sankar VH, Gupta N, Patil SJ, Mandal K, Tamhankar P, Aggarwal S, Agarwal M. Prenatal diagnosis in India is not limited to sex selection. *Genet Med*. 2015 Jan;17(1):88. doi: 10.1038/gim.2014.149. No abstract available.

208) Nandagopalan RS, **Phadke SR**, Dalal AB, Ranganath P. Novel mutations in PRG4 gene in two Indian families with camptodactyly-arthropathy-coxa vara-pericarditis (CACP) syndrome. *Indian J Med Res*. 2014 Aug;140(2):221-6.

209) Bidchol AM, Dalal A, Shah H, S S, Nampoothiri S, Kabra M, Gupta N, Danda S, Gowrishankar K, **Phadke SR**, Kapoor S, Kamate M, Verma IC, Puri RD, Sankar VH, Devi AR, Patil SJ, Ranganath P, Jain SJ, Agarwal M, Singh A, Mishra P, Tamhankar PM, Gopinath PM, Nagarajaram HA, Satyamoorthy K, Girisha KM. GALNS mutations in Indian patients with mucopolysaccharidosis IVA. *Am J Med Genet A*. 2014 Nov;164A(11):2793-801

210) **Phadke SR**, Kar A, Bhowmik AD, Dalal A. Complex Camptosynpolydactyly and Mesoaxialsynostotic syndactyly with phalangeal reduction are allelic disorders. *Am J Med Genet A*. 2016 Apr 4. @

211) Mandal K, Ray S, Saxena D, Srivastava P, Moirangthem A, Ranganath P, Gupta N, Mukhopadhyay S, Kabra M, **Phadke SR**. Pycnodynostosis: mutation spectrum in five unrelated Indian children. *Clin Dysmorphol*. 2016 Apr 18.

212) Ranganath P, Stephen J, Iyengar R, **Phadke SR**. Worsening of Callus Hyperplasia after Bisphosphonate Treatment in Type V Osteogenesis Imperfecta. *Indian Pediatr*. 2016 Mar 8;53(3):250-2.

213) Bhavani GS, Shah H, Shukla A, Gupta N, Gowrishankar K, Rao AP, Kabra M, Agarwal M, Ranganath P, Ekbote AV, **Phadke SR**, Kamath A, Dalal A, Girisha KM. Clinical and mutation profile of multicentric osteolysis nodulosis and Arthropathy. *Am J Med Genet A*. 2016 Feb;170(2):410-7.

214) Thevenon J, Duplomb L, **Phadke SR**, Eguether T, Saunier A, Avila M, Carmignac V, Brueal AL, St-Onge J, Duffourd Y, Pazour GJ, Franco B, Attie-Bitach T, Masurel-Paulet A, Rivière JB, Cormier-Daire V, Philippe C, Faivre L, Thauvin-Robinet C. Autosomal recessiveIFT57 hypomorphic mutation cause ciliary transport defect in unclassified oral-facial-digital syndrome with short stature and brachyomesophalangia. *Clin Genet*. 2016 Dec;90(6):509-517. @

215) Narayanan DL, Srivastava P, Mandal K, Gambhir PS, **Phadke SR**. Hunter Syndrome in Northern India: Clinical features and Mutation Spectrum. *Indian Pediatr*. 2016 Feb 8;53(2):134-6.

216) Mandal K, Agarwal M, Boggula VR, Patil SJ, **Phadke SR**. Complex chromosomal rearrangement involving five chromosomes: deciphering genomic imbalances in an apparently balanced chromosomal translocation. *Clin Dysmorphol*. 2016 Apr;25(2):63-7.

217) Boggula VR, Agarwal M, Kumar R, Awasthi S, **Phadke SR**. Recurrent benign copy number variants & issues in interpretation of variants of unknown significance identified by cytogenetic microarray in Indian patients with intellectual disability. *Indian J Med Res*. 2015 Dec;142(6):699-712

218) Tuteja M, Agarwal M, **Phadke SR**. Knowledge of Cord Blood Banking in General Population and Doctors: A Questionnaire Based Survey. *Indian J Pediatr*. 2016 Mar;83(3):238-41

219) Martin CA, Murray JE, Carroll P, Leitch A, Mackenzie KJ, Halachev M, Fetit AE, Keith C, Bicknell LS, Fluteau A, Gautier P, Hall EA, Joss S, Soares G, Silva J, Bober MB, Duker A, Wise CA, Quigley AJ, **Phadke SR**; Deciphering Developmental Disorders Study., Wood AJ, Vagnarelli P, Jackson AP. Mutations in genes encoding condensin complex proteins cause microcephaly through decatenation failure at mitosis. *Genes Dev*. 2016 Oct 1;30(19):2158-2172.:@

220) Gupta D, Rao R, Girisha KM, Stephen J, **Phadke SR**. Unusual skin manifestations in a patient with menkes disease. *Am J Med Genet A*. 2016 Nov;170(11):3039-3040.

221) Singh B, Mandal K, Lallar M, Narayanan DL, Mishra S, Gambhir PS, **Phadke SR**. Next Generation Sequencing in Diagnosis of MLPA Negative Cases Presenting as Duchenne/ Becker Muscular Dystrophies. *Indian J Pediatr*. 2017 Sep 12.

222) Lallar M, Srivastava A, **Phadke SR**. Hyperekplexia: A forgotten diagnosis clinched by next-generation sequencing. *Neurol India*. 2017 Sep-Oct;65(5):1065-1067.

223) Siddesh A, Gupta G, Sharan R, Agarwal M, **Phadke SR**. Spectrum of prenatally detected central nervous system malformations: Neural tube defects continue to be the leading foetal malformation. *Indian J Med Res*. 2017 Apr;145(4):471-478. \$

224) Mangla P, Gambhir PS, Sudhanshu S, Srivastava P, Rai A, Bhatia V, **Phadke SR**. Pyruvate Carboxylase Deficiency Mimicking Diabetic Ketoacidosis. Indian J Pediatr. 2017 Dec;84(12):959-960

225) Kruszka P, Porras AR, Addissie YA, Moresco A, Medrano S, **Phadke SR**, Obregon MG, Linguraru MG, Muenke M. Noonan syndrome in diverse populations. Am J Med Genet A. 2017 Sep;173(9):2323-2334.

226) Narayanan DL, Pandey H, Moirangthem A, Mandal K, Gupta R, Puri RD, Patil SJ, **Phadke SR**. Hotspots in PTPN11 Gene Among Indian Children With Noonan Syndrome. Indian Pediatr. 2017 Aug 15;54(8):638-643.

227) Narayanan DL, **Phadke SR**. A novel variant in MED12 gene: Further delineation of phenotype. Am J Med Genet A. 2017 Aug;173(8):2257-2260.

228) Lallar M, **Phadke SR**. Fetal intra abdominal umbilical vein varix: Case series and review of literature. Indian J Radiol Imaging. 2017 Jan-Mar;27(1):59-61. \$

229) Bruel AL, Franco B, Duffourd Y, Thevenon J, Jego L.....**Phadke SR**, Cormier-Daire V, ..... Thauvin-Robinet C. Fifteen years of research on oral-facial-digital syndromes: from 1 to 16 causal genes. J Med Genet. 2017 Jun;54(6):371-380. @

230) Moirangthem A, Tuteja Bhatia M, Srivastava P, Mandal K, Rai A, **Phadke SR**. Expansion of the phenotypic spectrum in three families of methyl CpG-binding protein 2 duplication syndrome. Clin Dysmorphol. 2017 Apr;26(2):73-77

231) Salian S, Cho TJ, **Phadke SR**, Gowrishankar K, Bhavani GS, Shukla A, Jagadeesh S, Kim OH, Nishimura G, Girisha KM. Additional three patients with Smith-McCort dysplasia due to novel RAB33B mutations. Am J Med Genet A. 2017 Mar;173(3):588-595.

232) Srivastava P, Gambhir PS, **Phadke SR**. KBG syndrome: 16q24.3 microdeletion in an Indian patient. Clin Dysmorphol. 2017 Jul;26(3):161-166.

233) Srivastava P, Pandey H, Agarwal D, Mandal K, **Phadke SR**. Spondyloepiphyseal dysplasia Omani type: CHST3 mutation spectrum and phenotypes in three Indian families. Am J Med Genet A. 2017 Jan;173(1):163-168.

234) Gupta A, Kapoor A, **Phadke SR**, Sinha A, Kashyap S, Khanna R, Kumar S, Garg N, Tewari S, Goel P. Use of strain, strain rate, tissue velocity imaging, and endothelial function for early detection of cardiovascular involvement in patients with beta-thalassemia. Ann Pediatr Cardiol. 2017 May-Aug;10(2):158-166

235) Verma A, Hemlata, Elhence P, **Phadke SR**, Neyaz Z. Posterior reversible encephalopathy syndrome following blood transfusion in a patient with factor X deficiency: Is it an unusual systemic manifestation of an adverse transfusion reaction? TransfusApher Sci. 2017 Dec 6.

236) Kar A, **Phadke SR**, Das Bhowmik A, Dalal A. Whole exome sequencing reveals a mutation in ARMC9 as a cause of mental retardation, ptosis, and polydactyly. Am J Med Genet A. 2018 Jan;176(1):34-40.

237) Tuteja M, Agarwal D, **Phadke SR**. Double Segment Chromosomal Imbalance due to Inherited Chromosomal Translocation: Detection by Cytogenetic Microarray. Indian Pediatr. 2017 Oct 15;54(10):879-881.

238) Moirangthem A, **Phadke SR**. Socio-demographic Profile and Economic Burden of Treatment of Transfusion Dependent Thalassemia. Indian J Pediatr. 2018 Feb;85(2):102-107

239) Lallar M, Rai A, Srivastava P, Mandal K, Gupta N, Kabra M, **Phadke SR**. Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Indian Pediatr. 2018 Feb 9. [Epub ahead of print]

240) Sheth J, Pancholi D, Mistri M, Nath P, Ankleshwaria C, Bhavsar R, Puri R, **Phadke S**, Sheth F. Biochemical and molecular characterization of adult patients with type I Gaucher

disease and carrier frequency analysis of Leu444Pro –a common Gaucher disease mutation in India. *BMC Med Genet.* 2018 Oct 1;19(1):178.

241) Kashyap S, Kumar S, Agarwal V, Misra DP, **Phadke SR**, Kapoor A. Gene expression profiling of coronary artery disease and its relation with different severities. *J Genet.* 2018 Sep;97(4):853-867.

