**CURRICULUM VITAE**

**Personal details:**

Name: Dr. Deepti Saxena

Educational qualification:

* M.B.B.S., 2007, Gandhi Medical College, Bhopal, Madhya Pradesh
* M.S. (Obstetrics and Gynecology), 2010, Sarojini Naidu Medical College, Agra, Uttar Pradesh
* D.M. (Medical Genetics), 2015, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, Uttar Pradesh

Present designation:

Associate Professor,

Department of Medical Genetics,

Sanjay Gandhi Postgraduate Institute of Medical Sciences,

Lucknow, Uttar Pradesh

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Phone: 0522-2494322, 9554364760

**Work Experience:**

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| **Period** | **Designation** | **Institution** |
| January, 2017 to present | Assistant Professor,  Department of Medical Genetics | SGPGIMS, Lucknow,  Uttar Pradesh |
| 2012 to 2015 | Senior Resident (D.M.), Department of Medical Genetics | SGPGIMS, Lucknow,  Uttar Pradesh |
| 2010 to 2011 | Senior Resident, Department of Obstetrics and Gynaecology | R.N.T. Medical College, Udaipur, Rajasthan |

**Membership of Societies:**

* 1. Member of American Society of Human Genetics
  2. Member of European Society of Human Genetics
  3. Member of International Society of Prenatal Diagnosis
  4. Member of International Society of Ultrasound in Obstetrics and Gynaecology
  5. Member of Federation of Obstetric and Gynaecological Societies of India (FOGSI)
  6. Lifetime member of Society of Indian Academy of Medical Genetics (SIAMG)
  7. Lifetime member of U.P. Chapter of Obstetricians and Gynaecologists
  8. Lifetime member of Society of Fetal Medicine.
  9. Lifetime member of Indian Society of Perinatology and Reproductive Biology
  10. Lifetime member of Indian Society of Prenatal Diagnosis and therapy
  11. Lifetime member of Stillbirth Society of India

