CURRICULUM VITAE (MEDICO-LEGAL): PROFESSOR DHAVENDRA KUMAR

APPOINTMENTS

Current Clinical Practice:

1. Consultant in Clinical Genetics with Special Interest in Cardiovascular Genetics & Genomic Medicine, Director- The Genome Clinic, Spire Cardiff Hospital, Spire Healthcare, Cardiff, UK.

2. Hon. Consultant in Cardiovascular Genetics, Inherited Cardiac Diseases Unit, Department of Cardiology, St. Bart's Hospital, London, UK.

3. Honorary Senior Consultant Adviser, Apollo Genomics Institutes, Apollo Group of Hospitals, India.

Current Academic appointment:

Hon. Clinical Professor, William Harvey Research Institute, Bart's and the London School of Medicine & Dentistry, Queen Mary University of London, UK

Honorary Professor, Faculty of Medicine, Swansea University School of Medicine, Swansea, Wales, UK

Recent NHS appointment:

- **Consultant in Clinical Genetics**, All Wales Medical Genetics Service, University Hospital of Wales, Institute of Medical Genetics, Cardiff University, UK (1 November 2204 to 31 October 2017)

Academic appointments:

- **Honorary Senior Research Fellow,** Institute of Cancer & Genetics (previously Institute of Medical Genetics), Cardiff University, 2008 to date.
- **Visiting Professor**, Genomic Policy Unit, Faculty of Health, Sport & Science, The University of South Wales (previously Univ. of Glamorgan), Wales, UK; 2010-date
- **Visiting Professor,** Education & Research, Chettinad Health University, Chennai, India; 2011 to date
- **Adjunct Professor,** Public Health/ Medical Genetics, China Capital Medical University, Beijing, China; 2010- date
- **Visiting Professor**, Ranbaxy Science Foundation, India; January/ February 2013
- **Senior Faculty Adviser,** Centre for Precision Medicine and Health, King George's Medical University, Lucknow, India, 2013- date
- **Adjunct Professor,** Kasturba Medical College, Manipal University, Karnataka, India, March 2017 to Date.

- **Adjunct Professor,** JSS Higher Education Academy (Deemed University), Mysore, Karnataka, India; 2019 to date.
- **Adjunct Professor**, Department of Biotechnology, Shri Mata Vashno Devi Unversity, Katra, Jammu & Kashmir, India.
- **Honorary Professor,** Faculty of Medicine, University of Colombo, Sri Lanka. January 2022 to date

PERSONAL

Nationality British- U.K. Citizen

Defence Organisation Medical Protection Society No. 281124

General Medical Council Full Registration No: 2351409

Professional Education:

Medical School/College attended- King George's Medical College, Lucknow University, Lucknow, India

Undergraduate: August 1968 - July 1973
Postgraduate: January 1976 - December 1977

The Medical School, University of Sheffield, UK

Postgraduate (Research): April 1982 - December1986

***Qualifications*:**

M.B.,B.S.,1973 (King George's Medical College, University of Lucknow, India)
MD 1977 in Paediatrics (University of Lucknow, India)
D.C.H 1979 (Royal Colleges of Surgeons and Physicians in Ireland- RCS&PI)
MRCPI.,1990; ***FRCPI***.,1992 (RCP Ireland);
MRCPCH ,1996 (RCPCH,UK)
FRCPCH 2006 (RCPCH, UK)
MMedSci 1987 (University of Sheffield, UK)
DABMG 1990, Diplomate American Board Certification in Clinical Genetics
PG Cert. Med Edu , 2003 (Staffordshire University, UK)
FACMG, 2003 (Fellow American College of Medical Genetics)
FRCP 2008 (Fellow RCP London, UK)
DSc (Honaris Causa) 2015, King George Medical University, Lucknow, India.

PROFESSIONAL

Professional and Specialist training:

Paediatrics*:* Five years post medical qualification and registration training in Paediatrics with 10 years experience in Community Paediatrics.

Medical/ Clinical Genetics:
Over 25 years experience in medical genetics including general clinical genetics, clinical dysmorphology (malformation syndromes), paediatric genetic disorders, neuro-genetics, cancer genetics, and cardiovascular genetics.