242) Rai A, Puri RD, **Phadke SR**. Extending the phenotype and an ECEL1 gene mutation in distal arthrogryposis type 5D. *Clin Dysmorphol.* 2018 Oct;27(4):130-13

243) Sharma A, Poddar U, Agnihotry S, **Phadke SR**, Yachha SK, Aggarwal R. Spectrum of genomic variations in Indian patients with progressive familial intrahepaticcholestasis. *BMC Gastroenterol.* 2018 Jul 4;18(1):107.

244) Mishra S, Srivastava A, Mandal K, **Phadke SR**. Study of the association offorkhead box P3 (FOXP3) gene polymorphisms with unexplained recurrent spontaneousabortions in Indian population. *J Genet.* 2018 Jun;97(2):405-410.

245) Lallar M, Srivastava P, **Phadke SR**. A large interstitial 11q deletion withisolated mild intellectual disability: review of the literature forgenotype-phenotype correlation. *Clin Dysmorphol.* 2018 Oct;27(4):142-144

246) Singh B, Srivastava P, **Phadke SR**. Sequence variations in TENM3 gene causingeye anomalies with intellectual disability: Expanding the phenotypic spectrum.*Eur J Med Genet.* 2018 May 9. pii: S1769-7212(17)30743-7.

247) Saxena D, Srivastava P, Tuteja M, Mandal K, **Phadke SR**. Phenotypiccharacterization of derivative 22 syndrome: case series and review. *J Genet.* 2018Mar;97(1):205-211.

248) **Phadke SR**, Puri RD, Ranganath P. Prenatal screening for genetic disorders: Suggested guidelines for the Indian Scenario. *Indian J Med Res.* 2017Dec;146(6):689-699.

249) Narayanan DL, **Phadke SR**. Concepts, Utility and Limitations of Cord Blood Banking: What Clinicians Need to Know. *Indian J Pediatr.* 2018 Mar 20.

250) Mishra S, Pandey H, Srivastava P, Mandal K, **Phadke SR**. Connexin 26 (GJB2)Mutations Associated with Non-Syndromic Hearing Loss (NSHL). *Indian J Pediatr.* 2018 Mar 15.

251) Puri RD, Kapoor S, Krishnani PS, Dalal A, Gupta N, Muranjan M, **PhadkeSR**, Sachdeva A, Verma IC, Mistry PK; Gaucher Disease Task Force. Diagnosis and Management of Gaucher Disease in India - Consensus Guidelines of the Gaucher Disease Task Force of the Society for Indian Academy of Medical Genetics and the Indian Academy of Pediatrics. *Indian Pediatr.* 2018 Feb 15;55(2):143-153.

252) Chang HR, Cho SY, Lee JH, Lee E, Seo J, Lee HR, Cavalcanti DP, Mäkitie O, Valta H, Girisha KM, Lee C, Neethukrishna K, Bhavani GS, Shukla A, NampoothiriS, **Phadke SR**, Park MJ, Ikegawa S, Wang Z, Higgs MR, Stewart GS, Jung E, Lee MS, Park, JH, Lee EA, Kim H, Myung K, Jeon W, Lee K, Kim D, Kim OH, Choi M, Lee HW, Kim Y, Cho TJ. Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. *Am J Hum Genet.* 2019 Mar 7;104(3):439-453. @

253) Narayanan DL, Matta D, Gupta N, Kabra M, Ranganath P, Aggarwal S, **Phadke SR**, Datar C, Gowrishankar K, Kamate M, Jain JMN, Dalal A. Spectrum of ARSA variations in Asian Indian patients with Arylsulfatase A deficient metachromatic leukodystrophy. *J Hum Genet.* 2019 Apr;64(4):323-331.

254) Dowsett L, Porras AR, Kruszka P, Davis B, Hu T, Honey E, Badoe E, Thong MK, Leon E, Girisha KM, Shukla A, Nayak SS, Shotelersuk V, Megarbane A, **Phadke S**, Sirisena ND, Dissanayake VHW, Ferreira CR, Kisling MS, Tanpaiboon P, Uwineza A, Mutesa L, Tekendo-Ngongang C, Wonkam A, Fieggen K, Batista LC, Moretti-Ferreira D, Stevenson RE, Prijoles EJ, Everman D, Clarkson K, Worthington J, Kimonis V, Hisama F, Crowe C, Wong P, Johnson K, Clark RD, Bird L, Masser-Frye D, McDonald M, Willems P, Roeder E, Saitta S, Anyane-Yeoba K, Demmer L, Hamajima N, Stark Z, Gillies G, Hudgins L, Dave U, Shalev S, Siu V, Ades A, Dubbs H, Raible S, Kaur M, Salzano E, Jackson L,

Deardorff M, Kline A, Summar M, Muenke M, Linguraru MG, Krantz ID. Cornelia de Lange syndrome in diverse populations. *Am J Med Genet A*. 2019 Feb;179(2):150-158.

255) Rai A, Narayanan DL, **Phadke SR**. Malan syndrome: Extension of genotype and phenotype spectrum. *Am J Med Genet A*. 2018 Dec;176(12):2896-2900

256) Lallar M, Srivastava P, Rai A, Saxena D, Mandal K, **Phadke SR**. Cytogenetic microarray in structurally normal and abnormal foetuses: a five year experience elucidating increasing acceptance and clinical utility. *J Genet*. 2019 Mar;98(1). \$

257) Sekiguchi F, Tsurusaki Y, Okamoto N, Teik KW, Mizuno S, Suzumura H, Isidor B, Ong WP, Haniffa M, White SM, Matsuo M, Saito K, **Phadke S**, Kosho T, Yap P, Goyal M, Clarke LA, Sachdev R, McGillivray G, Leventer RJ, Patel C, Yamagata T, Osaka H, Hisaeda Y, Ohashi H, Shimizu K, Nagasaki K, Hamada J, Dateki S, Sato T, Chinen Y, Awaya T, Kato T, Iwanaga K, Kawai M, Matsuoka T, Shimoji Y, Tan TY, Kapoor S, Gregersen N, Rossi M, Marie-Laure M, McGregor L, Oishi K, Mehta L, Gillies G, Lockhart PJ, Pope K, Shukla A, Girisha KM, Abdel-Salam GMH, Mowat D, Coman D, Kim OH, Cordier MP, Gibson K, Milunsky J, Liebelt J, Cox H, El Chehadeh S, Toutain A, Saida K, Aoi H, Minase G, Tsuchida N, Iwama K, Uchiyama Y, Suzuki T, Hamanaka K, Azuma Y, Fujita A, Imagawa E, Koshimizu E, Takata A, Mitsuhashi S, Miyatake S, Mizuguchi T, Miyake N, Matsumoto N. Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. *J Hum Genet*. 2019 Sep @

258) Mishra S, Rai A, Srivastava P, **Phadke SR**. A mild phenotype of LGI4-Related arthrogryposis multiplex congenita with intrafamilial variability. *Eur J Med Genet*. 2019 Sep 9:103756.

259) Gupta L, Ahmed S, Singh B, Prakash S, **Phadke S**, Aggarwal A. Novel NLRP12 variant presenting with familial cold autoimmunity syndrome phenotype. *Ann Rheum Dis*. 2019 Aug 24. pii: annrheumdis-2019-216158.

260) Moirangthem A, Mandal K, Ghosh A, **Phadke SR**. Vici Syndrome with a Novel Mutation in EPG5. *Indian Pediatr*. 2019 Jul 15;56(7):603-605.

261) Pani K, Sharma S, Murari M, Yadav M, **Phadke SR**, Agarwal S. Clinico-hematological Profile of Hb E-β Thalassemia-Prospective Analysis in a tertiary Care Centre. *J Assoc Physicians India*. 2018 Jun;66(6):42-45

262) Rauen KA, Alsaegh A, Ben-Shachar S, Berman Y, Blakeley J, Cordeiro I, Elgersma Y, Evans DG, Fisher MJ, Frayling IM, George J, Huson SM, Kerr B, Khire U, Korf B, Legius E, Messiaen L, van Minkelen R, Nampoothiri S, Ngeow J, Parada LF, **Phadke SR**, Pillai A, Plotkin SR, Puri R, Raji A, Ramesh V, Ratner N, Shankar SP, Sharda S, Tambe A, Vikkula M, Widemann BC, Wolkenstein P, Upadhyaya M. First International Conference on RASopathies and Neurofibromatoses in Asia: Identification and advances of new therapeutics. *Am J Med Genet A*. 2019, Jun;179(6):1091-1097.

263) Kruszka P, Addissie YA, Tekendo-Ngongang C, Jones KL, Savage SK, Gupta N, Sirisena ND, Dissanayake VHW, Paththinige CS, Aravena T, Nampoothiri S, Yesodharan D, Girisha KM, Patil SJ, Jamuar SS, Goh JC, Utari A, Sihombing N, Mishra R, Chitrakar NS, Iriele BC, Lulseged E, Megarbane A, Uwineza A, Oyenusiee, Olopade OB, Fasanmade OA, Duenas-Roque MM, Thong MK, Tung JYL, Mok GTK, Fleischer N, Rwegerera GM, de Herreros MB, Watts J, Fieggen K, Huckstadt V, Moresco A, Obregon MG, Hussen DF, Ashaat NA, Ashaat EA, Chung BHY, Badoe E, Faradz SMH, El Ruby MO, Shotelersuk V, Wonkam A, Ekure EN, **Phadke SR**, Richieri-Costa A, Muenke M. Turner syndrome in diverse populations. *Am*

J Med Genet A.2020 Feb;182(2):303-313. doi: 10.1002/ajmg.a.61461. Epub 2019 Dec 19. PMID:31854143.

264) Masih S, Moirangthem A, **Phadke SR**. Renpenning syndrome in an Indian patient. Am J Med Genet A. 2020 Feb;182(2):293-295. doi: 10.1002/ajmg.a.61457. Epub 2019 Dec 16. PMID: 31840915.

265) Arora V, Setia N, Dalal A, Vanaja MC, Gupta D, Razdan T, **Phadke SR**, Saxena R, Rohtagi A, Verma IC, Puri RD. Sialidosis type II: Expansion of phenotypic spectrum and identification of a common mutation in seven patients. Mol Genet Metab Rep. 2020 Jan 11;22:100561. doi: 10.1016/j.ymgmr.2019.100561. PMID:31956508; PMCID: PMC6957780.

