

**Curriculum Vitae**

**of**

**Dhavendra Kumar**

MD, MMedSci, FRCP, FRCPI, FRCPCH, FACMG

**CONSULTANT IN CLINICAL GENETICS  
HON. SENIOR RESEARCH FELLOW**

**INSTITUTE OF MEDICAL GENETICS**

**CARDIFF UNIVERSITY  
University Hospital of Wales,  
Cardiff and Vale NHS Trust, Cardiff**

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## **1. SUMMARY: CURRICULUM VITAE OF DR. DHAVENDRA KUMAR**

***Present post:*** CONSULTANT IN CLINICAL GENETICS/ HON. SENIOR RESEARCH FELLOW, INSTITUTE OF MEDICAL GENETICS, UNIVERSITY HOSPITAL FOR WALES, CARDIFF UNIVERSITY, UK.

***Qualifications:*** *M.B.,B.S.*,1973; *MD* 1977 in Paediatrics (University of Lucknow, India); *D.C.H* 1979 (Royal Colleges of Surgeons and Physicians in Ireland); *M.R.C.P.*,1990; *F.R.C.P.*,1992 (Fellow RCPI); *M.R.C.P.C.H* ,1996 (RCPCH); *M. Med. Sci* 1987 (University of Sheffield); *DABMG* certification in Clinical Genetics 1990 (ABMG); *PCME* (Postgraduate Certificate in Medical Education) 2003 (Staffordshire University); *FACMG, 2003* (Fellow American College of Medical Genetics); *FRCPCH* , 2006 (Fellow-RCPCH); *FRCP* , 2008 (Fellow RCP London)

***Postgraduate Medical Training/Experience in Paediatrics:*** Five years post medical qualification and registration training in Paediatrics with 10 years experience in Community Paediatrics.

### ***Specialist Training and Experience in Medical 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.

***Special interests:*** Clinical dysmorphology, Cardiovascular genetics, Genomic medicine

### ***Specialist Accreditation:***

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

***Academic achievements:*** Experience in organised research and applied clinical research as reflected in publications in peer-reviewed journal, contributions to books, invited lectures, and presentations at scientific and professional meetings. Recent achievements include:

\*Editor, “*Genetic disorders of the Indian subcontinent*”, Springer-Kluwer, Netherlands

\*Editor, “*Genomics and Clinical Medicine*” (in press), Oxford University Press, New York

\*Editor-in-Chief, ‘*The HUGO Journal*’ (formerly *Genomic Medicine*), published by Springer,NL

\*Editor: “*Clinical cardiovascular genetics-principles and practice*”, in preparation, a new book commissioned by Oxford University Press, New York.

Co-author: “*Oxford Hand Book on Inherited Cardiovascular Disease*” in preparation

\*Editor: “*Genetic disorders among peoples from developing countries*”, a new book commissioned by Oxford University Press, New York.

***Teaching:*** Undergraduate and postgraduate teaching in paediatrics and clinical genetics to medical students, specialist registrars and senior nurses.

***Audit in Clinical Genetics:*** Participation in various clinical audit projects relevant to clinical genetics, including patient satisfaction, patient notes review, outcome of specialist neurofibromatosis clinic, molecular testing in Rett syndrome, and clinical and molecular diagnosis of Fabry disease.

***Personal skills:*** Enjoy working as a team member with effective communication skills; equipped with practical computer skills including effective use of the internet; special communication skills in dealing with patients and families from the Indian subcontinent.

***Management experience:*** Broad general experience in administration and management acquired through working and attendance at management training courses.

***Career aims:*** **Clinical and academic career in Medical Genetics**

## 2. Personal and professional details

<b>Full Name:</b>	Dhavendra Kumar
<b>Nationality</b>	British- U.K. Citizen
<b>Defence Organisation</b>	Medical Protection Society No. 281124
<b>General Medical Council</b>	Full Registration No: 2351409
<b>General Practice accreditation</b>	<b>Entry in the GMC-GP Register</b>
<b>Specialist accreditation</b>	<b>GMC Specialist Register entry-Clinical Genetics, April, 2006</b>  <b>Board certified 'Clinical geneticist'- 1990 [American Board of Medical Genetics]</b>
<b>Clinical skills</b>	I am a caring, hardworking, knowledgeable and highly motivated professional.
<b>Academic skills</b>	I have wide research and teaching experience in medical genetics
<b>Management skills</b>	I have leadership, organisational, management, inter-personal and communication skills. I enjoy team working. I am computer literate and regularly use internet and computer databases.
<b>Languages</b>	English and major Indian languages (Hindi, Punjabi and Urdu)
<b>Hobbies and interests</b>	A family oriented person, I appreciate quality time with my family. I enjoy playing golf and working out in the gym on a regular basis. My other leisurely interests include reading, writing and theatre. I also participate in community and charitable activities.
<b>Career aims</b>	<b>Clinical and academic career in Medical Genetics</b>

### 3. Awards, Honours and Prizes

#### 3.1 Awards and Honours

- Awarded a **Gold Medal** for the highest percentage of marks at the second professional MBBS examination (1971), which covered the second MBBS professional examination included pharmacology, pathology, forensic medicine, and social and preventive medicine.
- Awarded a **Gold Medal** for the first position at the final professional MBBS examination (1973): Final MBBS examination subjects included general medicine, general surgery, ophthalmology and obstetrics and gynaecology.
- Awarded a **Gold Medal** for the best successful candidate for M.D. in Paediatrics.