**Publications:**

1. Kandasamy S, **Saxena D,** Kishore Y, Phadke SR. Williams syndrome: a case series. Indian Pediatr. 2014May;51(5):411-2.
2. **Saxena D.** Next generation sequencing: window to a new era of molecular diagnostics. Genetic Clinics (GeNeXprESS). 2014 Oct-Dec;7(4):15-16.
3. **Saxena D**, Phadke SR. Prader – Willi syndrome due to an unbalanced chromosomal rearrangement. Genetic Clinics (Clinical Vignette). 2015 Jan-March;8(1):3-5.
4. **Saxena D**, Misra MK, Parveen F, Phadke SR, Agrawal S. The transcription factor Forkhead Box P3 gene variants affect idiopathic recurrent pregnancy loss. Placenta. 2015 Feb;36(2):226-31.
5. Srivastava P, **Saxena D**, Joshi S, Phadke SR. Consanguinity as an adjunct diagnostic tool. Indian J Pediatr.2016 March;83(3):258–260.
6. Mandal K, Ray S, **Saxena D**, Srivastava P, Moirangthem A, Ranganath P, Gupta N, Mukhopadhyay S, Kabra M, Phadke SR. Pycnodysostosis: mutation spectrum in five unrelated Indian children. Clin Dysmorphol 2016 Jul;25(3):113-20.
7. **Saxena D**, Srivastava P, Phadke SR. A novel heterozygous missense mutation in uromodulin gene in an Indian family with familial juvenile hyperuricemic nephropathy. Indian J Nephrol 2016 Sep;26(5):364‐7.
8. **Saxena D**, Agarwal M, Gupta D, Agrawal S, Das V, Phadke SR. Utility and limitations of multiplex ligation-dependent probe amplification technique in the detection of cytogenetic abnormalities in products of conception. J Postgrad Med 2016 Oct-Dec;62(4):239-41.
9. **Saxena D**, Srivastava P, Tuteja M, Mandal K, Phadke SR. Phenotypic characterization of derivative 22 syndrome: case series and review. J Genet. 2018 Mar;97(1):205-211.
10. 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;98:6.
11. Rai A, Mandal K, **Saxena D**, Lallar M, Phadke SR. Distal Arthrogryposis: A Clue to the Etiology of Neonatal Cholestasis. Indian J Pediatr. 2020 Oct;87(10):869-870.
12. **Saxena D**, Phadke SR. Prenatal diagnosis of congenital high airway obstruction syndrome: our experience from a tertiary care center. Int J Reprod Contracept Obstet Gynecol 2020;9:3858-61.
13. Rawool A, Gupta S, Singh B, Phadke SR, **Saxena D**, Mandal K. Recurrent miscarriage in North Indian population: a study of association of polymorphisms in genes coding for the natural killer: cell receptor natural killer group 2, member D and its ligand MHC class I chain-related protein A. Int J Reprod Contracept Obstet Gynecol 2020;9:3665-71.
14. Rawool A, Nilay M, **Saxena D**, Srivastava P, Moirangthem A, Mandal K, Phadke SR. A Case Series of Double Segment Imbalances: Delineation of Phenotypes and Comparison with Phenotypes of Isolated Copy Number Variations. Genetic Clinics 2020 Oct-Dec;13(4):7-15.
15. Singh S, Sardhara J, Raiyani V, **Saxena D**, Kumar A, Bhaisora KS, et al. Craniovertebral junction instability in Larsen syndrome: An institutional series and review of literature. J Craniovert Jun Spine 2020;11:274‐84.
16. **Saxena D**, Moirangthem A, Shambhavi A, Phadke SR. Koolen-de Vries syndrome: First report of two unrelated Indian patients. Am J Med Genet Part A. 2020;1–4. https://doi.org/10.1002/ajmg.a.62008.
17. 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.
18. 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 May 1;64(7):104235. doi: 10.1016/j.ejmg.2021.104235
19. 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 May 4. doi: 10.1002/ajmg.a.62241.
20. 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. 2021 Sep 29. doi: 10.1111/ijlh.13715.
21. 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 Sep 29;64(12):104345. doi: 10.1016/j.ejmg.2021.104345.
22. Srivastava S, Manisha R, Dwivedi A, Agarwal H, **Saxena D,** Agrawal V, Mandal K. Meckel Gruber and Joubert Syndrome Diagnosed Prenatally: Allelism between the Two Ciliopathies, Complexities of Mutation Types and Digenic Inheritance. Fetal Pediatr Pathol. 2021 Nov 25:1-11. doi: 10.1080/15513815.2021.2007434.
23. 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. 2021 Nov 29. doi: 10.1097/MCD.0000000000000407.
24. 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 Jan 6. doi: 10.1002/ajmg.a.62630.
25. 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 Feb 8;65(3):104447. doi: 10.1016/j.ejmg.2022.104447.
26. 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.
27. Sait H, Srivastava S, Saxena D. Integrated Management Strategies for Epidermolysis Bullosa: Current Insights. Int J Gen Med. 2022 May 24;15:5133-5144. doi: 10.2147/IJGM.S342740.
28. Dwivedi A, Moirangthem A, Pandey H. *et al.* Von Hippel–Lindau (VHL) disease and VHL-associated tumors in Indian subjects: VHL gene testing in a resource constraint setting. *Egypt J Med Hum Genet* **23**, 126 (2022). https://doi.org/10.1186/s43042-022-00338-1

**Research Projects:**

1. To study the phenotypic spectrum of a cohort of patients with Parry Romberg syndrome and to identify the copy – number variations by cytogenetic microarray.

**Principal Investigator, Intramural grant under SGPGIMS, 2018-2020,**

**Rs. 5,00,000**

1. Use of Next Generation Sequencing Techniques in identification of causative etiology in fetuses with abnormal ultrasound findings and identification of Novel Genes in Lethal Disorders.