266) Pasumarthi, D., Gupta, N., Sheth, J. *et al.* Identification and characterization of 30 novel pathogenic variations in 69 unrelated Indian patients with Mucolipidosis Type II and Type III. *J Hum Genet* (2020). <https://doi.org/10.1038/s10038-020-0797-8>

267) Nilay M, **Phadke SR**. Pearson Syndrome: Spontaneously Recovering Anemia and Hypoparathyroidism [published online ahead of print, 2020 Jun 15]. *Indian J Pediatr*. 2020;10.1007/s12098-020-03333-9. doi:10.1007/s12098-020-03333-9

268) Danda S, Mohan S, Devaraj P, *et al.* Founder effects of the homogentisate 1,2 dioxygenase (HGD) gene in a gypsy population and mutation spectrum in the gene among alkaptonuria patients from India. *Clin Rheumatol*. 2020;39(9):2743-2749. doi:10.1007/s10067-020-05020-8

269) Rai A, Mandal K, Saxena D, Lallar M, **Phadke SR**. Distal Arthrogryposis: A Clue to the Etiology of Neonatal Cholestasis. *Indian J Pediatr*. 2020;87(10):869-870. doi:10.1007/s12098-020-03248-5

270) Girisha KM, Pande S, Dalal A, **Phadke SR**. Untapped opportunities for rare disease gene discovery in India [published online ahead of print, 2020 Sep 10]. *Am J Med Genet A*. 2020;10.1002/ajmg.a.61866. doi:10.1002/ajmg.a.61866

271) Finsterer J, Nilay M, **Phadke SR**. Pearson Syndrome: Spontaneously Recovering Anemia and Hypoparathyroidism - Correspondence. *Indian J Pediatr*. 2021 Feb;88(2):209-210.doi: 10.1007/s12098-020-03467-w. Epub 2020 Aug 7

272) Narayanan DL, Tuteja M, McIntyre AD, Hegele RA, Calmels N, Obringer C, Laugel V, Mandal K, **Phadke SR**. Clinical and Mutation Spectra of Cockayne Syndrome in India. *Neurol India*. 2021 Mar-Apr;69(2):362-366. doi: 10.4103/0028-3886.314579.

273) Gupta L, Ahmed S, Singh B, Prakash S, **Phadke SR**, Aggarwal A Novel NLRP12 variant presenting with familial cold autoimmunity syndrome phenotype. *Ann Rheum Dis*. 2021 Jul;80(7):e117. doi: 10.1136/annrheumdis-2019-216158. Epub 2019 Aug 24. PMID: 31446425 No abstract available.

274) Suri D, Rawat A, Jindal AK, Vignesh P, Gupta A, Pilania RK, Joshi V, Arora K, Kumrah R, Anjani G, Aggarwal A, **Phadke SR**, Aboobacker FN, George B, Edison ES, Desai M, Taur P, Gowri V, Pandrowala AA, Bhattad S, Kanakia S, Gottorno M, Ceccherini I, Almeida de Jesus A, Goldbach-Mansky R, Hershfield MS, Singh S. Spectrum of Systemic Auto-Inflammatory Diseases in India: A Multi-Centric Experience. *Front Immunol*. 2021 Mar 19;12:630691. doi: 0.3389/fimmu.2021.630691eCollection 2021. PMID: 33815380 **Free PMC article.**

275) Agrawal N, **Phadke SR**. Peters-Plus with Anal Atresia and a Novel Frameshift Mutation. Indian J Pediatr. 2021 Feb;88(2):184-185. doi: 10.1007/s12098-020-03416-7. Epub 2020 Jul 11. PMID: 32654102 No abstract available.

276) Masih S, Moirangthem A, **Phadke SR**. Homozygous Missense Variation in *PNPLA8* Causes Prenatal-Onset Severe Neurodegeneration. Mol Syndromol. 2021 Jun;12(3):174-178. doi: 10.1159/000513524. Epub 2021 Mar 19. PMID: 34177434 . \$

277) Agrawal N, Srivastava P, **Phadke SR**. Desbuquois dysplasia Kim variant: a rare case report syndrome. Clin Dysmorphol. 2021 Jan;30(1):62-65. doi: 10.1097/MCD.0000000000000356. PMID: 33136656 No abstract available.

278) Sait H, Gangadharan H, Gupta A, Aggarwal A, Jain M, **Phadke SR**. Monogenic Lupus with IgA Nephropathy Caused by Spondyloenchondrodysplasia with Immune Dysregulation. Indian J Pediatr. 2021 Aug;88(8):819-823. doi: 10.1007/s12098-020-03636-x. Epub 2021 Mar 13. PMID: 33712926

279) Rai A, Patil SJ, Srivastava P, Gaurishankar K, **Phadke SR**. Clinical and molecular characterization of four patients with Robinow syndrome from different families. Am J Med Genet A. 2021 Apr;185(4):1105-1112. doi: 10.1002/ajmg.a.62082. Epub 2021 Jan 26. PMID: 33496066

280) Nilay M, Saxena D, Mandal K, Moirangthem A, **Phadke SR**. Novel pathogenic variants in an Indian cohort with epidermolysis bullosa: Expanding the genotypic spectrum. Eur J Med Genet. 2021 Dec;64(12):104345. doi: 10.1016/j.ejmg.2021.104345. Epub 2021 Sep 29 PMID: 34597860

281) Deshpande D, Gupta SK, Sarma AS, Ranganath P, Jain S JMN, Sheth J, Mistri M, Gupta N, Kabra M, **Phadke SR**, Girisha KM, Dua Puri R, Aggarwal S, Datar C, Mandal K, Tilak P, Muranjan M, Bijarnia-Mahay S, Rama Devi A R, Tayade NB, Ranjan A, Dalal AB. Functional characterization of novel variants in *SMPD1* in Indian patients with acid sphingomyelinase deficiency. Hum Mutat. 2021 Oct;42(10):1336-1350. doi: 10.1002/humu.24263. Epub 2021 Aug 3. PMID: 34273913

282) Moirangthem A, **Phadke SR**. Novel FOXP1 pathogenic variants in two Indian subjects with syndromic intellectual disability. Am J Med Genet A. 2021 Apr;185(4):1324-1327. doi: 10.1002/ajmg.a.62083. Epub 2021 Jan 11. PMID: 33427368

283) Sait H, Srivastava P, Gupta N, Kabra M, Kapoor S, Ranganath P, Rungsung I, Mandal K, Saxena D, Dalal A, Roy A, Pabbati J, **Phadke SR**. Phenotypic and genotypic spectrum of CTSK variants in a cohort of twenty-five Indian patients with pycnodysostosis. Eur J Med Genet. 2021 Jul;64(7):104235. doi: 10.1016/j.ejmg.2021.104235. Epub 2021 May 1. PMID: 33945887

284) **Phadke SR**, Srivastava P, Sharma P, Rai A, Masih S. Homozygosity stretches around homozygous mutations in autosomal recessive disorders: patients from nonconsanguineous Indian families. J Genet. 2021;100:2. PMID: 33707353

285) Moirangthem A, Mandal K, Saxena D, Srivastava P, Gambhir PS, Agrawal N, Shambhavi A, Nampoothiri S, **Phadke SR**. Genetic heterogeneity of disorders with overgrowth and intellectual disability: Experience from a center in North India. Am J Med Genet A. 2021 Aug;185(8):2345-2355. doi: 10.1002/ajmg.a.62241. Epub 2021 May 4. PMID: 33942996

286) Gangadharan H, Singh K, **Phadke S**, Aggarwal A. Clinical Sequencing Solves a Diagnostic Dilemma by Identifying a Novel Pathogenic Variant in *USB1* Gene Causing Poikiloderma

with Neutropenia. *Indian J Pediatr.* 2021 Mar;88(3):270-271. doi: 10.1007/s12098-020-03502-w. Epub 2020 Sep 16. PMID: 32936385 No abstract available.

287) Nilay M, Srivastava P, Rai A, **Phadke SR.** Partial Trisomy of Chromosome 8q and Partial Monosomy of Chromosome 6p with Robinow Syndrome-Like Phenotype. *Indian J Pediatr.* 2021 Aug;88(8):813-818. doi: 10.1007/s12098-021-03763-z. Epub 2021 May 22. PMID: 34021867

288) Nilay M, Moirangthem A, Saxena D, Mandal K, **Phadke SR.** Carrier frequency of SMN1-related spinal muscular atrophy in north Indian population: The need for population based screening program. *Am J Med Genet A.* 2021 Jan;185(1):274-277. doi: 10.1002/ajmg.a.61918. Epub 2020 Oct 14. PMID: 33051992

289) Kausthubham N, Shukla A, Gupta N, Bhavani GS, Kulshrestha S, Das Bhowmik A, Moirangthem A, Bijarnia-Mahay S, Kabra M, Puri RD, Mandal K, Verma IC, Bielas SL, **Phadke SR**, Dalal A, Girisha KM. A data set of variants derived from 1455 clinical and research exomes is efficient in variant prioritization for early-onset monogenic disorders in Indians. *Hum Mutat.* 2021 Apr;42(4):e15-e61. doi: 10.1002/humu.24172. Epub 2021 Mar 1. PMID: 33502066

290) Masih S, Moirangthem A, **Phadke SR.** Twins with PEX7 related intellectual disability and cataract: Highlighting phenotypes of peroxisome biogenesis disorder 9B. *Am J Med Genet A.* 2021 May;185(5):1504-1508. doi: 10.1002/ajmg.a.62110. Epub 2021 Feb 14. PMID: 33586206

291) Jacob P, Bhavani GSL, Shah H, Galada C, Nampoothiri S, Kamath N, **Phadke SR**, Muranjan M, Datar CA, Shukla A, Girisha KM. Pseudoachondroplasia: Phenotype and genotype in 11 Indian patients. *Am J Med Genet A.* 2022 Mar;188(3):751-759. doi: 10.1002/ajmg.a.62566. Epub 2021 Nov 9. PMID: 34750995

292) Sait H, Srivastava P, Dabadghao P, **Phadke SR.** Kallmann Syndrome and X-linked Ichthyosis Caused by Translocation Between Chromosomes X and Y: A Case Report. *J Reprod Infertil.* 2021 Oct-Dec;22(4):302-306. doi: 10.18502/jri.v22i4.7657. PMID: 34987993 **Free PMC article.**

293) Agrawal N, Kumar R, Masih S, Srivastava P, Singh P, Jaiswal SK, Moirangthem A, Saxena D, **Phadke SR**, Mandal K. Molecular analysis of severe hemophilia B in Indian families: Identification of mutational hotspot and novel variants. *Int J Lab Hematol.* 2022 Feb;44(1):186-192. doi: 10.1111/ijlh.13715. Epub 2021 Sep 29. PMID: 34590426

294) Sait H, Sajjan SM, **Phadke SR.** Haemophilia management programme: Transformation during COVID-19. *Indian J Med Res.* 2022 May-Jun;155(5&6):472-477. doi: 10.4103/ijmr.ijmr\_1941\_21. PMID: 36124495