#### 3.2 Travel Fellowships

- **“The Guarantors of “Brain”** Travel scholarship in 1986 to visit the major Neurogenetics centres in the U.S.A.: A travel scholarship of £500 was awarded by the ‘Lord Brain Trust’ for my work on the clinical and genetic heterogeneity in hereditary spinocerebellar ataxia and Charcot-Marie-Tooth disease.
- Travel grant of £150 was awarded by the **Skeletal Dysplasia Group (UK)** to attend the First meeting of the US BONE DYSPLASIA (1993).
- Travel grant of £250 from the **UK Neurofibromatosis Association** to attend the 11<sup>th</sup> Clinical Care Symposium of the National Neurofibromatosis Foundation, 5<sup>th</sup> March 1995, Los Angeles, USA.
- Travel grant of £200 from the **UK Clinical Genetics Society** to attend the 2<sup>nd</sup> Joint Annual Clinical Genetics Conference of the March of Dimes Clinical Genetics meeting and the American College of Medical Genetics, 6-9 March 1995, LA
- Travel fellowship from **NIH, Bethesda, USA** to attend the Joint meeting of the March of Dimes Birth Defects and the American College of Medical Genetics, 6-9 March 1995, LA.
- Travel grant from **British Society of Human Genetics** to attend the Human Genome meeting (HGM2005) in Kyoto, Japan (18-21 April,05)

### 4. Education and Qualifications

#### 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 - December 1986

#### Qualifications

1. MB, BS            1973   King George's Medical College, University of Lucknow, India
2. MD                1977   University of Lucknow, India
3. DCH              1979   Royal College of Surgeons & Physicians in Ireland
4. M Med Sci        1987   University of Sheffield, UK  
(Medical Genetics)
5. MRCPi            1990   Royal College of Physicians in Ireland
- FRCPI            1992   Royal College of Physicians in Ireland
6. DipABMG        1990   American Board of Medical Genetics  
(Clinical Genetics)
7. MRCPCH        1996   Royal College of Paediatrics & Child Health (UK)
- FRCPCH         2006
8. PCME            2003   Staffordshire University, UK
9. FACMG          2003   Fellow: American College of Medical Genetics
10. FRCP            2008   Fellow   Royal   College   of   Physicians-London,  
England

## 5. Employment History

### 5.1 Present post in Clinical Genetics

**CONSULTANT IN CLINICAL GENETICS: 1 December 2006 to date**

**University Hospital of Wales, Cardiff and Vale University Health Board, Cardiff.**

This is the substantive Consultant post- major activities in this post include:

#### 1. Clinics

General –

University Hospital of Wales, Cardiff  
Ystrad Mynach Hospital, Ystrad Mynach  
Nevill Hall Hospital, Abergavany

Special cardiovascular genetics- University Hospital of Wales, Cardiff

2. **On-call**            Ward referrals- Neonatal unit; Children's ward  
Telephone enquiries- professional, patients and others

#### 3. Clinical meetings

Genetic counselors- review of referrals/ clinic appointments  
Consultants/SpRs- dysmorphology slides/cases discussion  
Clinical molecular genetics meetings  
Multi-disciplinary cardiac genetics meetings (Cardiff, Swansea, Gwent)  
Structural heart disease  
Arrhythmias

#### 4. Administration

Review of clinical referrals  
Clinic appointments

Database (SHIRE) management- outcomes etc.

**5. Management**

Consultants meeting  
Clinic planning and administration  
Secretarial supervision  
Clinical Governance issues  
Strategy planning discussions

6. **Service Developments-** Preparation of the business plan for organising and providing specialist cardiovascular genetic service across Wales:  
This is a new service development. It has the approval from Cardiac Services Network for Wales. Full implementation would require developing a mutli-disciplinary All Wales Cardiac Genetics Team.

**7. Postgraduate meetings**

Journal club- general and cancer genetics, monthly  
Weekly clinical case meetings, Thursdays 11-12  
Weekly Lunch-time meetings, Thursdays 1-2  
Weekly Academic meetings, Fridays 1-2  
Postgraduate grand round, UHW, Wednesdays 1-2  
Other postgraduate seminars, for example at Henry Wellcome Centre and Cardiff University

**8. Audit**

Clinic outcome- Patient satisfaction survey  
Fabry disease- clinical and molecular management  
Participation in other SWOB audit programs

**9. Teaching**

Specialist Registrars  
Genetic counsellors  
MBChB and Intercalated BSc

**8. Research and Academic**

- Book project- '*Genomics and Clinical Medicine*' Publishers Oxford University Press, New York (In press)
- Editor-in Chief- new journal - '*Genomic Medicine*' to be published by Springer-Verlag publishing group.

**9. Publications**

Book- 'Genetic disorders of the Indian subcontinent', published by Kluwer-Springer, October 2004  
Book- 'Genomics and Clinical Medicine' Oxford University Press, November 2007  
New Journal-'Genomic Medicine', Springer, The Netherlands

**10. Attendance/participation at professional meetings/conferences**

- Manchester Birth defects meeting 9-12 November, 2005
- London dysmorphology group meetings Feb 2005; June 2005
- Human Genome Meeting (HGM2005, Kyoto, Japan & HGM2008, Hyderabad, India)
- American Society of Human Genetics- Salt Lake City, 2005 & San Diego, 2007)
- British Society of Human Genetics, 2005-2009

**11. Organising professional meetings/courses/workshop**

- Two day workshop at Millennium Centre, Cardiff '*Genetic disorders and people from the Indian subcontinent-Challenges for clinical services in the NHS*' 22-23 November 2005
- Symposium on '*Clinical Cardiovascular Genetics*', 22-23 November, 2007 and 23-24 November 2009, Cardiff.
- Two day seminar on '*Multi-Disciplinary Care for Inherited Heart Disease*', 30-31 January 2009, Cardiff.
- Two day International symposium on '*Clinical Medicine in the Genome Era*', 26-27 February 2009, Cardiff.