Specialist Accreditation*:*

- **General Medical Council (UK): Specialist Register- Clinical Genetics**
- **American Board of Medical Genetics- Clinical Genetics**

Medico Legal Practice

- Harley Street Genetics Clinic, 10 Harley Street, London, W1 (1997- 2000)
- The Genome Clinic, Spire Cardiff Hospital, Cardiff (2012 to date)
- Medico-legal practice (1994 to date)-

1. Mobile-Doctors Ltd.- Assessment and reports on personal injury cases, alleged medical negligence and expert witness reports (1996-2003)
2. Individual expert reviews and reports on wide ranging paediatric and Adult genetic conditions on instructions from solicitors in Sheffield, Cardiff, London, Scotland and many others from all over UK (names of instructing solicitors withheld, can be provided on request)
3. Expert reviews and reports including expert witness presentation and cross-examinations in Family & Civil Courts related to alleged non-accidental injury vs. genetic susceptibility for recurrent childhood fractures. (Details available on request, subject to confidentiality)
4. Expert review, report and Court witness evidence in Criminal cases related to serious injuries and child's death on behalf of both criminal prosecution service and defence team alleging related to possible underlying genetic or inherited medical condition mitigating circumstances and evidence.
5. Medical/Clinical negligence cases arising from missed genetic diagnosis and/or inappropriate and delayed care and treatment.
6. Expert review and secondary opinion on previous cases under review or appeals process.

Examples of cases dealt with include-

Neurofibromatosis type 1: Alleged medical negligence for incorrect diagnosis and information

Ehlers- Danlos syndrome: Father accused for non-accidental injury related to recurrent bruising and haematomas

Chromosomal translocation: Expert report on adoption and child protection case

Fetal alcohol syndrome: Alleged maternal alcohol consumption during pregnancy leading to growth and developmental delay

Brittle bone disease/ Osteogenesis imperfect: Alleged parental neglect and role in non-accidental injury (recurrent fractures); One case young girl died with multiple fractures

Child with recurrent bruising due to inherited connective tissue disease- Alleged parental negligence and accusation of non-accidental injury; exclusion of Ehlers-Danlos syndrome by the next generation sequencing multi-gene panel or targeted whole exome sequencing.

X-linked mental retardation syndrome (excess of affected males with learning/ developmental delay in the family): Defence of a mother accused of causing her sons' learning and behavioural problems due to her drug and alcohol misuse during all pregnancies. In fact she was confirmed to be a carrier for an X-linked mental retardation syndrome, cause for her sons' developmental problems.

Inappropriate medication in a dysmorphic syndrome: Report on a known case with a genetic syndrome where long standing medical neglect and inappropriate medication led to unusual and unexpected complications.

Clinical negligence in the care pregnant mother with epilepsy: Child is suspected to have Fetal Valproate Syndrome

Genetics review in a deceased child with multiple fractures and brain injury: major criminal investigation.

Alleged clinical negligence by a GP for **incorrect diagnosis of an inherited chronic neurological disease.**

Alleged negligence on the part of an NHS Trust for **inappropriate surgical intervention based on misinterpretation of a breast/ ovarian cancer genetic (BRCA) testing.**

Alleged negligence for **the care of a young person with Prader- Willi syndrome**

Causation reports following acceptance of the Breach of Duty with specific reference to a known or suspected genetic or inherited disease.

Exclusion of genetic condition and/or inherited susceptibility manifesting with internal bleeding, particularly in a new born with bleeding inside the brain, such as subdural or periventricular haemorrhages.

Genetic causation testing/ opinion in pregnancy related perinatal or neonatal complications. These are mostly replated to the baby born with suspected hypoxic ischemic encephalopathy.

- **Attendance and active participation in Experts' meeting with the Counsel and the Legal team**

- **Court attendances-**

- Five Family Courts- Derby (1), Bristol (2), Oxford (1), York (1)
- Two Civil dispute- Royal Courts of Justice, London & District Court, Leeds
- Three Criminal cases- Cardiff, London & Swansea Crown Courts

Medico-Legal Courses & Training:

1. Attended Bond Solon 'Expert Report' writing course- Birmingham, April 2001
   Continued self- learning and development through regular reviews and online information

2. MBL Seminars: Course on Report Writing and Court Room skills for Expert Witnesses
   By Augustus Ullstein QC, 16th Feb. 2016, London

3. Bond Solon 'Expert Witness Report Writing Course', accredited by the Cardiff University, Feb 2018, London.
4. Continuous professional development through reviews of articles, web searches and participating in webinars on relevant subjects related to a particular case.