295) Sait H, Sharma L, Dabadghao P, **Phadke SR.** Congenital Hyperinsulinemia of Infancy: Role of Molecular Testing in Management and Genetic Counseling. *Indian J Pediatr.* 2022 Apr;89(4):395-398. doi: 10.1007/s12098-021-04014-x. Epub 2022 Feb 19. PMID: 35182381

296) Moirangthem A, Saxena D, Masih S, Shambhavi A, Nilay M, **Phadke SR.** Variable neurological phenotypes of homocystinuria caused by biallelic methylenetetrahydrofolate reductase variants. *Clin Dysmorphol.* 2022 Apr 1;31(2):59-65. doi: 10.1097/MCD.0000000000000407. PMID: 34845156

297) Agrawal N, Verma G, Saxena D, Kabra M, Gupta N, Mandal K, Moirangthem A, Sheth J, Puri RD, Bijarnia-Mahay S, Kapoor S, Danda S, H SV, Datar CA, Ranganath P, Shukla A, Dalal A, Srivastava P, Devi RR, **Phadke SR**. Genotype-phenotype spectrum of 130 unrelated Indian families with Mucopolysaccharidosis type II. *Eur J Med Genet*. 2022 Mar;65(3):104447. doi: 10.1016/j.ejmg.2022.104447. Epub 2022 Feb 8. PMID: 35144014

298) Gupta N, Kaur R, **Phadke S**, Sharma P, Nampoothiri S, Saxena D, Kabra M. Monosomy 1p36: Report of a cohort of 13 Asian Indian patients. *Am J Med Genet A*. 2022 Apr;188(4):1317-1322. doi: 10.1002/ajmg.a.62630. Epub 2022 Jan 6. PMID: 34989134

299) Masih S, Moirangthem A, Shambhavi A, Rai A, Mandal K, Saxena D, Nilay M, Agrawal N, Srivastava S, Sait H, **Phadke SR**. Deciphering the molecular landscape of microcephaly in 87 Indian families by exome sequencing. *Eur J Med Genet*. 2022 Jun;65(6):104520. doi: 10.1016/j.ejmg.2022.104520. Epub 2022 May 12. PMID: 35568357

300) Srivastava P, Chaudhry C, Kaur A, **Phadke SR**, Panigrahi I. How Experts Make a Call: Copy Number Variation Analysis in Unusual/Rare Case Scenarios. *Neurol India*. 2022 Jan-Feb;70(1):148-154. doi: 10.4103/0028-3886.338651. PMID: 35263867

301) Mishra R, Kulshreshtha S, Mandal K, Khurana A, Diego-Álvarez D, Pradas L, Saxena R, **Phadke S**, Moirangthem A, Masih S, Sud S, Verma IC, Dua Puri R. COASY related pontocerebellar hypoplasia type 12: A common Indian mutation with expansion of the phenotypic spectrum. *Am J Med Genet A*. 2022 Aug;188(8):2339-2350. doi: 10.1002/ajmg.a.62768. Epub 2022 May 2. PMID: 35499143

302) Sait H, Moirangthem A, Agrawal V, **Phadke SR**. Autosomal recessive spinocerebellar ataxia-20 due to a novel SNX14 variant in an Indian girl. *Am J Med Genet A*. 2022 Jun;188(6):1909-1914. doi: 10.1002/ajmg.a.62701. Epub 2022 Feb 23. PMID: 35195341

303) Chaudhary AK, Gholse A, Nagarajaram HA, Dalal AB, Gupta N, Dutta AK, Danda S, Gupta R, Sankar HV, Bhavani GS, Girisha KM, **Phadke SR**, Ranganath P, Bashyam MD. Ectodysplasin pathogenic variants affecting the furin-cleavage site and unusual clinical features define X-linked hypohidrotic ectodermal dysplasia in India. *Am J Med Genet A*. 2022 Mar;188(3):788-805. doi: 10.1002/ajmg.a.62579. Epub 2021 Dec 4. PMID: 34863015

304) Sait H, Shambhavi A, Pandey M, Ravichandran D, **Phadke SR**. T2 olivary nuclei hyperintensities: A characteristic neuroimaging finding in FIG4 related leukoencephalopathy. *Am J Med Genet A*. 2022 Dec 18. doi: 10.1002/ajmg.a.63084. Online ahead of print. PMID: 36529678

305) Solomon BD, Adam MP, Fong CT, Girisha KM, Hall JG, Hurst ACE, Krawitz PM, Moosa S, **Phadke SR**, Tekendo-Ngongang C, Wenger TL. Perspectives on the future of dysmorphology. *Am J Med Genet A*. 2023 Mar;191(3):659-671. doi: 10.1002/ajmg.a.63060. Epub 2022 Dec 9. PMID: 36484420 PMCID: PMC9928773.

306) Nagral A, Mallakmir S, Garg N, Tiwari K, Masih S, Nagral N, Unavane O, Jhaveri A, **Phadke S**, ArunKumar G, Aggarwal R. Genomic Variations in ATP7B Gene in Indian Patients with Wilson Disease. *Indian J Pediatr*. 2023 Mar;90(3):240-248. doi: 10.1007/s12098-022-04250-9. Epub 2022 Sep 16. PMID: 36112267.

307) Varshney K, Narayananachar SG, Girisha KM, Bhavani GS, Narayanan D, **Phadke S**, Nampoothiri S, Udupi GA, Raghupathy P, Nair M, Geetha TS, Bhat M. Clinical, radiological and molecular studies in 24 individuals with Dyggve-Melchior-Claussen dysplasia and Smith-

McCort dysplasia from India. *J Med Genet*. 2023 Apr 27:jmedgenet-2021-108098. doi: 10.1136/jmedgenet-2021-108098. Online ahead of print. PMID: 35477554

308) Pranav Chand R, Vinit W, Vaidya V, Iyer AS, Shelke M, Aggarwal S, Magar S, Danda S, Moirangthem A, **Phadke SR**, Goyal M, Ranganath P, Mistri M, Shah P, Shah N, Kotecha UH. Proband only exome sequencing in 403 Indian children with neurodevelopmental disorders: Diagnostic yield, utility and challenges in a resource-limited setting. *Eur J Med Genet*. 2023 May;66(5):104730. doi: 10.1016/j.ejmg.2023.104730. Epub 2023 Feb 15. PMID: 36801247.

309) Saxena D, Srivastava S, Maurya RK, Moirangthem A, Mandal K, **Phadke S**. Retrospective diagnosis by parental testing in the next generation sequencing era and utility of reanalysis of exome data. *Prenat Diagn*. 2023 May;43(5):579-595. doi: 10.1002/pd.6346. Epub 2023 Apr 3. PMID: 36964991.

310) Sait H, Srivastava S, Pandey M, Ravichandran D, Shukla A, Mandal K, Saxena D, Shambhavi A, Majethia P, Rao LP, Sharma S, **Phadke SR**, Moirangthem A. Neurodegeneration with brain iron accumulation: a case series highlighting phenotypic and genotypic diversity in 20 Indian families. *Neurogenetics*. 2023 Apr;24(2):113-127. doi: 10.1007/s10048-023-00712-0. Epub 2023 Feb 15. PMID: 36790591.

311) Tiwari AK, Srinivasan VM, **Phadke SR**, Saxena D. Variants in DOK7 results in fetal akinesia deformation sequence: A case report and review of literature. *Clin Genet*. 2023 Oct 17. doi: 10.1111/cge.14431. Epub ahead of print. PMID: 37849383.

312) Yadav S, Madhumita RC, Gupta N, Chauhan S, Kusmakar S, Balakrishnan P, Jana M, Puri RD, Phadke SR, Kabra M. Isolated Lateralized Overgrowth - Phenotypic Spectrum and Molecular Alterations. *Indian J Pediatr*. 2024 Oct 19. doi: 10.1007/s12098-024-05273-0. Epub ahead of print. PMID: 39425824.

313) Verma A, Verma MK, Priyanka VL, Naranje K, Singh A, Roy A, Paul A, Phadke S, Kumar B. Epidemiology and management of congenital anomalies in neonates in a hospital in Northern India. *J Trop Pediatr*. 2024 Oct 4;70(6):fmae038. doi:

314) Sait H, Pandey M, Phadke SR. COQ7 splice site variant causing a spastic paraparesis phenotype in siblings. *J Genet*. 2024;103:26. PMID: 39080983.

315) Banu S, Mk K, George JK, Siby E, Bhagat R, Ms S, Patil SJ, Phadke SR, Sowpati DT, Tallapaka KB. Enhanced resolution of optical genome mapping utilizing telomere-to-telomere reference in genetic disorders. *Eur J Hum Genet*. 2024 Dec 9. doi: 10.1038/s41431-024-01763-z. Epub ahead of print. PMID: 39653745.

316) Chauhan S, Kusmakar S, Balakrishnan P, Jana M, Puri RD, Phadke SR, Kabra M. Isolated Lateralized Overgrowth - Phenotypic Spectrum and Molecular Alterations. *Indian J Pediatr*. 2024 Oct 19. doi: 10.1007/s12098-024-05273-0. Epub ahead of print. PMID: 39425824.

317) Jacob P, Singh S, Bhavani GS, Gowrishankar K, Narayanan DL, Nampoothiri S, Patil SJ, Soni JP, Muranjan M, Kapoor S, Dhingra B, Bhat BV, Bajaj S, Banerjee A, Mamadapur M, Hariharan SV, Kamath N, Shenoy RD, Suri D, Shukla A, Dalal A, Phadke SR, Nishimura G, Mortier G, Shah H, Girisha KM. Genetic and allelic heterogeneity in 248 Indians with skeletal dysplasia. *Eur J Hum Genet*. 2024 Dec 20. doi: 10.1038/s41431-024-01776-8. Epub ahead of print. PMID: 39706863.

318) Manisha R, Phadke SR. Long-Term Outcomes of Disease Modifying Therapies in Gaucher Disease. *Indian J Pediatr.* 2025 Jun;92(6):585-591. doi: 10.1007/s12098-023-04986-y. Epub 2024 Feb 5. PMID: 38315376.

319) Shambhavi A, Moirangthem A, Mishra P, Phadke SR. Understanding and issues related to next-generation sequencing among educated laypersons in India. *J Genet Couns.* 2025 Apr;34(2):e2008. doi: 10.1002/jgc4.2008. PMID: 40110627.

320) Adarsha N, Shambhavi A, Sait H, Moirangthem A, Saxena D, Phadke SR. Spectrum of Inherited Childhood-Onset Dystonia: Case Series of 19 Families with Genotype and Phenotype Characterization Highlighting the Treatable Causes. *Clin Genet.* 2025 Apr 30. doi: 10.1111/cge.14762. Epub ahead of print. PMID: 40302693.