## 12. Appraisal

Educational with Dr. Sally Davies, dated 2 December 2005

Clinical with Dr. Annie Procter, 2005, 2006, 2007, 2008

## 5.2 Previous posts in Clinical Genetics

### 5.2.1 Consultant in Clinical Genetics,

**Institute of Medical Genetics, University Hospital for Wales, Cardiff, UK**

**1/11/2004 to 31/12/2006**

**Including Clinical Lecturer/Hon. Consultant, Cardiff University, 1 November 2005 to 31 March 2006.**

The clinical commitment of this post included general and cancer genetics. Special interests include dysmorphology, metabolic diseases and cardiovascular genetics. In addition, this post provides opportunities for teaching, research and management.

### 5.2.2 Specialist Registrar in Clinical Genetics

**1/11/2003 to 31/10/2004**

**Oxford Regional Genetics Service, The Churchill Hospital, Oxford**

The main components of this post included improving upon generic skills, carrying out supervised clinical work, obtaining relevant laboratory experience, acquiring managerial and administrative skills and experience in clinical audit on one of the selected topics. The clinical work involved general genetics, special genetic clinics (cancer, neurofibromatosis, VHL, craniofacial and neuro-genetics) and attending the ward referrals. In addition, this post provided opportunities of teaching the junior specialist registrars and trainee genetic counsellors.

### 5.2.3 Consultant in Clinical Genetics

**Oxford Regional Genetics Service, The Churchill Hospital, Oxford**

**03/03/2003 to 31/10/2003**

This post involved participation in the delivery of clinical genetics service through general genetic and specialist genetic clinics to the Oxford region serving a population of approximately 2.5 million. In addition, the post offered an opportunity to teach medical students and trainees in clinical genetics, as well as acquiring administrative and management experience.

### 5.2.4 Associate Specialist in Medical Genetics at North Trent Regional Genetics Service, Sheffield Children's Hospital, Western Bank, Sheffield.

*Part time (5 sessions per week) 8 June 1988 to 31 July 1990*

***Whole time from 1 August 1990 to 2 March 2003***

***Consultants: Dr. Oliver Quarrell and Dr. J. Cooke***

The clinical commitment included a wide range of general and special genetic clinics, prenatal diagnosis, neonatal and paediatric ward referrals. This post helped to develop special interests in dysmorphology, metabolic diseases, neurogenetics, cancer genetics and neurofibromatosis. In addition, the post offered opportunities for teaching, administrative and managerial duties.

**5.2.5 Hon. Clinical Tutor in Medical Genetics, Division of Genomic Medicine,  
The Medical School, University of Sheffield, Sheffield.  
*From 1 November 1990 to 1 June 1998***

This honorary position offered opportunities to participate in teaching and research.

**5.2.6 Consultant in Clinical Genetics, Nottingham Clinical Genetics Service, City  
Hospital Nottingham, Nottingham:  
*11 August 1997 to 31 October 1997***

Fixed term Consultant post serving the Nottingham clinical genetics service; estimated population about 2 million people across the Mid-Trent NHS region.  
Lead Consultant: Prof. Sandy Raeburn.

**5.2.7 Consultant Clinical Geneticist, Oxford Radcliffe Hospitals NHS Trust,  
Oxford. *1 May 1997 to 31 July 1997***

This 3 month locum Consultant appointment was to provide the Oxford Regional Clinical Genetics Service to cover leave for one of the Consultants.  
Lead Consultant: Dr. Cyril Chapman

**5.2.8 Consultant Clinical Geneticist, Royal Devon and Exeter Hospital, Exeter**

Two appointments: *1-30 November 1992 and 1 May to 31 July 1993.*

This consultant post was to deliver a clinical genetics service to the South-West region based at the Royal Devon and Exeter Hospital serving a population of over 1.5 million. In addition to providing the clinical genetics service, I participated in discussions over future planning and management of the service. Such plans included expansion of Genetics Clinics, the provision of pre- and post- genetic counselling with the help of the Genetic Nurse Specialists, a centralised secretarial service, computerisation of records, and setting up and maintenance of the Regional Genetic Registers.  
Lead Consultant: Dr. Peter Lunt, Bristol Children's Hospital, Bristol

**5.3 Previous posts in other specialties**

**5.3.1 Clinical Medical Officer (Child Health),  
Community Child Health, Sheffield Health Authority, Sheffield.  
*Whole time: 1 Feb 1980 to 31 July 1980*  
*Part time (7 sessions per week): 1 Aug. 1980 to 31st October 1980*  
*Part time (5 sessions per week): 1 Nov. 1980 to 31 July 1990***

This post provided me with ample opportunity to work within a team caring for pre-school and school children. The duties included developmental assessment, advising parents and teachers on common childhood health problems, planning and carrying out immunisations, taking part in health education activities, teaching medical and nursing trainees, writing medical advice on education and social aspects for children with special needs, and dealing with some administration work.

**5.3.2 Senior House Officer in Psychiatry**

North Tees General Hospital, Stockton on Tees, Cleveland:

*1 August 1979 to 31 January 1980;*

Six months training post in general psychiatry.

