

Shubha Phadke, Professor and Head, Department of Medical Genetics,
Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, India.

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Qualifications: M.B.B.S. (1982), M.D. (Pediatrics) 1986, DM (Medical Genetics) 1992

About Dr Shubha Phadke

Dr Shubha Phadke has an experience of 30 years in the field of clinical genetics and laboratory genetics. She runs the first DM in Medical Genetics program in India since 1996 and has contributed to the establishment of the specialty of Medical Genetics in India by training most of the medical geneticists [more than 40] in India. Her interests are pre and postnatal dysmorphology, genetic hematology and she has 262 publications in national & international journals to her credit. She has reported 10 new syndromes and has identified causative gene 4 of them. ‘Genetics for Clinicians’ is the book written by her and has popularized genetics amongst clinicians. She is the founding president of Society for Indian Academy of Medical Genetics and founding editor of Genetics Clinics, a three monthly publication of SIAMG. She is on the research committees of ICMR, DBT and on the editorial boards of renowned journals like American Journal of Medical Genetics, Medical Genetics & Genomic Medicine, etc. A two week long yearly training program for clinicians, ‘ICMR Course in Medical Genetics & Genetic Counseling’ is held regularly for last 16 years and has introduced more than 500 medical doctors to the fascinating world of medical genetics. She is recipient of prestigious awards like Hargobind Foundation Fellowship [1994], **International Scholarship of Clinical Genetics Society of UK [2009]** and Dr I C Verma Outstanding Researcher Award [2016]. Dr Phadke has established Newborn Screening Program for Uttar Pradesh funded by National Health Mission and is successfully running since 2015. Dr Phadke is involved actively with the activities of patient support groups for thalassemia, hemophilia, Down syndrome and rare diseases.

Dr Phadke is heading a department of Medical Genetics in an academic institute providing tertiary healthcare to a large state of Uttar Pradesh. The department of Medical Genetics being the only department in the state, gets referral from Uttar Pradesh and neighboring states. The department provides clinical and laboratory services to patients of all spectrum of genetic disorders and presentations of all organ systems and has a large load of patients. The department has facilities for prenatal diagnosis, fetal autopsy in addition to molecular (Sanger sequencing, analysis of NGS) and cytogenetic laboratories (including in-house cytogenetic microarray).

Research and Achievements

- Publications: 262, OMIM citations – 22 < <https://www.omim.org/entry/607539>>
- Ten novel syndromes and identification of genes for 5 novel phenotypes
- Citations in and contributions to London Dysmorphology Databases
- Recipient of I C Verma Award for Excellence in Research [2016]
- Funding till date – Rs 760 lac (760,00,000-00)
- Founding President of Society for Indian Academy of Medical Genetics < <http://iamg.in/>>
- Founding Editor of Genetics Clinics < http://iamg.in/genetic_clinics/index.php>
- On Editorial board of American Journal of Medical Genetics, Indian Pediatrics, Molecular Genetics & Genomic Medicine

EXPERIANCE

Institution	Position	Period	Duration
Department of Medical Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow, India	Professor & Head of the department	Jan 2006	Till date
	Additional Professor	July 2004	Jan 2006
	Associate Professor	July 2000 to June 2004	4 years
	Assistant Professor	April 1996 to July 2000	4 years
Lucknow [Private clinic]	Private practice	Aug 1994 to April 1996	1 year 11 months
Department of Pathology, University of Washington, Seattle, USA	Fellowship	April 1994 to July 1994	3 months
Department of Medical Genetics, SGPGIMS, Lucknow, India	Research associate	1 st Feb 1993 to 31 st Jan 1994	1 year
	Senior Resident (Medical Genetics)	Jan 1990 to Dec 1993	3 years
	Research officer (Medical Genetics)	Feb 1989 to Dec 1989	11 Months
Nagpur, India [Private clinic]	Private Practice	Jan 1987 to Jan 1989	2 years

Recent Research Projects:

1. Centre for Molecular Medicine: Indian Council of Medical Research. Rs 499 lac, for 5 years. Sanctioned on 21-3-12. (Completed)
 2. Evaluation of utility of cytogenetic microarray in detection of etiology of Prenatally detected malformation Rs 3 lac. Intramural. 1st Nov 2012 to 31st oct 2014 (Completed)
 3. Multicentric Collaborative Study of the Clinical, Biochemical and Molecular Characterization of Lysosomal Storage Disorders in India. PI – Shubha Phadke. Indian Council of Medical Research (Rs 31.3 lac for 3 years- Sanctioned on 10 th Dec 2014)
 4. Genomic studies into limb malformations and related syndromes. Department of Biotechnology. Sanctioned on 03/03/2015 for 3 years. Rs 97,97,160. PI – Shubha Phadke. (Completed)
 5. The Indian Movement Disorder Registry and Biobank: Clinical and Genetic Evaluation of Movement Disorders in Indian Patients” Sanctioned by Department of Biotechnology for 3 years in Oct 2018 (PI – Shubha Phadke) (Rs. 4755520)
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