321) Sait H, Adarsha N, Moirangthem A, Saxena D, Sharma L, Dabadgao P, Gupta A, Phadke SR. Molecular and Clinical Landscape of Osteogenesis Imperfecta: Unraveling Autosomal Recessive Forms, Therapeutic Outcomes, and Bone Mineral Density in Carriers. *Clin Genet.* 2025 Jul 12. doi: 10.1111/cge.70003. Epub ahead of print. PMID: 40650436.

## 18. BOOK AND CHAPTERS IN BOOK

1. “**GENETICS FOR CLINICIANS**” Published in June 2007 by **PRISM BOOKS PVT LTD, 1865, 32nd Cross, 10th main, BSK II Stage, Bangalore, 560070**
2. “Genetic Counseling” in “Update in Pediatrics” edt I C Verma
3. “Malformation syndromes in India” in ‘Genetic disorders in Indian subcontinent’ edited by Dhavendra Kumar, Kluwer Academic Publishers, 2005 and Second edition in 2011
4. “Molecular techniques in prenatal diagnosis” in ‘Medicine Update2003’ vol VII pp42-45
5. “Handbook of clinical genetics” for ICMR course in Medical Genetics and Genetic Counseling. 2002
6. “Chromosomal disorders” in ‘ Pediatrics & Neonatology’ edit Agrawal KN, Modern Publishers, New Delhi, 2000
7. “Congenital Malformations” in ‘Human genetics for students’ by Moving Academy of Medicine & Biomedicine, Pune, edited by Deo MG, Ganguli S, Kher A, 2002
8. “Genetic counseling and prenatal diagnosis” in API Textbook of Medicine, ninth edition . Editor: Yash Pal Munjal, Japee brothers, 2012, pp209 -214
9. Section Editor for ‘**GENETICS**’ in IAP color atlas of Pediatrics. Editor: A Parthsarathy. Japee brothers, 2012. Pp260-280
10. ‘Genetic metabolic disorders’ in IAP textbook of pediatrics. Editor: Parthsarathy, fifth edition, 2013
11. Section editor for **GENETICS** in IAP textbook of pediatrics. Editor: Parthsarathy, JAPEE Publishers, fifth edition, 2013, 2018 edition
12. ‘**Clinical Genetics**’ in Textbook of Pediatrics, Editor: Piyush Gupta. CBS publishers 2013
13. ‘Inborn Errors of Metabolism Presenting in the Newborn Period: Representative Phenotypes & Diagnostic Approach’ in ‘Medical Emergencies in Newborn’ Editor: Meharban Singh, 2016
14. “Genetic counseling and prenatal diagnosis” in API Textbook of Medicine, ninth edition. Editor: Dr. Sandhya Kamath, Japee brothers, 2019, Eleventh Edition, pp209 - 214

15. Section Editor for Medical GENETICS, in API Textbook of Medicine, ninth edition. Editor: Dr. Sandhya Kamath, Japee brothers, 2019, Eleventh Edition,
16. 'Genetic Metabolic Disorders' Authors: Ranjana Mishra, Shubha R Phadke. In 'TEXTBOOK OF PEDIATRICS 3rd EDITION' Editor: Dr Piyush Gupta, 2023. Japee Brothers

## 18. 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 43<sup>rd</sup> Short course in Medical & Experimental Mammalian Genetics at Bar Harbor, USA 15<sup>th</sup> July to 26<sup>th</sup> 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 15<sup>th</sup> March 2011 to 20<sup>th</sup> March 2011
7. 'Clinical genetics: Indian Scenario" in Twelfth ICHG and annual conference of American Society of Human Genetics at Montreal, Canada, 11<sup>th</sup> to 15<sup>th</sup> 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. "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 Mucopolysaccharidosis II (Hunter Syndrome) by Sequencing of IDS gene' and got award
10. '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 16<sup>th</sup> Oct to 20<sup>th</sup> Oct 2018
11. '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
12. '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.
13. "Exome Sequencing in Understanding the Etiologies of Ataxia in Children, " in 14<sup>th</sup> International Congress of Human Genetics, held from 22<sup>nd</sup> Feb to 26<sup>th</sup> Feb 2023, at Cape Town South Africa
14. "Polydactyly – A 20 Year Journey from Clinical to Molecular Diagnosis" in 19th Manchester Dysmorphology Conference 2023, held from 16<sup>th</sup> to 18<sup>th</sup> Oct 2023, at Hilton Hotel, Manchester, UK.

15. The International Genomics Education and Training Summit, held at Conference Centre on the Wellcome Genome Campus, Hinxton, Cambridge on 27<sup>th</sup> and 28<sup>th</sup> Nov 2023

## 20. 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 Cytogenetics (06-to 09.12.99) (Treasurer)
6. ICMR Course on Genetic Counseling (18<sup>th</sup> to 30<sup>th</sup>, Sept. 2000) (Course Convenor)
7. Camp for Hemophilia children 9<sup>th</sup> to 30<sup>th</sup> Dec. 2000 (Organizer) at SGPGIMS.
8. Foundation Day Cultural program on 14<sup>th</sup> December 2000 at SGPGIMS.
9. 2<sup>nd</sup> ICMR course on Genetics Counseling (10<sup>th</sup> to 21<sup>st</sup>, April 2001) (Course coordinator)
10. 3<sup>rd</sup> ICMR course on Medical Genetics & Genetic Counseling(11<sup>th</sup> to 22<sup>nd</sup> Dec. 2001) (Course Convenor)
11. 4<sup>th</sup> ICMR course on Medical Genetics & Genetic Counseling (9<sup>th</sup> to 20<sup>th</sup>, April 2002) (Course Convenor)
12. Camp for Hemophilia Children at Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow 25<sup>th</sup> –28<sup>th</sup>, Dec. 2002.
13. 5<sup>th</sup> ICMR course on Medical Genetics and Genetic Counseling. 9<sup>th</sup> Feb. to 21stFeb. 2004.
14. Sixth ICMR course on Medical Genetics and Genetic Counseling . 11<sup>th</sup> April to 23<sup>rd</sup> April 2005
15. Seventh ICMR course on Medical Genetics and Genetic Counseling July 17<sup>th</sup>-July 22<sup>nd</sup>, 2006
16. ‘GENETICS FOR PAEDIATRICIANS’ A CME organized on behalf of Genetics Chapter of Indian Academy of Paediatrics, at Sir GangaRam Hospital, New Delhi, 15<sup>th</sup> and 16<sup>th</sup> Dec 2007.
17. Eighth ICMR course on Medical Genetics and Genetic Counseling. 20<sup>th</sup> July 2008 to 2<sup>nd</sup> August 2008
18. ‘Ninth ICMR Course on Medical Genetics and Genetic Counseling’ (2<sup>nd</sup> August 2010 to 14<sup>th</sup> August 2010)
19. **‘First Indo US Symposium on Skeletal Dysplasia’ at Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow on 12<sup>th</sup> and 13<sup>th</sup> Feb 2011 [In collaboration with Dr Rimoin]**
20. Workshop on DNA diagnostics 2011, in Medical Genetics, SGPGIMS, on 24<sup>th</sup> to 26<sup>th</sup> Nov 2011
21. ‘Tenth ICMR Course on Medical Genetics and Genetic Counseling’ from 9<sup>th</sup> to 20<sup>th</sup> August 2011
22. ‘Eleventh ICMR Course on Medical Genetics and Genetic Counseling’ from 29<sup>th</sup> July 12 to 11<sup>th</sup> August 2012
23. ‘Twelfth ICMR Course on Medical Genetics and Genetic Counseling’ from 29<sup>th</sup> July 2013 to 13<sup>th</sup> Aug 2013

24. 'Thirteenth ICMR Course on Medical Genetics and Genetic Counseling' from 28<sup>th</sup> July 2014 to 9<sup>th</sup> 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 25<sup>th</sup> July 2016 to 6<sup>th</sup> August 2016
27. Sixteenth ICMR Course on Medical Genetics and Genetic Counseling' from 24<sup>th</sup> July 2017 to 5<sup>th</sup> August 2017
28. Seventeenth ICMR Course on Medical Genetics and Genetic Counseling' from 20<sup>th</sup> August 2018 to 1<sup>st</sup> September 2018
29. Eighteenth ICMR Course on Medical Genetics and Genetic Counseling' from 26<sup>th</sup> August 2019 to 7<sup>th</sup> September 2019
30. Organized online World Thalassemia Day on 8<sup>th</sup> May 21
31. Organized on line World Hemophilia Day – 17<sup>th</sup> April, 2021
32. 'Nineteenth ICMR Course on Medical Genetics and Genetic Counseling' from 1<sup>st</sup> August 2022 to 13th August 2022
33. Organized online World Thalassemia Day on 8<sup>th</sup> May 22, in SGPGIMS, Shruti Auditorium
34. Organized The First Medical Camp for Down syndrome patients and families at the department of Medical Genetics, SGPGIMS, Lucknow on the 15th & 16th of October 2022
35. Organized 7th Annual conference of Society of Indian Academy of Medical Genetics, "SIAMGCON 2022" on 9th and 10<sup>th</sup> December 2022 in Lecture theatre complex of SGPGIMS, Lucknow
36. Organized Workshop on CliGen on 8<sup>th</sup> December 2022 at Telemedicine Auditorium, SGPGIMS, Lucknow
37. Organized World Down Syndrome Day on 21<sup>st</sup> March 2023 for the children with Down syndrome and their families
38. On the occasion of World Hemophilia Day on 17th April 2023 with the theme, " To Assess For All" an awareness and recreational program was organised for Hemophilia patients by the entire team of Medical Genetics department under the leadership of Dr. S R Phadke (HOD- Medical Genetics). The program was organized in the Telemedicine Conference Room, SGPGIMS campus.
39. 5<sup>th</sup> July, 2023 – Dr S S Agarwal Remembrance meet. Public lecture by Dr Madhulika Kabra
40. Twentieth ICMR Course on Medical Genetics and Genetic Counseling' from 31<sup>st</sup> July to 12<sup>th</sup> Aug 2024
41. 29<sup>th</sup> Feb 24, Rare disease day celebration- educational program for patients with lysosomal storage disorders and spinal muscular atrophy
42. 21<sup>st</sup> March 24 – Celebration with patients and families of trisomy 21

43. 8<sup>th</sup> May 24, Celebration of World Thalassemia Day and awareness program- Dr S S Agarwal lecture hall in Lecture theatre complex, SGPGIMS, Lucknow

44. 5<sup>th</sup> July 24- Dr S S Agarwal Remembrance Meet – A public lecture on ‘Navigating the translational route of gene editing therapies in India’ by Dr Debojyoti Chakraborty, IGIB, New Delhi and musical program, ‘Finding Gandhar in science and medicine’