Supervising Consultant- Dr. M. Sutherland

**5.3.3 Senior House Officer in Paediatrics, Leicester Royal Infirmary, Leicester: 1 July 1979 to 31 July 1979**

**5.3.4 Consultant Paediatrician (Private practice), Moradabad, UP.**

*1 March 1979 to 30 May 1979.*

**5.3.5 Hospital training appointments in Paediatrics:**

Approved post graduate training in Paediatrics included various hospital appointments based at the Department of Paediatrics, King George's Medical College and Gandhi Memorial and Associated Hospitals, Lucknow, U.P., India. This programme included carrying out wide clinical duties and studying for an MD by thesis.

<i>Chief Resident in Paediatrics</i>	<i>1 March 1978 to 28 Feb 1979</i>
<i>Hon. Research Fellow</i>	<i>1 Jan 1978 to 29 Feb 1978</i>
<i>Senior Resident in Paediatrics</i>	<i>1 Jan 1977 to 31 Dec 1977</i>
<i>Junior Resident in Paediatrics</i>	<i>1 Jan 1976 to 31 Dec 1976</i>
<i>House Officer in Paediatrics</i>	<i>1 Sept 1974 to 31 Dec 1974</i>
	<i>1 Jan 1975 to 31 Dec 1975</i>

**5.3.6 Pre-Registration Houseman, District Hospital, Moradabad, UP**

*1 Sept. 1973 to 31 Aug. 1974.*

This one-year compulsory pre-registration internship (house officer) included 3 monthly rotations between general medicine, general surgery, obstetrics and gynaecology and public/ community health.

## **6. Clinical genetic experience and training**

After completing the general professional and specialist training in paediatrics, I joined medical genetics as a part-time Clinical Assistant. This post evolved alongside the part-time post in Community Child Health.

My paediatric background helped me to develop an interest in paediatric genetic disorders particularly in developing skills and special interest in metabolic diseases, paediatric neurological diseases and dysmorphology. I participated in providing the clinical genetic service for diagnosis and genetic assessment on malformation syndromes. I developed close links with the biochemical diagnostic and neonatal screening laboratories, and acquired experience and understanding in various paediatric metabolic conditions. As

a member of the metabolic team at the Sheffield Children's Hospital, I gained useful practical experience of working within the multi-disciplinary metabolic team.

During my tenure as Clinical Assistant and subsequently as Associate Specialist in medical genetics, I gained extensive experience in general genetics, genetic counselling and dysmorphology, and developed a special interest in neurogenetics. I also gained laboratory experience in cytogenetics and molecular genetics. Apart from the clinical commitment in general genetics, I was involved with various patient/family support groups, management and administration and organising the regional genetics meetings in the Trent NHS region.

My research project on inherited cerebellar ataxias helped me to develop a special interest in neurogenetic problems. I established a multi-disciplinary specialist team in Sheffield for managing patients and families with inherited spinocerebellar ataxias. My other special interests in neurogenetics include neurofibromatosis and Charcot-Marie-Tooth disease.

In addition, I was actively involved in developing the Cancer Genetics service in Sheffield for the North Trent region. This service is now fully established and includes regular cancer genetic clinics and has close links with oncologists, surgeons and supported by the local diagnostic molecular genetic laboratory.

I spent one-year as Specialist Registrar [LAT] in Clinical Genetics at The Oxford Clinical Genetics department. This post provided opportunity to undergo formal training. This helped me in improving generic skills, carrying out the supervised clinical work, obtain the relevant laboratory experience, acquire managerial and administrative experience and conduct an audit on Rett syndrome. Clinical work included general genetics, cancer genetics, cardiac genetics and ward referrals. In addition, I also participated in the teaching of specialist registrars and trainee genetic counsellors. Experience and training in this post was accepted for accreditation by the General Medical Council (UK) and entry on the Specialist Register in Clinical Genetics.

My present post in University Hospital of Wales, Cardiff as Consultant in Clinical Genetics offers excellent clinical and educational opportunities in furtherance to my professional career development [see [Appendix-A](#)]. I have kept myself updated on recent developments and new research in genetics and related disciplines through professional development and participating in continued medical education. I regularly attend various workshops and courses and actively participate in professional meetings and conferences. I am enrolled with the Continued Professional Development [CPD] plan of the Royal College of Physicians, London.

My aim is to develop further clinical and academic career in Clinical Genetics with a special interest in cardiovascular genetics and genomic medicine.

#### **Special interest and career development in cardiovascular genetics:**

I have a special interest in cardiovascular genetics and intend to develop as my future career aim. This interest generated from my time spent in Oxford Regional Medical Genetics unit. I was involved with the Oxford Marfan Clinic, which is a multi-disciplinary NHS service activity co-ordinated by Prof. Wordsworth and jointly staffed with clinical geneticist team, cardiologist, rheumatologist and an ophthalmologist. In addition, I have closely watched the functioning of the Oxford Molecular Genetic Service for inherited cardiomyopathies and arrhythmias. I worked closely with Dr. Edward Blair, who runs the Cardiac Genetics clinic with Prof. Hugh Watkins. During my stay in Oxford I worked on a large family with primary pulmonary hypertension and helped the 'at-risk' family members in predictive genetic testing and specialist clinical management.