5. Regular viewing and reflections on Webinars related to medico-legal cases and advisory reports offered by professional organisations such as Medical Protection Society and MBL Seminars

6. MBL Seminar- Court Room Skills- Preparation and Presentation including cross examination

Medico-Legal Expert Witness Database/ Directory entries-

Medico-legal / Expert Witness profile is listed in the following professional resources:

Genome Clinic- Spire Healthcare (www.spirehealthcare.com)

The Expert Witness Directory (www.expertwitness.co.uk)

National Expert Witness Agency (www.NEWA.expert)

Speed Medical (www.speedmedical.com)

Primex plus (www.primexplus.com)

UK Register for Expert Witnesses (www.jspubs.com)

Medical Expert Witness Agency (www.MEWA.org.uk)

CLINICAL GENETICS SKILLS AND COMPETENCIES

A. Clinical Genetics Experience- Consultant in Clinical Genetics

- General Clinical Genetics
- Paediatric genetics- clinical dysmorphology & Neuro-developmental disorders
- Inherited connective tissue diseases including skeletal dysplasia and Ehlers-Danlos syndrome.
- Clinical cardiovascular genetics

As the Lead Consultant for Cardiovascular Genetics service for Wales, I was actively involved in a number of clinical, academic and professional activities. I organised weekly multi-disciplinary Inherited Cardiac Conditions team for developing an integrated 'All Wales Service for Inherited Cardiovascular Conditions" within the AWMGS:

- Actively pursued service development for Wales cardiac genetic service
- Prepared and implemented the clinical service plan and protocols
- Prepared and implemented the clinical care pathways for clinical referrals, molecular genetic testing and managing family members with a history of 'sudden cardiac death'.
- Organized the NHS multi-disciplinary team for providing a World class service for patients and families with inherited heart disorders across Wales
- Active participation in setting up the National level service for families affected with sudden cardiac death in a close relative as laid down in the National Service Framework for England and Wales.
- Organised and successfully led the All Wales Cardiovascular Genetics Interest Group
- Contacts with Wales section of major patient support groups- for example SADS, CMA, CRY and AA
- Organised highly successful and popular the International symposium on clinical cardiovascular genetics in Cardiff (2007 to 2017).
- Founder member and active leadership for the UK Association for Inherited Cardiovascular Conditions (AICC) affiliated to BSHG and British Cardiovascular Society (BCVS)
- Member of the UK Cardiac Genetics Advisory Group of the Public Health Genomic Foundation, Cambridge

B. Academic commitments:

- **Teaching & Training**
  - Undergraduate (MB) teaching Cardiff & Swansea
  - Intercalated BSc in Medical Genetics, Cardiff University
  - Educational supervisor- Clinical Genetics
  - MSc in Genetic Counselling, Cardiff University
  - Co-Supervisor, PhD, University of Bristol (2009 to present)
  - External examiner DSc., Queens University Belfast, N-Ireland, 2010.
- **Accredited courses and seminars (since 2005)-** active participation in developing, leading and organisation of the following professional meetings, seminars and conferences:
  - **Genetic disorders and people from the Indian subcontinent-challenges for clinical**

services in the NHS. Two-day symposium organised in Cardiff, with Wales Gene Park, 22-23 November, 2005.