45. Twenty first ICMR Course on Medical Genetics and Genetic Counseling’ from 29<sup>th</sup> July to 10<sup>th</sup> Aug 2024

46. Workshop on Rare Diseases –“Care for Rare: Together we win” . Organized in Raman auditorium, Lecture theatre complex, SGPGIMS, in collaboration with rare disease cell, Ministry of Health & Family Welfare, Government of India on 22<sup>nd</sup> Jan 2025

47. Down syndrome day celebration at Lecture theatre complex, SGPGIMS, Lucknow, 21<sup>st</sup> March 2025

## **21. 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]
5. Joubert syndrome 30 [617622]
6. Microcephaly 21 [# 617983]

## **22. 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. European Journal Of Human Genetics (Section Editor since 2022)
9. European J of Medical Genetics
10. Prenatal diagnosis [official journal of International Society of Prenatal Diagnosis]
11. Indian Journal of Human Genetics
12. Gene
13. BMC journal
14. Molecular Genetics and Genomic Medicine

## **22. 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]
5. Member of the National Assisted Reproductive Technology and Surrogacy Board, 2023
6. Member of Rare Disease Technical Committee under National Policy for Rare Diseases
7. Chairperson of ICMR committee for Skeletal Dysplasia
8. Member of IEC, King George Medical University, Lucknow
9. Member of IEC, Centre for Bio Medical Research, SGPGIMS Campus, Lucknow
10. Chairman of Rare Disease Committee of SGPGIMS, FOR National Policy for Rare Diseases, 2021 till 31<sup>st</sup> March 25

### **23. INVITED LECTURES**

1. “Prenatal sex determination” In workshop on “Female Feticide” 6<sup>th</sup> 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. “Genetic Counseling for deafness”, CME at Annual conference of ISHA, Post Graduate Institute of Medical Sciences, Chandigarh, 12Feb. 1999.
5. “Hemophilia-Struggle with a lifelong problem” Keynote address at engineering college. Sultanpur on the occasion of concluding function of the Health week. April 1999.
6. “Genetics in Pediatrics Practice”: Lucknow Branch of Indian Association of Pediatrics. October’99
7. “Hemophilia common problems and management”: Annual function of Hemophilia Society Lucknow 1999
8. “Clinical Genetics-Indian perspective: at 25<sup>th</sup> Annual conference of Indian Society of Human Genetics 9<sup>th</sup>-10<sup>th</sup>, 2000 Nagpur
9. “Hemophilia-A lifelong struggle” A lecture for school children in St. Paul’s School on the occasion of Hemophilia Day Celebration 6<sup>th</sup> 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, 7<sup>th</sup>-10<sup>th</sup> Feb. 2001.
12. “DNA Diagnosis of Genetic Disorders” Guest Lectures at XXXVIII National Conference of Indian Academy of Pediatrics at Patna, 7<sup>th</sup>-10<sup>th</sup> Feb. 2001.

13. "Dysmorphology Diagnosis in fetus & a neonate" Guest Lectures at XXXVIII National Conference of Indian Academy of Pediatrics at Patna, 7<sup>th</sup>-10<sup>th</sup> 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 18<sup>th</sup> 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 28<sup>th</sup> Oct 2002
18. "Genetics in clinical practice" at CME, Indian Association of Pediatrics, Gorakhpur branch on 25<sup>th</sup> Dec 2002
19. "Prenatal diagnosis of hemophilia" at Hemophilia Workshop, by Hemophilia Society, Nagpur, on 19<sup>th</sup> 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 23<sup>rd</sup> Feb 2003
22. "Cytogenetic techniques & applications" GENTICON by Indian Association Medicine, Rajkot on 23<sup>rd</sup> 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 18<sup>th</sup> 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 23<sup>rd</sup> -25<sup>th</sup> 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 23<sup>rd</sup> -25<sup>th</sup> Sept 2003
27. "Prenatal diagnosis of CNS malformations" in Neuropedicon 2003 14<sup>th</sup> Annual conference of Indian Society of Pediatric Neurosurgery at Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, on 6<sup>th</sup> Nov 2003
28. "Genetic counseling – Illustrative cases" in Pediatric Multispeciality Update at Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, on 15<sup>th</sup> nov 2003
29. "Molecular diagnosis in clinical genetics: scope and limitations" lecture in Pediatric subspeciality symposium in PEDICON 2004, on 10<sup>th</sup> 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 14<sup>th</sup> 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 7<sup>th</sup> 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, 30<sup>th</sup> 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, 1<sup>st</sup> oct 2004
34. "Ethical issues in fetal medicine and Perinatology" Lecture in 01 course of Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow , 8<sup>th</sup> dec 2004
35. "Clinical cytogenetics and role of FISH" in Workshop on techniques in genetic toxicology at Indian Institute of Toxicological Research , Lucknow on 12<sup>th</sup> dec 2004.
36. "Congenital Malformations :Diagnosis & counseling" Annual conference of Indian Association o Pediatrics – PEDICON 2005, at Kolkota,8<sup>th</sup>jan 2005.
37. "Diagnosis and prevention of congenital malformations" R N Ganguli public forum in 21<sup>st</sup> conference of Indian society of perinatology and reproductive biology, in Lucknow , 6<sup>th</sup>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, 21<sup>st</sup>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<sup>TH</sup> Sept 2005.
40. "Molecular techniques in cancer: Research and management" in Department of Radiotherapy, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow. 23<sup>rd</sup> sept 2005.
41. "Congenital malformations: A gateway to developmental genetics" In 74<sup>th</sup> Annual meeting of Society of biological chemists, India. Central Drug Research Institute, Lucknow. 8<sup>th</sup>nov 2005.
42. "Congenital malformations: USG and autopsy correlation" in 8<sup>th</sup> Conference of Indian Society of Prenatal Diagnosis and Fetal Therapy at New Delhi. 19<sup>th</sup> 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 1<sup>st</sup> August 2006
45. "USG in prenatal diagnosis of malformations" in Conference on antenatal diagnosis, at Vivekanand polyclinic, Lucknow on 26<sup>th</sup> August 2006
46. "Role of perinatal autopsy in genetic counseling" in Workshop on Prenatal Medicine in Deenanath Mangeshkar Hospital and Research Centre, Pune on 6<sup>th</sup> October 2006.
47. "DNA diagnosis of monogenic disorders" in Workshop on Prenatal Medicine in Deenanath Mangeshkar Hospital and Research Centre, Pune on 6<sup>th</sup> October 2006.

48. "Common presentations or rare genetic disorders" in PEDICON 2007, Mumbai on 14<sup>th</sup>jan 2007
49. "Skeletal Dysplasia" in PEDICON 2007, Mumbai on 12<sup>th</sup>jan 2007
50. "Five lectures in workshop" of CLINICAL GENETICS at Rainbow hospital, Hyderabad.
51. "Malformation Syndromes: Indian scenario' on 5<sup>th</sup> June 2007, in Pediatric Genetics Department, Shands Medical Centre, Gainesville, USA
52. "DNA diagnosis in clinical practice" in ISPAT 2007 Chennai , 23<sup>rd</sup> to 25<sup>th</sup> Nov 2007
53. "Genetics of artificial reproductive techniques" in ISPAT 2007 Chennai , 23<sup>rd</sup> to 25<sup>th</sup> Nov 2007
54. "Chromosomal abnormalities: How to choose the best test" in ISPAT 2007 Chennai , 23<sup>rd</sup> to 25<sup>th</sup> Nov 2007
55. "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, 15<sup>th</sup> and 16<sup>th</sup> Dec 2007
56. "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, 15<sup>th</sup> and 16<sup>th</sup> Dec 2007
57. "Dysmorphology diagnosis" in Indo European CME on Clinical Genetics and Dysmorphology, at CMC, Vellore on 2<sup>nd</sup> October 2008
58. "Medical Genetics: Indian scenario" in PERICON, at Ernakulum, Kerala on 5<sup>th</sup> October 2008
59. "Fetal autopsy: Indications & utility" in North India conference on fetal medicine and prenatal diagnosis , on 16<sup>th</sup> Nov 2008, at AIIMS, New Delhi
60. "Malformation Syndromes: Our experience" Spring Conference of Clinical Genetics Society, London, UK, 12<sup>th</sup> March 2009
61. 'Genetics of Short Stature' in Fourth International Conference on Birth Defects and Disabilities in Developing World, New Delhi, 4 – 7<sup>th</sup> October 2009
62. 'Genetic Screening' in Fourth International Conference on Birth Defects and Disabilities in Developing World, New Delhi, 4 – 7<sup>th</sup> October 2009
63. 'Ethical issues in Genetics and stored tissue research' in 'ICMR-FERCAP Human subject protection course' held on 7<sup>th</sup> 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 5<sup>th</sup> Nov 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 23<sup>rd</sup> Nov 2010
66. 'New tools for evaluation of mental retardation' at Annual conference of ISHG at Manipal on 16<sup>th</sup> Feb 2011
67. 'Mental Retardation: An unsolved Mystery' in 'The current practice of genetics and genomic medicine' held at Apollo Hospital , New Delhi on 26<sup>th</sup> and 27<sup>th</sup> March 2011

68. 'Fetal autopsy: a must' in 'The current practice of genetics and genomic medicine' held at Apollo Hospital , New Delhi on 26<sup>th</sup> and 27<sup>th</sup> March 2011
69. " Molecular Medicine in resource poor countries' in 'Synergy and diversity of Molecular Medicine' at PSG Centre for Molecular Medicine, Coimboor on 27<sup>th</sup> and 28<sup>th</sup> June 2011.
70. "Genetic disorders with joint problems" in National Conference of Pediatric Rheumatology on 3<sup>rd</sup> 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 4<sup>th</sup> Sept 2011
72. "Skeletal Dysplasia: Clinical Approach" in workshop "Dysmorphology: Unmystified" organized by Genetic Chapter of IAP in AIIMS, New Delhi on 4<sup>th</sup> Sept 2011
73. "DNA to disease: Genetic counseling for retinal disorders" in Retinaware 2011, on 24<sup>th</sup> Sept 2011, In Vigyan Bhawan, New Delhi
74. "Malformation syndromes in fetus" in First international Congress of Society of Fetal Medicine, at New Delhi, 1<sup>st</sup> and 2<sup>nd</sup> Oct 2011
75. 'Molecular diagnostics in Clinical Practice" in Workshop on DNA diagnostics 2011, in Medical Genetics, SGPGIMS, on 24<sup>th</sup> to 26<sup>th</sup> Nov 2011
76. "Medical Genetics Services: Need for National & Regional Networks for Genetic Services" in Expert Group Meeting on Birth Defects in WHO SEAR ' from 13<sup>th</sup> to 15<sup>th</sup> Dec 2011 at AIIMS, New Delhi
77. "Fetal Malformations:Illustrative cases" in Workshop on fetal medicine and genetics at New Delhi, by NARCI on 14<sup>th</sup> Sept 12
78. 'Clinical approach in mental retardation' in Indo US Symposium on Disorders of Developing Brain' at Manipal, 27<sup>th</sup> -28th Oct 12
79. 'Prenatal diagnosis of Malformation syndromes' in Lucknow Ultrasound Course 2012 in Lucknow, 3<sup>rd</sup> Nov 12
80. 'Genetics of Endocrine Disorders' I 42<sup>nd</sup> Annual Conference of Endocrine Society of India, in Kolkota, 13<sup>th</sup> Dec 2012
81. "Pearls of Dysmorphology" in GENECON 2012 at Raipur, on 1<sup>st</sup> and 2<sup>nd</sup> Dec 12
82. 'Molecular cytogenetics" in GENECON 2012 at Raipur, on 1<sup>st</sup> and 2<sup>nd</sup> 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" 31<sup>st</sup> January 2013, At Dr.RMLIMS, Lucknow.
85. 'Newborn Screening in India: Why and How' in the North Zone conference of Indian Society of Perinatology and Reproductive Biology' in Lucknow on 6<sup>th</sup> Feb 2013
86. '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
87. 'Genetics of Epilepsy' in International conference on Cerebral Palsy and Developmental Medicine, 6<sup>th</sup> to 10<sup>th</sup> March 2013 at Lucknow.