During my tenure in Cardiff, I have been associated with Dr. Sally Davies who is the

Lead Clinical Geneticist for the cardiac genetics. I have conducted few cardiac genetic clinics independently. I have understanding of various protocols that are commonly used in dealing with inherited heart disease. I work closely with Mrs. Mary Nicol, who is the designated cardiac genetic counsellor. In addition, I have established links with Arrhythmia (Dr. Peter O'Callghan) and Cardiomyopathy (Dr. Zaheer Yousef) clinics. There is overwhelming support for developing a multi-disciplinary cardiac genetics team in Cardiff.

I have established close links with the Inherited Cardiovascular Service unit at the The Heart Hospital of the University College London. This has helped me in expanding my experience and understanding and balancing the service provision and meeting with challenges of academic research. I have worked with Dr. Perry Elliot (Reader and Hon. Consultant) and developed a comprehensive text book on principles and practice of cardiovascular genetics (published by Oxford University Press).

I carried out a clinical and genetic audit in Fabry disease, a multi-system X-linked metabolic disorder with major cardiac manifestations including sudden cardiac death. The outcome of this audit has been appreciated by the metabolic and public health units. This has also been used in discussions with the Health Commission for Wales in securing funding for enzyme replacement therapy in Fabry disease. Part of this audit data was used in a poster presentation at the annual conference of the British Society of Human Genetics (18-20 September, 2006).

My commitment to team working has led me to consultations on the structure and organisation of the *All Wales Cardiovascular Genetics Service*. I am also co-opted with the Cardiovascular Interdisciplinary Research Group of the School of Medicine, Cardiff University and the Wales Cardiovascular Research Network. My role is to lead clinical cardiovascular genetics research within these two major research groups.

I am keen on promoting and strengthening clinical cardiovascular genetics and have organised two consecutive International symposia to review developments and challenges of inherited cardiovascular conditions.

## **7. Teaching experience**

### **7.1 Undergraduate students**

I participated in student tutorials during the paediatric, community medicine and obstetrics and gynaecology electives. In addition, I have also helped the MB ChB students during the elective attachment and preparation of the dissertation on a topic related to clinical genetics. In my current post I am one of the Clinical Tutors for Cardiff University for the third year MB students. In addition, I am also assigned teaching responsibility to medical students (Graduate entry programme) of the Swansea University. In addition I also teach medical genetics to BSc intercalated programme within the MB course of Cardiff University.

### **7.2 Post graduate students and trainees**

Participate in the post-graduate teaching and training in Clinical Genetics and Genetic Counselling:

1. Specialist Registrars in Clinical Genetics
2. Specialist trainees in other specialties- Paediatrics, Obstetrics & Gynaecology, Fetal Medicine.
3. M.Sc Genetic Counselling, Cardiff University

### 7.3 Post graduate courses/workshop:

1. Course for General Practitioners: Since 1990, I have organised educationally **approved courses for General Practitioners**: These courses were aimed at offering a broad view of the practice of Medical Genetics as applied to the primary medical care.

2. I have organised a CME approved postgraduate course for specialist clinicians and nurses entitled **“Genetic predisposition for Human Cancer”**.

3. **Neurofibromatosis Workshop - October 1997**: This workshop was arranged with the help of the Neurofibromatosis Association and covered a range of problems and information on NF1 and NF2. It was attended by both medical and nursing personnel dealing with patients and families affected with neurofibromatosis.

4. **Genetics and Modern Medical Practice - April 1998**: This one-day symposium was organised to give medical practitioners from various backgrounds basic and applied insight into the practice of modern clinical genetics and its impact on fast changing trends in the practice of medicine.

5. **Community management of Huntington’s Disease - April 2001**: This half day educationally approved course was offered to general practitioners and other interested professionals to update them on Huntington’s disease with an aim to focus upon the concept of the community approach in management.

6. **First Asian Workshop on Genomics and Community Genetics, 12-22 April 2004**, held at the Peking Hong Kong University Hospital, Shenzhen, PR China.

I participated in this post-graduate workshop as a Faculty member. Fellows included students and geneticists from various Asian countries.

7. **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. Approved for 10 CPD credits.

8. **“Challenges of genetics and genomics for the Indian subcontinent”**, organised a pre-congress satellite symposium at the 11<sup>th</sup> International Congress of Human Genetics, 6-10 August, Brisbane, Australia.

9. **“Challenges of cardiovascular genetics”**- 22/23 November 2007, Cardiff. A two day educationally approved symposium to focus on recent developments, current practice and issues in delivering the cardiac genetic services in the UK.

10. **“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.

### 7.4 Faculty advisor for ‘Postgraduate Diploma in Medical Genetics’ and ‘Medical Genetics Workshops’ offered by the Vidyasagar Institute of Genetic Studies in Mumbai, India. Key features of this course are as follows:

**PROGRAM NAME: POST GRADUATE DIPLOMA IN MEDICAL GENETICS**

**TARGET GROUPS:** \*Medical practitioners in primary, secondary and tertiary care with a special interest in medical genetics

- \*Medical graduates with career interest in medical genetics
- \*Post graduate trainees in general medicine, pediatrics, obstetrics and gynecology, oncology and preventive medicine
- \*Postgraduate science students with career interests in laboratory genetics, biotechnology, molecular biology and bio-informatics

**ACADEMIC STANDARD:** Masters level course

**ACCREDITATION:** To be applied for- Indira Gandhi National University [IGNU]

**PROGRAM COMMENCES-** AUGUST 2007

**AIM** To learn the principles and practice of medical genetics in modern medicine

### **OBJECTIVES**

- Knowledge about basic principles of genetics
- Application of basic principles of genetics in understanding human diseases
- Clinical diagnosis of human genetic diseases
- Laboratory diagnosis of human genetic diseases
- To understand and be able to apply medical genetics knowledge in clinical medicine
- Genetic counselling
- Health and related issues in medical genetics
- Scope and applications of the human genome sequencing
- Applications of genome science and technology in future medicine and public health
- Understanding the ethical, legal and social issues in genetics and genomics