**Principal Investigator, ICMR, 2020-2023, Rs. 94,37,898.**

1. Analysis of sequence variations in MEOX1, GDF3 and GDF6 genes in cases with congenital craniovertebral junction anomalies.

**Co-Investigator, Intramural grant under SGPGIMS, 2018-2020,**

**Rs. 5,00,000**

1. The Indian Movement Disorder Registry and Biobank: Clinical and Genetic Evaluation of Movement Disorders in Indian Patients

**Co-Investigator, Department of Biotechnology, 2018-21, Rs. 47,55,520**

1. Clinical and genotypic profile of arthrogryposis multiplex congenita.

**Co-Investigator, Intramural grant under SGPGIMS, 2019-2021,**

**Rs. 4,90,000**

1. Training of in-service Clinicians from Government Hospitals and Outreach Program for Aspirational Districts.

**Co-Investigator, Department of Biotechnology, 2019-2022, Rs. 1,23,64,000**

1. National Registry for Rare and other  inherited disorders (NRROID).

**Co-Investigator, ICMR, 2019-2024, Rs. 39,76,000**

**Awards:**

1. First prize for poster presentation at FOGSI International Conference Saving Mothers held at Gorakhpur, U.P. from 2nd to 4th August, 2019
2. Best poster presentation at 30th Annual conference of U.P. Chapter of Obstetrics and Gynaecology held at M.L.N. Medical College, Prayagraj, U.P. from 15th to 17th November, 2018
3. Certificate of Merit for securing highest marks in E.N.T. in M.B.B.S. examination
4. Certificate of honor and Gold Medal for getting distinction in the subject of E.N.T. in M.B.B.S. examination
5. Certificate of Merit in 1st Professional M.B.B.S. examination

**Invited Talks:**

1. Delivered a talk on “**Approach to a Neural Tube Defects, Hydrocephalus & interesting case studies**” in DHR sponsored training course on **“Pediatric Medical Genetics”** organized by **“Genetic research Centre, ICMR-NIRRCH, Mumbai”** from 20th June to 15th July, 2022.
2. Participated as a panelist in Webinar on **“Unravelling Fetal Mysteries”** organized by Lucknow ISOPARB in association with Kolkata ISOPARB on 18 May, 2022.
3. Varanasi
4. SBSI WEBINAR 26 March
5. Participated as faculty in Mid Term International Hybrid conference of ISOPARB on **“Prevention of Stillbirth”** organized by Gorakhpur Chapter of ISOPARB and Gorakhpur Obstetrics and Gynaecology Society on 13-14 November, 2021
6. Delivered a lecture on, **“An approach to neural defects and hydrocephalus”** in Virtual Training Course on Medical Genetics organized by **“Genetic Research Centre” at “ICMR- National Institute for Research in Reproductive Health, Mumbai”** from 5th to 30th July, 2021.
7. Delivered a talk on, **“Common fetal disorders – how to prevent?”** in International health Summit organized by Youth News online on 19 June, 2021.
8. Participated in panel discussion on **“Congenital Diaphragmatic hernia and Miscellaneous thoracic lesions”** in Webinar on Genetics of Fetal thoracic anomalies organized by Society of fetal medicine on 15 April, 2021.
9. Participated in panel discussion on **“Multicystic dysplastic kidneys and Hydronephrosis”** in Webinar on Genitourinary tract: Anomalies, Presentation, Genetics and Management organized by Society of fetal medicine on 18 March, 2021.
10. Talk on **“NIPS”** in SIAMG – ORDI Webinar Series on Unraveling Genetic Disorders on 7 November, 2020
11. Participated as faculty in workshop titled, **“Role of Imaging in Obgyn”** at 63rdAll India Congress of Obstetrics and Gynaecology” held at Lucknow from 29 January to 2 February, 2020
12. **“TORCH positive- What next?”** at “FOGSI International conference Saving Mothers” held at Gorakhpur, U.P from 2-4 August, 2019
13. **“Contingent screening for aneuploidy in the 1st trimester – what is intermediate risk” and “cf-DNA practical implications”** in CME on “Maternal – Fetal Medicine: an Update 2018” at Shri Ram Murti Institute of Medical Sciences, Bareilly, U.P on 4 Feb, 2018
14. **“Introduction of Hemophilia – Clinical aspects”** in CME on “Physiotherapy in Hemophilia” organized by Department of Transfusion Medicine, SGPGIMS, Lucknow on 29 April, 2017
15. **“Recent Advances in PCR”,** in the DBT Course in Biotechnology **“Techniques in Molecular Biology”**held at SGPGIMS, Lucknow from 15-30 September, 2015