- "**Challenges of genetics and genomics for the Indian subcontinent**", organised a pre-congress satellite symposium at the 11th International Congress of Human Genetics, 6-10 August, Brisbane, Australia.
- **"Challenges of cardiovascular genetics- The Cardiff symposium"-** 22/23 November 2007 and 23/24 Nov. 2009 Cardiff. Two day educationally approved symposium to focus on recent developments, current practice and issues in delivering the cardiac genetic services in the UK.
- **"Clinical Medicine in the Genome Era",** 26-27 February 2009, Cardiff. A two day International symposium to review recent developments in genome sciences and technologies as applied to the practice of modern medicine.
- **"Short course on clinical cardiovascular genetics",** 30-31 January 2010, Bangalore, India
- **"Current trends in Clinical Genetics & Genomic Medicine",** 21-24 March 2011, Chettinad Health City, Chennai, India
- **"Current practice of genetic and genomic medicine",** 26-27 March 2011, Apollo Hospital, New Delhi, India
- **"Genetics and genomics in developing countries',** 5-7 September 2011, Annual conference of the British Society of Human Genetics, University of Warwick, UK
- **"Inaugural meeting of the Association for Inherited Cardiac Conditions",** 23 November 2011, Cardiff, UK
- **"The Third Cardiff Symposium on Cardiovascular Genetics",** 24-25 November 2011, Cardiff, Wales, UK
- **"FH and Cardiac study day",** part of the local training and continuous professional development programme for genetic counsellors and related health professionals; 26th January 2012, Institute of Medical Genetics, Cardiff**.**
- **"Genes and Human Malformations- Indian Birth Defects Conference", 27-29 February 2012, Bangalore, India; part of the Indo-UK Genetic Education Forum,** jointly supported by CGS/BSHG and hosted by the Centre for Human Genetics, Indian Institute of Science, Bangalore, India
- **"The Second International Genomics Conference- Genomics for Healthcare and Socioeconomic progress", 13-14 September 2012,** sponsored by the Human Genome Organisation-International and hosted by the Wales Gene Park, Cardiff University.
- **"The First Indian Cancer Genetics Conference",** 23-25 January 2013, Advanced Center for Treatment, Research and Education for Cancer (ACTREC), Tata Memorial Cancer Trust, Mumbai, India; jointly organised by the Indo-UK Genetic Education Forum and ACTREC.
- **"The Next Revolution of Genetics and Genomics",** 27-29 January 2013, Postgraduate Education Centre, Ram Manohar Lohia Hospital, New Delhi; jointly organised by the Indo-UK Genetic Education Forum with the Indian Academy of Medical Genetics and the Centre for Genetic Medicine, Sir Ganga Ram Hospital, New Delhi, India.
- **"Current Trends in Genetic and Genomic medicine",** 31 January 2013, Dr Ram Manohar Lohia Post Graduate Institute of Medical Sciences, Gomti Nagar, Lucknow; jointly organised by the Indo-UK Genetic Education Forum

- **"Gains of Genomic Research for Medicine and Biology",** The Ranbaxy Science Foundation Symposium, 4th February 2013, Institute of Immunology, New Delhi, India.
- **"The Fourth Cardiff Cardiovascular Genetics Symposium",** organised by the Wales Gene Park, Cardiff University, 21-22 November 2013.
- "International Birth Defects Conference", Jointly organised on behalf of the Indo UK Genetic Education Forum, Wales Gene Park and the Human Genetics Unit, The University of Colombo, Sri Lanka; 9-11 Feb. 2014.
- Ophthalmic Genetics Seminar, 11 Feb 2014, Jointly organised on behalf of the Indo UK Genetic Education Forum, Wales Gene Park and the Human Genetics Unit, The University of Colombo, Sri Lanka.
- Indian Ophthalmic Genetics Conference, 15-16 Feb 2014, Jointly organised on behalf of the Indo UK Genetic Education Forum, Wales Gene Park and Narayana Nethralaya, Bangalore. India.
- "Genes and Genome", 21st Feb 2014, invited speaker to the Seminar organised by the Dept. of Human Genetics, Guru Nanak Dev University, Amritsar, Punjab, India.
- "Joint UK-Dutch Clinical Genetics 2014", 17-19 March 2014, Leiden, The Netherlands.
- Symposium on 'Genomics in Developing Countries', at the Annual Conference of the British Society of Genetic Medicine (BSHG), 17-19 September 2014, Liverpool Convention Centre, Liverpool, UK
- "Familial heart and blood disorders- International symposium", 31 Jan-1 Feb 2015, Post Graduate Institute of Medical Education and Research, Chandigarh, India.
- "Seminar on Genomic and Molecular Medicine:, 3 Feb 2015, Himalayan Institute of Medical Sciences, Dehradun, Uttarakhand, India.
- "Genetics and Genomics basis of Clinical Medicine", 9-10 Feb 2015, King George's Medical University, Lucknow, UP, India.
- Clinical Genetics Society (UK), Spring Conference, 3rd March 2015, Royal College of Physicians, London.