#### **7.5 Other teaching activities:**

- **Nursing and paramedical:** Teaching medical genetics to nursing and midwifery students (pre and post registration). This includes mental health nurses, general nursing students, and nurses in special nursing courses.
- **Lectures to Lay Organisations and Patient Support Groups:** I have assisted students studying for GCSE and 'A' levels with projects related to Genetics. I have given lectures to lay groups and Patient Support Groups, including Down syndrome, Motor Neurone disease, Friedreich's Ataxia, Charcot-Marie-Tooth disease, Neurofibromatosis, Fragile X syndrome and Prader Willi syndrome.

## **8. Audit and Clinical governance**

### **8.1 Audit in Clinical Genetics**

I have regularly participated in the audit. These include:

- Protocols in common genetic diseases- Fragile-X syndrome, neurofibromatosis, familial adenomatous polyposis coli [FAP], and familial breast/ovarian cancers. (Sheffield)
- Review of patient notes (Sheffield)
- Readability of patient letters (Sheffield)
- Patient satisfaction surveys (Sheffield)
- Outcome of special neurofibromatosis clinic. (Sheffield)
- Clinical and molecular diagnosis in Retts syndrome. (Oxford)
- Clinical and molecular diagnosis in Fabry disease (Cardiff)

## 8.2 Clinical governance

I am interested in '**clinical governance in clinical genetics**'. I have experience in '**risk management**' and '**untoward incident reporting**' relevant to clinical genetics.

## 9. Administration and Management

### 9.1 General administration:

- Clinic appointments and administration
- Organising postgraduate lectures/ seminars, journal club and departmental clinical meetings including Regional Genetics meetings involving the three Genetic Centres of the Trent NHS Region (Sheffield, Leicester and Nottingham).
- Managing the departmental library
- Providing support to routine administrative tasks
- As a member of the Department management team, I regularly contribute in planning and management decisions concerning staff recruitment and retention, maintenance and purchase of new equipment and issues related to the future development of the Clinical Genetics services.

### 9.2 Management

I have regularly participated in the management the activities that included serving on sub-committees for building and library management. In the present post, I serve on the '**clinical unit**' and '**information technology**' sub-committees of the department. In addition, I have kept myself informed about management skills and changes through attendance at various management training courses and seminars specifically on clinical governance, appraisal and revalidation:

- **Management training course for Senior Registrar:** One week residential course for management training aimed at Senior Registrars and Career grade doctors - August 1994, organised by the Trent Institute of Health Management and Administration.
- **Nuts and Bolts of Health service management**', 7-8 July 2004, organised by Oxford Postgraduate medical and dental education (PGMDE).

### 9.3 Developing cardiovascular genetic service for Wales

**I am the Lead Consultant Clinical Geneticist for Cardiovascular Genetics for Wales. In this role I am actively involved in the following clinical, academic and professional activities. I have organised a multi-disciplinary team for developing an integrated [‘The Wales Centre for Inherited Cardiovascular Diseases’](#). The centre will have the following key roles and commitments:**

- Preparing the clinical service plan and protocols
- Preparation of clinical care pathways for clinical referrals, molecular genetic testing and managing family members with a history of ‘sudden cardiac death’.
- Organizing 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.
- Organising the All Wales Cardiovascular Genetics Interest Group
- Contacts with Wales section of major patient support groups- for SADS, CMA, CRY and AA
- Organising the International symposium on clinical cardiovascular genetics in Cardiff (22-23 Nov. 2007)
- Active participation in the UK Cardiac Genetics Group of BSHG
- Member of the UK Cardiac Genetics Advisory Group of the Public Health Genomic Foundation, Cambridge
- Active participation in service development initiatives of BHF for cardiac genetic service in Wales

In addition to the above roles/ activities, I am also working to develop a new text book and a hand book on clinical cardiovascular genetics jointly with Dr. Perry Elliott of the Heart Hospital of University College London (see section below ‘New Books’).

## 10. Research and Publications

### 10.1 Research

During my tenure in Clinical Genetics I have carried out number of independent and joint research projects. The outcome of my research is reflected in publications in peer reviewed journals and presentations at professional and scientific meetings

#### 10.1.1 Research grants and awards

- 1 Council of Scientific and Industrial Research – India 1978-1981 **“Cytogenetic studies in Childhood Diseases”** at the Department of Paediatrics, K.G.Medical College, Lucknow, India.
- 2 Trent Regional NHS Locally Organised Research Fund: 1982-1986 **“Clinical and Genetic study in hereditary spinocerebellar ataxia with special reference to genetic linkage with HLA”**
- 3 Children’s Hospital Research in Sheffield Fund [CHRIS Fund]: 1995-1996 **“Application of molecular cytogenetics and DNA methods in the**

**diagnosis of Charcot-Marie-Tooth disease.”**

#### **10.1.2 Recent research projects:**

- **A clinical and genetic study of abnormal Mullerian duct development and associated multiple congenital anomalies**

This study is aimed at systematic collection of information on syndromic association of abnormal Mullerian development with cranio-facial and skeletal abnormalities. This study has been approved by the local Ethics Committee.