88. 'Fabry Disease' in 'International conference on inborn errors of metabolism & 2<sup>nd</sup> national conference of ISIEN' at New Delhi on 5<sup>th</sup> -7<sup>th</sup> April 2013
89. 'Fabry disease: Diagnosis & ERT' in
90. 'aCGH, NIPD, FISH, QF PCR Demystifying the New Genetic Alphabets' in 2<sup>nd</sup> International Congress of Society of Fetal Medicine, on 31<sup>st</sup> August 2013 at Hyderabad
91. 'Genetic approach to short stature' in Growth hormone symposium organized by IAP, Mumbai branch on 1<sup>st</sup> 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 – 29<sup>th</sup>, 2013
93. 'Dysmorphology diagnosis: Face speaks' in MAHAPEDICON 2013, annual conference of Maharashtra API, on 19<sup>th</sup> Oct 13 at Nagpur
94. 'Sonological evaluation of fetal intracranial anatomy' in a workshop on "Maternal Fetal Medicine" from 25<sup>th</sup> – 27<sup>th</sup> November 2013, In the department of Obstetrics and Gynecology in King George's Medical College, Lucknow.
95. ': 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 28<sup>th</sup> , 29<sup>th</sup> and 30<sup>th</sup> Nov 2013.
96. "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.
97. 'Genetics: A Science of Probabilities' in State level symposium 'Exploring statistics applications in Diverse Fields' organized by Department of Statistics, Hislop college, Nagpur on 7<sup>th</sup> Dec 2013.
98. "Statistics in Genetics" in "Exploring Statistics Applications in Diverse Fields" at Nagpur, 7<sup>th</sup> Dec 13
99. 'Next Generation sequencing in Clinical Practice' in 'Genetics for Clinicians' at Kasturba Medical College, Manipal on 5<sup>th</sup> 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 29<sup>th</sup> Jan 2015
102. 'When to order a new generation test' in "Genetics for the Practicing Pediatrician " 21st January 2015, Sir Ganga Ram hospital, New Delhi
103. 'Newborn Screening' in International Symposium on 'Genetics and Genomics in Modern Clinical Medicine' at King George's Medical University, Lucknow on 9<sup>th</sup> Feb 2015
104. 'Nonsyndromic Mental Retardation' in PediGen2015, at Deenanath Mangeshkar Hospital, Pune ON 15<sup>TH</sup> Feb 15
105. 'Genomic techniques in diagnostics' in National Conference on " Biotechnology and Human Welfare: New Vista' in VBS Purvanchal University, Jaunpur on 22<sup>nd</sup> March 2015

106. "Genetics: Bench to bedside' in CME on Molecular Genetics for Practicing Clinicians on 19 April 2015, Mumbai
107. "Treatment of Genetic 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 14<sup>th</sup> 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 7<sup>th</sup> Sept 2015
110. 'USG and Genetics' in Sixth Ultrasonography Course in Lucknow Under Aegis of Ultrasound Education & Research Foundation, 1<sup>st</sup> Nov 2015
111. 'Clinical Genomics' in Second Annual conference of Society for Indian Academy of Medical Genetics, at Jodhpur on 12<sup>th</sup> and 13<sup>th</sup> Dec 2015
112. 'Genomic Techniques and Consanguinity in Rare Disorders' in MEDLAB [Arab Health Conference] on 26<sup>th</sup> 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 22<sup>nd</sup> 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, 7<sup>th</sup> , 8<sup>th</sup> May 16
116. 'Syndromes and Congenital Heart Disease' in International Congress on Advances in ObGyn Ultrasound 2016 in Hyderabad, 7<sup>th</sup> , 8<sup>th</sup> May 16
117. 'In ILLUMNATI at Armed Forces Medical College, Pune on 5<sup>th</sup> Aug 2016
118. ' In ILLUMNATI at Armed Forces Medical College, Pune on 5<sup>th</sup> Aug 2016
119. ' In ILLUMNATI at Armed Forces Medical College, Pune on 5<sup>th</sup> Aug 2016
120. 'Technological Milestones in (*Applications in*) Fetal Genetics' in FetalMed2016 on 10<sup>th</sup> September 2016, in Kolkata
121. 'Genetic Factors in Fetal Growth Restriction' in in FetalMed2016 on 10<sup>th</sup>September 2016, in Kolkata
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 15<sup>th</sup> , 2016<sup>th</sup> 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 1<sup>st</sup> and 2<sup>nd</sup> Dec 16
125. 'Molecular Diagnostics in Clinical Dysmorphology: Indian Experience' in third Annual Conference of Society for Indian Academy of Medical Genetics, at All India Institute of Medical Sciences, New DELHI on 8<sup>th</sup> and 9<sup>th</sup> Dec 16
126. 'Approach to neurometabolic disorders' in IV National Conference of Indian Society of Inborn Errors of Metabolism, in Chennai, 11<sup>th</sup> to 12<sup>th</sup> Feb 17

127. 'Genomic Diagnosis : Journey with Patients' in Manipal Genetics Update IV on Genetic Counseling at Kasturba Medical College, Manipal [3<sup>rd</sup> and 4<sup>th</sup> March 2017]
128. 'Genetics for Clinicians- Pedigree to Genome' in Faculty Development Program, School of Medicine, KIIT University, Bhubaneshwar, on 29<sup>th</sup> June 17
129. 'Genetics of Multifactorial Disorders: Learning from Age Related Multifactorial Disorders' in Faculty Development Program, School of Medicine, KIIT University, Bhubaneshwar, on 29<sup>th</sup> June 17
130. What aneuploidy screening can be offered to a woman who presents after 14 weeks' in FetalMed 2017[13<sup>th</sup> Asia Pacific Congress of Maternal Fetal Medicine] on 2<sup>nd</sup>& 3<sup>rd</sup> sept2017 at Leela Ambiance Hotel, Gurugram, New Delhi
131. 'Aneuploidy screening: Indian scenario' in FetalMed 2017[13<sup>th</sup> Asia Pacific Congress of Maternal Fetal Medicine] on 2<sup>nd</sup> 3<sup>rd</sup> sept2017 at Leela Ambiance Hotel, Gurugram, New Delhi
132. 'Learning Dysmorphology: Noonan Syndrome' in An international conference on RASopathies, Crowne Plaza, Kochi from 27<sup>th</sup> Nov to 29<sup>th</sup> Nov, 2017
133. 'Prenatal Growth Retardation: Diagnostic Challenges' in Annual conference of Society for Indian Academy of Medical Genetics, on 8<sup>th</sup> and 9<sup>th</sup> 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, 5<sup>th</sup>, 6<sup>th</sup> and 7<sup>th</sup> Jan 2018
136. 'Medical Genetics: Indian Scenario' in PEDICON 2018, AT Nagpur, 5<sup>th</sup>, 6<sup>th</sup> and 7<sup>th</sup> Jan 2018
137. 'Prenatal Diagnosis' in PEDICON 2018, AT Nagpur, 5<sup>th</sup>, 6<sup>th</sup> and 7<sup>th</sup> Jan 2018
138. 'Down Syndrome & More' in Mayo Medical College, Barabanki on 21<sup>st</sup> 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 25<sup>th</sup> 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 20<sup>th</sup> 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, 19<sup>th</sup> to 21<sup>st</sup> Nov 2018.
145. 'Genetics of facial clefts' in APSI Accredation Course on Cleft Lip Palate &Cranio Facial Anomalies – 2018 , 19<sup>th</sup> Nov 2018

146. "Future of Genetics in Neuro-disability" in Child Neurodisability Conference- 2018 on 3<sup>rd</sup> Dec 2018
147. 'Current Consensus And Practical Tips To Apply Newborn Screening In Clinical Practice' in Annual conference of ISIEM, at Pune 19<sup>th</sup> Jan 2019
148. 'Panels, panels everywhere; which test to choose - Genetic testing' in Genetics Workshop, PEDICON 2019, at KEM Hospital, Mumbai on 6<sup>th</sup> Feb 2019
149. 'Charting and interpreting pedigrees' in Genetics Workshop, PEDICON 2019, at KEM Hospital, Mumbai on 6<sup>th</sup> Feb 2019
150. 'Genetic diagnosis: Selecting the right tests ' in PEDICON 2019, at Mumbai, 9<sup>th</sup> March 2019
151. 'Genetics in Medicine' in "Latest Trends in Biotechnology for the benefit of the People of Uttar Pradesh" on 12th March, 2019.
152. '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)
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 17<sup>th</sup> March 2019
154. 'Genetics: You Need to Know for PCPNDT Implementation: Our Mission' on 23<sup>rd</sup> March 2019 for medical officers at CMO office Lucknow
155. 'Roadmap for Management of Rare Disorders in India', 13<sup>th</sup> April 2018, 7th International conference on Rare and Undiagnosed diseases. Addressing patient needs for Rare Disorders in India, 13-15<sup>th</sup> April 2019, New Delhi, India, Hotel Leela Ambience
156. 'Genetics in Autism' on World Autism Day – Keynote address o 2<sup>nd</sup> April 2019 at program organized by Association of Child Brain Research in M B Club, Lucknow
157. ' Genetics - the Present and The Future of Medicine" on 7th April 2019 , at Genetics for Pediatricians in Nanavati Hospital, Mumbai
158. "Treatable Lysosomal Storage Disorders-Diagnosis and Management " on 27<sup>th</sup> 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 15<sup>th</sup> Annual Conference of Academy of Pediatrics (**NAPCON 2019**) & National Mid-Term CME of Infectious Diseases Chapter of IAP held on 29<sup>th</sup> 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 22<sup>nd</sup> Nov 2019
162. 'Autosomal recessive inheritance' in "Decoding Genetic Investigations: Interpretation of NGS and other Genetic Tests in the Clinical Setting"on 24<sup>th</sup> Nov 2019 in Centre for DNA Diagnostics and Fingerprinting, Hyderabad