**Poster presentations:**

1. **Saxena D**, Srivastava P, Phadke SR. A novel mutation in UMOD gene in an Indian family with familial juvenile hyperuricemic nephropathy. Indo - US Symposium on Genomic Insights into Human Morphogenesis Prenatal, Postnatal and Molecular Dysmorphology, 7-9 November, 2014, Hyderabad
2. **Saxena D**,Rawool A, Agarwal N, Phadke SR. Prenatal diagnosis of CHAOS – Report of 3 cases. 30th Annual conference of U.P. Chapter of Obstetrics and Gynaecology , 15-17 November, 2018, M.L.N. Medical College, Prayagraj, Uttar Pradesh
3. **Saxena D,** Mandal K, Phadke SR. Retrospective diagnosis by parental testing in NGS era. 5th Annual conference of SIAMG, 13-15 December, 2018, Christian Medical College, Vellore, Tamil Nadu
4. **Saxena D**, Rawool A, Phadke SR. Prenatal diagnosis of Emanuel syndrome. INSUOG, 3-5 May, 2019, Agra, Uttar Pradesh
5. **Saxena D**, Rawool A, Phadke SR. Prenatal microarray – solution or confusion? FOGSI International conference Saving Mothers, 2-4 August, 2019, Gorakhpur, Uttar Pradesh
6. Retrospective diagnosis by parental testing in NGS era**.** 26th International Conference on Prenatal Diagnosis and Therapy, organized by International Society for Prenatal Diagnosis, held virtually on 23 June, 2022.
7. Resolving fetal hydrops – a rare entity.26th International Conference on Prenatal Diagnosis and Therapy, organized by International Society for Prenatal Diagnosis, held virtually on 23 June, 2022.