- **Kaufman-McKusick syndrome**

Gene mapping and molecular studies in this autosomal recessive multiple malformation syndrome and phenotypically related Bardet-Beidl syndrome -in collaboration with Dr. Leslie Biesecker, NIH, Bethesda, USA.

- **AD Anonychia-Brachydactyly syndrome**

Gene mapping for type b brachydactyly in collaboration with Prof. Andrew Wilkie, WIMM, University of Oxford.

#### **10.1.3 Current Research Interests**

My recent research interests have largely focussed on collecting information to compile books, two of these books are published and two are progressing as per schedule. These book projects required extensive research to select relevant subjects and appropriate authors.

My current research interests are in cardiovascular genetics. The following projects are in under discussion and expected to commence soon:

1. **‘Clinical and genetic study in familial thoracic and abdominal aneurysm’**. This study is currently in the planning stage reflecting my special interest in cardiovascular genetics. The study aims to collect clinical and genetic data on heterogeneity in non-syndromal form of aortic aneurysms.
2. **‘A study on gene expression signatures in cardiovascular medicine’**. This study aims to apply gene expression array technology in diagnosing and assessing the clinical progress in chronic heart failure specifically related to dilated cardiomyopathy. Funding application will be made with the appropriate ethical approval.
3. **‘A study on genetic heterogeneity in Wolfe-Parkinson-White syndrome’**- this study aims to focus on heterogeneity in Wolfe-Parkinson-White (WPW) syndrome. Apart from collecting clinical data, the study will investigate patients and family members using molecular genetic methods. It is possible the study might lead to identifying new aetiological genetic factors.
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.

#### 10.1.4 Book projects:

- ***‘Genomics and Clinical Medicine’***, Series- Oxford Monographs in Medical Genetics, *Oxford University Press, New York- published January 2008.*

This multi-author book provides comprehensive information on general aspects of genomics and applications of genome-based science and technology in clinical medicine and population health.

In addition to planning, selecting various authors/contributors and editing the book, I have also written or co-authored several chapters (see below section 10.2.1).

- ***‘Clinical Cardiovascular Genetics- Principles and Practice’***, Series- *Oxford Monographs in Medical Genetics, published by Oxford University Press, New York. Editors: Dhavendra Kumar and Perry Elliot*

This new multi-author edited textbook is aimed at providing a comprehensive account on all aspects of principles and practice of inherited cardiovascular disease. The book consists of contributions from several internationally renowned experts on individual diseases or group of disorders.

- ***“Genomic variation and Genetic disorders of developing countries”*** – **to be published by Oxford University Press, New York.**

This is new book project which aims at providing a detailed and comprehensive account on the physical and genomic structure of peoples from developing countries spanning about two-third of the World. The book will include information on data on genetic epidemiology and common genetic disorders. This will be first book of its kind. Several eminent geneticists working in different parts of the World will lead respective sections exclusively assigned to one of the major geo-political regions such as China, Indian subcontinent, Africa, South America etc. My role is to lead this project as the Chief Editor.

- ***“Oxford Handbook on Inherited Cardiovascular Disease”***, Co-Editor, commissioned by Oxford University Press.

This book aims at providing concise and practical information on inherited cardiovascular conditions focussing on genetic aspects, clinical management and surveillance of the ‘at risk’ family members.

#### 10.1.5 New Journal **“GENOMIC MEDICINE”**

This is a new ‘peer-reviewed’ international bio-medical journal recently launched by Springer from The Netherlands. I am appointed as ***Editor-in-Chief*** for five years. The journal is supported by several eminent persons from human genetics, genomics, bioinformatics, clinical medicine and population health. The first issue of the journal was published in September

2007. The journal is published both on-line ( [www.springer.com/11568](http://www.springer.com/11568)) and in print (quarterly). The manuscript submission and management for the journal is wholly managed on-line using [www.editorialmanager.com/genomed](http://www.editorialmanager.com/genomed). So far 4 issues in the first volume and two issues of the second volume are published online and as well as in the print format. The journal is now indexed in PubMed and can be accessed free on PubMed Central. This journal is now the official journal of the Human Genome Organisation (HUGO) appropriately named '**The HUGO Journal**'.

## 10.2 Publications

### 10.2.1 Chapters in books

**“Genetic Disorders of the Indian Subcontinent”** - I have edited this reference book to cover various aspects of genetic variation and genetic diseases among people from the Indian subcontinent. Several authors from India, UK and other countries have contributed various chapters. [Publishers- Kluwer-Springer Academic, The Netherlands, 2004].

In addition to editing the book, I wrote the following chapters:

- **Kumar D:** ‘The Indian subcontinent and genetic disorders: An introduction’.
- Ishwar C Verma and **Dhavendra Kumar:** ‘Epidemiology of genetic diseases in the Indian Subcontinent’
- **Kumar D:** ‘Congenital developmental anomalies’.
- **Dhavendra Kumar,** Michael Patton, Ajith Kumar: ‘Genetic neurological and psychiatric disorders’.
- Sanjay S Madan, James A Fernandes and **Dhavendra Kumar:** ‘Inherited skeletal dysplasias and collagen diseases’.
- **Dhavendra Kumar,** Carani B Sanjeevi, V. Radha and V. Mohan: ‘Diabetes mellitus and related disorders’.
- **Kumar D:** ‘Genetic counselling for families from the Indian subcontinent’.
- **Kumar D:** ‘The genetics of cancer-the Indian subcontinent perspective’

### **“Genomics and Clinical Medicine”**

This is a multi-author edited text book largely focussing on applications of genome science and technology in clinical medicine and population/public health. The book includes sections on general genomics, clinical genomics and health genomics. Several leading experts have contributed chapters. My own contribution in the book includes the following chapters/ sections. The book offered my excellent opportunity for editing the book.