163. ‘Cytgenetic microarray’ on 24<sup>th</sup> 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 10<sup>th</sup> to 12<sup>th</sup> Jan 2020, at Sri Ram Cancer Center, Mahatma Gandhi Medical College & Hospital, Sitapura, Jaipur.
165. ‘Medical Genetics & Treatable Lysosomal Storage Disorders – Lessons learnt and Centre's Experience “interactive session with Sanofi Genzyme Rare Diseases Team of India on Friday, May 22<sup>nd</sup> , 2020 between 3:00-4:00pm
166. ‘Genetic testing When and what in pediatric office practice ‘ in IAP Central Zone E-Pedicon 2020 , 24.08.2020
167. ‘Genetic testing in Pediatric Practice: When and How?’ Moderator of Panel discussion IAP Annual Conference, 6<sup>th</sup> Feb 2021
168. ‘Cytogenetics and Molecular Cytogenetics’ in *IAP Fundamental Course: Medical Genetics* on 28<sup>th</sup> May 2021 [virtual platform]
169. ‘The will and the way to reach the unreached: our experience with newborn screening’ Virtual platform on 12<sup>th</sup> May 2021.
170. ‘Unexpected results on Non invasive Prenatal Testing’ Panel discussion organized by Society of Fetal Medicine, on virtual platform on 22<sup>nd</sup> Sept 2021
171. ‘**More for the Rare: An Era for Monogenic Disorders**’ **Dr P K Mishra** orations on 68th Foundation day celebrations of the Department of Pediatrics in KGMU, Lucknow on 12 Nov 2021
172. ‘Gene to Genome: My Journey’ On SGPGIMS Alumni meet on 13<sup>th</sup> Dec 2021
173. ‘Uncertainties in Prenatal Diagnosis: Dealing with the Challenges’ in 14<sup>th</sup> Biennial conference of ISPAT, in Clarks Awadh, Lucknow on **15th May 2022**
174. ‘Stillbirths Etiology and Genetic Counseling’ Panel discussion on virtual platform on 4<sup>th</sup> March 2022, organized by Stillbirth Society of India
175. ‘Panel Discussion on Rare Diseases’ Organized by IAP, UP chapter [virtual platform] on 27<sup>th</sup> Aug 2022
176. ‘Overview of Medical Genetics’ on 20<sup>th</sup> June 2022 in Genetic Research Centre, ICMR - NIRRH, Mumbai on inaugural function of the training course on Pediatric Medical Genetics.
177. Panel discussion ‘Panel Discussion: How can we achieve optimal drug management for al PwH A with or without Inhibitors’ in National Hemostasis Update on 20th August 2022, at Bangalore.
178. ‘ Monogenic disorders” PEDICON 2022 , Noida
179. “Gaucher Disease *Learning from cases & Stories of patients*” online 28<sup>th</sup> Oct 2022 Organized by the Team of Sanofi -Genzyme
180. “Uncertainties in Prenatal Diagnosis: Dealing with the Challenges: Annual Conference of ISPAT, AT Clark’s Awadh, Lucknow
181. ‘**Keynote lecture-Monogenic Disorders in India; The Influence of Endogamy & Consanguinity on Their Mutational Spectrum- In National Symposium on Genetic**

**disorders- From Bench to bedside which will be held on 15<sup>th</sup> & 16<sup>th</sup> Oct 2022 at Leela Ambience, Gurugram, Haryana.**

182. Medicine in Post Human Genome Project: *An Era for Monogenic Disorders* Keynote speaker in “Recent Trends in Health and Diseases” on Jan 19, 2022. Organized by Institute of Advanced Molecular Genetics & Infectious Diseases (IAMGID), ONGC Centre of Advanced Studies, University of Lucknow, Virtual platform

183. ‘Fetal Malformations’ in West Zonal FOGSI conference with YUVA 2022-23 at Ranthambore on 18<sup>th</sup> Nov 2022

184. ‘Issues in Prenatal Diagnosis’ at West Zonal FOGSI conference with YUVA 2022-23 at Ranthambore on 19<sup>th</sup> Nov 2022

185. Prenatally Detected Malformation: What Next? *ATGC of Genetics & Genetic Counseling SIAMG SYMPOSIUM ON BIRTH DEFECTS on 11<sup>th</sup> Dec 23 at Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow*

186. ‘Medicine in Genomic Era: Changing Paradigm: in Indian Science Congress, at Nagpur University on 5<sup>th</sup> January 2023

187. ‘Elegant Genetic Counseling for Test Results – Positive and Negative’ in NeuroPedicon at Hotel Shereton Grand, Pune on 2<sup>nd</sup> Feb 2023

188. “Diagnosis of Heatic Disorders in Genomic Era” in Brain-storming and updates in liver disorders, at Hotel Trident, Mumbai on 4<sup>th</sup> Feb 2023

189. TEDx, Ambazari 30<sup>th</sup> April, 2023, in Nagpur - “Whorl of DNA.....CHAKRAVYUH”

190. ‘Genetic counselling in Haemophilia and PCPNDT rules’, on the 17<sup>th</sup> of April 2023, at 10:00 am -10:45 am [virtual] in ‘Laboratory Workshop on Molecular Genetic Diagnosis of Haemophilia’, organized by the ICMR- National Institute of Immunohaematology, Mumbai

191. “Extended Half Life and Non-factor therapies in management of Hemophilia” Lucknow

192. “DNA Diagnostics by Sequencing: Diagnosis of Hepatic Disorders in Genomic Era” on 16<sup>th</sup> August 2023 in Department of Pediatrics, SGPGIMS, Lucknow

193. “Spectrum of genetic disorders: Steps in diagnosis” in “NEUROPEDICON 2023” held on 1st to 3rd September 2023 at Hotel Clarks Shiraz, Agra

194. “Genetics in Clinical Practice” Webinar organized by National Board of Examinations, on 25<sup>th</sup> January, 2024

195. “Implementation and utilization of genetics in Acute Care Setting” on 9<sup>th</sup> (Friday) of February, 2024 in Precision Medicine and Intensive Care Conference 2024 at Convention Centre, Lucknow

196. “The Diagnostic armamentarium in the Genomic era: How to choose the correct test? ’ in National Conference on Rare Diseases (RarDisESICON) ORGANIZED ON 2<sup>ND</sup> March 2024 at ESIC Medical College & Hospital at Faridabad, Haryana

197. “Policy & Advocacy for Birth Defects: Changing Needs in Prevention & Care” in SAMVAAD organized by Indian Society of Prenatal Diagnosis & Therapy on 3<sup>rd</sup> March 24 at Lady Hardinge Medical College, New Delhi

198. “Genetics in Clinical Practice” at 14<sup>th</sup> Annual Conference of GAPIO 2024 at Centrum, Lucknow on 17<sup>th</sup> March 2024

199. "Genetic testing of asymptomatic children" 'Dr IC Verma oration' in 3rd National Symposium on Genetic Diseases "RDIFCON2024" on 13th July 2024 at Ayurvigyan Auditorium, Army Hospital Research and Referral, New Delhi.
200. "Dysmorphology diagnosis in molecular era" in workshop "Dysmorphology in Genetic Evaluation" organized by SMBT Institute of Medical Sciences and Research Centre (IMS and RC), Nashik on 16<sup>th</sup> August 2024.
201. "Selecting a Right Screening Test for Screening for Down Syndrome & Interpreting" in Certificate Course in Fetal Radiology & Genetics organized by Indian Radiological Imaging Association, West Bengal Branch in Hotel Stadel on 14<sup>th</sup> Sept 2024
202. "Genomic Tests in Evaluation of a Fetus Right test & Interpretation" in Certificate Course in Fetal Radiology & Genetics organized by Indian Radiological Imaging Association, West Bengal Branch in Hotel Stadel on 14<sup>th</sup> Sept 2024
203. "Hemoglobinopathy, Spinal Muscular Atrophy & More" in Certificate Course in Fetal Radiology & Genetics organized by Indian Radiological Imaging Association, West Bengal Branch in Hotel Stadel on 14<sup>th</sup> Sept 2024
204. "NGS and other genetic tools in pediatric endocrinology- Use and interpretation" on 21<sup>st</sup> September 2024 in Pediatric Endo Clinics-2024 held at Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow
205. "Becoming a mother in genomic era: New dilemmas" in the International Conference and SRBCE annual meeting entitled "International Conference on Reproductive Sciences and Molecular Medicine: Innovations in Therapeutics and Technologies" (ICRSMM-2024) and 41st Annual Meet of Society for Reproductive Biology and Comparative Endocrinology (SRBCE), on Nov. 16, 2024 at University of Delhi (North Campus), Delhi
206. "Dysmorphology Diagnosis in AI & NGS Era" in annual conference of Society for Indian Academy of Medical Genetics, on 5<sup>th</sup> Dec 2024, at Ahmadabad
207. "Chromosomal Disorders: An Overview" online talk in the webinar series on Clinical Genetics for Pediatricians organized by Indian Academy of Pediatrics - Telangana State Chapter is organizing. 1<sup>st</sup> Jan 2025
208. "An Update on Lysosomal Storage Disorders" in Workshop on rare diseases in SGPGIMS, Lucknow on 22<sup>nd</sup> Jan 2025
209. "Newborn Screening: SGPGIMS Experience" in Workshop on rare diseases in SGPGIMS, Lucknow on 22<sup>nd</sup> Jan 2025"
210. "Pedigree drawing" in 29th Indian Society of Assisted Reproduction (ISAR) 2025 Annual Conference, at Indira Pratithan, Lucknow on 9<sup>th</sup> Feb 2025
211. "Moderator for Panel discussion on Reproductive Genetics' in 29th Indian Society of Assisted Reproduction (ISAR) 2025 Annual Conference, at Indira Pratithan, Lucknow on 9<sup>th</sup> Feb 2025
212. "Genetic Counseling: The Right Career Choice for Bright Minds with Caring Hearts" in The National Genetic Counseling Symposium, 2025; held on 7th–8th March, 2025, at the Centre for DNA Fingerprinting and Diagnostics (CDFD), Hyderabad.