Other relevant information

1: Academic and Professional publishing

- **New Journals:**

1. ***The HUGO Journal*** (formerly- Founding Editor in Chief ***Genomic Medicine),*** Springer, Dordrecht, The Netherlands

The scope and remit of The HUGO Journal is very wide and includes:

- Genomic sciences and human disease: e.g., pharmacogenomics, cancer genomics, genetics of Mendelian disorders, genetics of complex diseases
- Genome structure and genetic variation
- Comparative genomics between humans and other organisms
- Microbial genomics of organisms pertinent to human disease
- Population genetics
- Human evolution and migration
- Genome-to-systems strategies in systems biology
- Genomic technologies
- Genomics and genetic epidemiology
- Computational genomics
- Ethical, legal, and societal research relevant to genomics and human genetics
- Science policy analyses pertinent to genomics and genetics.
- Genomic and genetic research from emerging and developing countries.

[*Kumar and Liu (2009). The emergence of the HUGO Journal. Hugo J. 2009 December; 3(1-4):1–2]*

2. "Applied and Translational Genomics", Elsevier, Online & Open Access

The journal publishes reviews and original research papers in many genomic areas including- Comparativegenomics • Applied bio-systems • Applied biotechnology (laboratory development and support) • Clinical cytogenomics (molecular cytogenetics) • Clinical genomics (diagnostics) • Genomic and personalized medicine (evidence-based medicine) • Public/population health (Screening and targeting selected population; large scale infection control and preventive measures) • Pharmaceutical industry (new drugs and vaccines) • Pharmacogenomics • Plant and crop genomics (Agricultural genomics) • Marinegenomics • Animal and Veterinary genomics • Bioeconomy • Education and media (professional and public education; bio-information through on-line/media resources) • Ethical, legal and social issues in translational genomics • Legal and Regulatory including Intellectual property rights

- **Books (Author/Editor)**

  - "*Genetic disorders of the Indian subcontinent*", Springer-Kluwer, Dordrecht, 2004
  - "*Genomics and Clinical Medicine",* Oxford University Press, NewYork, 2008
  - "*Principles and Practice of Clinical cardiovascular genetics",* Oxford Univ. Press, New York, 2010
  - "*Oxford Handbook of Inherited Cardiac Disease*", Oxford Univ. Press, Oxford (2011)
  - "*Genomics and Health in the Developing World'*, Oxford Univ. Press, New York (2012)
  - **"***Genomic and Molecular Medicine*", E-book series, Morgan and Claypool, USA (2012- contd.)
  - **"***Principles and Practice of Genomic Medicine", Oxford University Press, New York, 2015.*
  - *"Medical and Health Genomics", Academic Press, Elsevier, Cambridge, 2017*

- *"Genomics and Society", Academic Press, Elsevier, Cambridge, 2018.*
- *"Clinical Molecular Medicine- Principles and Practice", Academic Press Elsevier, 2019.*

- **Theses**

1. **MD (Paediatrics**) University of Lucknow, India 1977.
"A study of plasma and cerebrospinal fluid electrolytes in certain childhood neurological disorders with special reference to magnesium"

2. **M Med Sci (Medical Genetics**) University of Sheffield 1987.
"Genetic studies in hereditary cerebellar ataxia and related disorders"

3. **D.Sc.,** King George's Medical University, Lucknow, India. 2015.
"The Clinical and Genetic Heterogeneity of Inherited Human Diseases- Personal observations and Interpretations"

- **List of Publications- Book chapters, original papers, reviews etc.**

Around 100 published in various journals, books and web resources- full list available on 'Google Scholar' search; hard copies available on request.

2: Research & Development

Research Management & Governance

1. R&D Lead for All Wales Medical Genetics Service, 2011- date
2. R&D Lead for the Specialist Services Division, Cardiff & Vale University Health Board (CVUHB), 2011 to date
3. Member of the CVUHB Research Governance Board, 2011- date.
4. Member of the Steering Committee, Research Design and Conduct Service (RDCS), National Institute of Social and Healthcare Research (NISCHR)
5. External referee for research grants and extra-mural funding- MRC UK; Wellcome Trust UK, European Research Council, South African Medical Research Council, Leading research funding foundations in Germany, The Netherlands, Belgium and Italy.
6. External assessor and adviser on higher academic appointments- Glasgow, Cardiff, Leicester, UCL London, King's College London, Johns Hopkins University USA, University of Cape Town and University of Jordan.