**Workshops and Conferences attended:**

1. **Attended the course on “Exome sequencing” and “Non Invasive Prenatal Testing: The hot topics in 2022” at 26th International Conference on Prenatal Diagnosis and Therapy**, organized by International Society for Prenatal Diagnosis, held virtually on 23 June, 2022.
2. **26th International Conference on Prenatal Diagnosis and Therapy**, organized by International Society for Prenatal Diagnosis, held virtually from 20-22 June, 2022.
3. Varanasi
4. SAFOG Ian Donald School International Academic Exchange Webinar on **“Imaging the fetus”** held virtually on 27th January, 2022.
5. **ISUOG advanced neurosonography course: the dark side of the brain and changing the paradigm,** held virtually on 21 August, 2021.
6. **“AICC RCOG Master Class” on Maternal Fetal Medicine**, held virtually on July 4, 2021.
7. 6th Annual International Conference on**, “Genomics and genetic counselling – Value in Health Care”,** held virtually from 2-4 July, 2021.
8. **ISPD 25th International Conference on Prenatal Diagnosis and Therapy,** held virtually from 6-8 June, 2021.
9. **ISUOG Education course: Diagnosis and Management of Congenital Infections,** held virtually on 15 May, 2021.
10. **ISPD-Virtual Education Series,** held virtually from 7 October- 11 November, 2020.
11. **2020 Annual meeting of the American Society of Human Genetics**, held virtually from 26-30 October, 2020.
12. **63rdAll India Congress of Obstetrics and Gynaecology**, 29 January- 2 February, 2020, Lucknow, Uttar Pradesh.
13. **Fetal Cardiocon 2019**, 23-25 August, 2019, Gurugram, New Delhi.
14. **FOGSI International Conference Saving Mothers**, 2-4 August, 2019, Gorakhpur, Uttar Pradesh.
15. **INSUOG 2019**, 3-5 May, 2019, Agra, Uttar Pradesh.
16. 4th International conference on Birth defects (**ICBD 2018),”Birth defects in Genomic Era”,** 12-15 December, 2018, Christian Medical College, Vellore, Tamil Nadu.
17. 30th Annual conference of U.P. Chapter of Obstetrics and Gynaecology (**UPCOG-2018**), 15-17 November, 2018, M.L.N. Medical College, Prayagraj, Uttar Pradesh.
18. **Fetal Cardiocon 2018**, 10-12 August, 2018, Gurugram, Haryana.
19. CME on **“Rare disease update 2018”** organized by Indian Society of Inborn Errors of Metabolism, 28 July, 2018, Lucknow, Uttar Pradesh.
20. CME on **“Maternal – Fetal Medicine: An update 2018”,** 4 Feb, 2018, Shri Ram Murti Institute of Medical Sciences, Bareilly, Uttar Pradesh.
21. 13th Asia Pacific Congress of Maternal Fetal Medicine, **“FetalMed 2017,”** 1-3 September, 2017, Gurugram.
22. CME on **“Diagnosis of Genetic Disorders in NGS Era”,** 21 August, 2017, SGPGIMS, Lucknow.
23. International Symposium on **“Genetics and Genomics in Modern Clinical Medicine”,** 9-10 February, 2015, King George Medical University, Lucknow.
24. Indo - US Symposium on **“Genomic Insights into Human Morphogenesis Prenatal, Postnatal and Molecular Dysmorphology”**, 7-9 November, 2014, Hyderabad.
25. **“Workshop on Molecular Cytogenetics”**, 6 November, 2014, Hyderabad.
26. International Symposium by Indo – UK Genetic Education Forum on **“Current Trends in Genetic and Genomic in Medicine”** 31 January, 2013, King George Medical University, Lucknow.

**Courses done:**

1. **“ISUOG advanced neurosonography course: the dark side of the brain and changing the paradigm”,** held virtually on 21 August, 2021.
2. **“Human and Mammalian Genetics and Genomics: The McKusick short course”** held virtually from 19-30 July, 2021.
3. **“Basic course in Biomedical Research”** mandated by National Medical Commission.
4. Training course on **“Genome Sequencing: Methods and Applications”** at ICAR-National Bureau of Fish Genetic Resources, Lucknow, 12-17 March, 2018.
5. DBT Course in Biotechnology **“Techniques in Molecular Biology”** at SGPGIMS, Lucknow, 15-30 September, 2015.
6. Training in ultrasonography from Department of Radiodiagnosis, R.N.T. Medical College, Udaipur, Rajasthan, 1-30 April, 2011.
7. **“5th Ian Donald Obs Gyn Ultrasound Course”** at Agra, 5-6 September, 2009.

**Reviewer for journals:**

* American Journal of Medical Genetics
* Indian Journal of Pediatrics
* Neurology India

**Organization of workshop/ conferences:**

1. Sixteenth ICMR Course on Medical Genetics and Genetic Counseling, from 24th July to 5th August, 2017 at SGPGIMS, Lucknow
2. Seventeenth ICMR Course on Medical Genetics and Genetic Counseling, from 20th August to 1st September, 2018 at SGPGIMS, Lucknow
3. Eighteenth ICMR Course on Medical Genetics and Genetic counseling, Pedigree to Genome from 26th August to 7th September, 2019 at SGPGIMS, Lucknow