Cardiovascular genetics research:

The current lead for cardiovascular genetics research in collaboration with the Institute of Molecular and Experimental Medicine & the Wales Cardiovascular Registered Research Group of NISCHR.

The current research projects include:

1. **'Aortic dilation prevention by Ibesartan in Marfan Syndrome (AIMS)**- Lead investigator for Wales in this BHF funded multi-centre clinical trial coordinated by the Royal Brompton Hospital, London.
2. **'Genomic profiling in chemotherapy related cardiomyopathy'. T**his study aims to carry out genomic profiling in chronic heart failure due to chemotherapy related cardiomyopathy using the next generation sequencing methods.
3. '**Molecular autopsy in sudden cardiac death'** a collaborative multi-centre study with 'The Heart Hospital', London to explore the utility of molecular autopsy in assisting coroners in establishing the cause of unexplained sudden death.
4. **'Unmet needs of families in Wales affected by sudden arrhythmic death syndrome (SADS): A focus group and interview study'-** this study aims at studying to identify unmet information and support needs of families across Wales who have experienced a sudden unexplained death in a close family member. Part of higher degree (DEd) thesis, the University of Bristol, UK.
5. '**Molecular heterogeneity of ion channel genes in sudden arrhythmic death syndrome (SADS)**'- This study is focussed on extending molecular analysis in affected patients with SADS including whole genome sequencing.
6. **'Family and community experiences of sudden unexplained/cardiac death including the role of coroner'**, funded by the UK Education and Social Research Council (ESRC) and the Sudden Arrhythmic Death Syndrome (SADS-UK), charity; registered as a PhD project, Centre for social and economic applications of genomics (Cesagen), Cardiff University, UK

3: Presentations at Professional meetings and Invited talks

Regular participation and presentations at several national and international professional and scientific conferences- List available on request

4: Other professional activities related to genetics and genomics:
Actively associated with several leading Professional and Scientific Genetic and Genomic Organizations-

1. Human Genome Organisation International (HUGO)
    - Founding Editor for the HUGO Journal (2009 to date
    - Member of the HUGO Publications Committee (2009 to date)
    - Member of the HUGO Council (2010 – 2012)
    - Member of the HUGO Educational and Public Policy Committee (2011)
    - Chair- HUGO Education Committee (2022)
2. British Society of Human Genetics- 2011:Organised and leading a dedicated session on '**genetics and genomics in developing world'** at the annual meeting (5-7 September 2011, University of Warwick)
3. **'Genomic research and teaching collaboration'**, Visiting Professor in Clinical Genetics: organised and leading the Clinical Genetics module for postgraduate degree

course with Chettinad Health University, Academy for Research and Education, Chennai, India.

4. **'Genetic and genomic education for health professionals'**- active participation in teaching and training of nursing and allied health professionals for developing basic skills and competencies in medical genetics; this is delivered through the Genomic Policy Unit at the University of Glamorgan in collaboration with the National Centre for Genetic Education on behalf of the Department of Health, UK.
5. '**Promoting genetics and genomics in developing countries'**
   Established close links with several genetic and clinical colleagues in India, China, The Phillipines, Thailand, United Arab Emirates, Saudi Arabia, Israel, Brazil, South Africa and Argentina. Currently I am actively pursuing genetic and genomic education through dedicated educational seminars and workshops. I led the dedicated session on this at the BSHG annual conference (5-7 September 2011) held in Warwick University. Founded and Lead the '**Indo-UK Genetic Education Forum'**, 2010 to date.

5. Member, Expert Panel for **WHO Grand Challenges Project on Genomics and Public Health in Developing Countries**, July 2011 to date
6. Member of the International Advisory Board, **Indian Academy of Medical Genetics**, September 2012 to date.
7. Founder of **the Genomic Medicine Foundation UK** (www.genomicmedicine.org)
8. Founder/ Chair- **Global Consortium for Genomic Education (GC4GE)**
9. **Founder/ Chair- Global Familial Heart Challenge (GFHC)**
10. **Founder/ Chair- UK India Genomic Medicine Alliance (UKIGMA).**

Written and Revised by Professor Dhavendra Kumar
Cardiff, Revised 1 May 2023