1. Preface
2. Genomic perspectives of human development
3. A taxonomy for human disease- genetics and genomics approaches
4. 4. Clinical medicine in the genomic era- an introduction
5. Neuropsychiatric diseases II: Affective disorders
6. Genomics and global health (Co-author, Prof. David Weathrall)
7. Glossary (compiled from various sources)

### **“Principles and Practice of Clinical Cardiovascular Genetics”**

This is a joint project with Dr. Perry Elliott, Reader/Consultant in Inherited Heart Disease at the Heart Hospital, University College Hospital, London. Apart from planning and

editing, I have also co-authored the following chapters in this book:

- Challenges of clinical cardiovascular genetics- an Introduction
- The classification of inherited cardiovascular conditions
- Thoracic and abdominal aortic aneurysms

### 10.2.2 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”

### 10.2.3 Chapters in other books

- **Kumar D, Clark J W, Blank C E:** Acrocephalosyndactyly and fragile site 12q13 segregating in the same family. In **‘Recent Trends in Medical Genetics’**(Eds.K.M.Marumuthu, P.M. Gopinath) Pergaman Press, Oxford1986
- **Kumar D:** Grebe Syndrome. In **“BirthDefects Encyclopaedia”**. Ed. Mary Louise Bruyse. Alan R. Liss Inc.New York, 1990.
- **Kumar D:** Moebius syndrome In **“Congenital Malformations”** Eds.D.Donnai and R.Winter, Chapman and Hall, London, 1995

### 10.2.4 Publications in peer reviewed journals

#### 10.2.4.1 Papers

1. **Kumar D et al.** Cerebrospinal fluid cations in convulsive disorders in children; Indian Pediatrics 1980, 17, 765.
2. **Kumar D et al.** Plasma and CSF electrolytes in Tuberculous Meningitis: A concurrent study. Indian Pediatrics 1981 18: 473-477
3. Gray R G F, **Kumar D** , Whitfield A E, Glycogen Phosphorylase b Kinase deficiency in three siblings. J.Inherited Metabolic Disease 1983, 6, 107
4. **Kumar D**, Curtis D, and Blank C E, Grebe Chondrodysplasia and Brachydactyly in a family. Clinical Genetics 1984, 25(1), 68-72
5. Blank C E, **Kumar D**, Johnson M. Multivitamins and prevention of Neural Tube Defects: A need for detailed counselling. Lancet 1984, February 4, 291
6. **Kumar D** Genetics of Indian Childhood Cirrhosis. Tropical and Geographic Medicine 1984, 36, 313-316
7. Barnes ICS, **Kumar D**, Bell R J M. A child with recombinant of chromosome 8 inherited from her carrier mother. Journal Medical Genetics, 1985, 22(1), 67-70
8. Gray RGF and **Kumar D**. Mitochondrial Malic enzyme in Friedreich’s Ataxia – Failure to demonstrate reduced activity in cultured fibroblasts. J.NeurNeurosurgPsych.Jan1985, 48(1),70-74.
9. **Kumar D**, Blank C E, Griffiths B L. Cornelia de Lange syndrome in several members of the same family. Journal Med Genet 1985, 22:296-300
10. **Kumar D**, Blank C E, Gelsthorpe K. Hereditary Cerebellar Ataxia and Genetic Linkage with HLA. Human Genetics 1986, 72: 327-332
11. **Kumar D** and Blank C E. Hereditary Spinocerebellar Ataxia and genetic linkage with HLA. Journal Medical Genetics, 1984, 21, 301
12. **Kumar D**, Clark J W, Blank C E, Patton M A. A family with Craniofrontonasal

- dysplasia and fragile site 12q13 segregating independently. Clin Genetics, 1986, 29(6): 530-537
13. **Kumar D**, Levick R K. Autosomal dominant onychodystrophy and anonychia and brachydactyly type 'B' with ectrodactyly. Clin Genetics, 1986, 30: 219-225.
  14. **Kumar D**, Heath P R and Blank C E. Clinical manifestations of trisomy 5q J.Medical Genetics, 1987, 24(3): 180-184
  15. **Kumar D**, Papiha S S, Gelsthorpe K. Cell mediated immunological status and association of genetic markers in Hereditary Cerebellar Ataxia. Disease Markers 1987, 5: 31-41
  16. **Kumar D**. Genetic aspects of congenital cerebellar ataxia: Ind. J. Pediatrics 1986, 53: 761-773
  17. **Kumar D**, Timperley W R. Neurilemmomatosis and Neurifibromatosis Syndrome Ind. Journal Pediatrics 1985, 52:523-526
  18. **Kumar D**, Timperley W. Late onset sporadic Cerebellar Ataxia: The clinical, genetic and pathological aspects of some observations on a series of ten patients. Acta Neurol. Scand. 1988 77:181-186.
  19. Hurst J, Mankiewicz M, **Kumar D**, Winter R M. Hirschsprung's disease, microcephaly and Iris coloboma. A new syndrome of defective neuronal migration Journal Med. Genetics, 1988,25: 494-500.
  20. **Kumar D** Syndrome of the Month: Moebius syndrome J.Med Genet 1990, 27:122-126
  21. Nour S, **Kumar D**, Dickson J A. Anorectal malformations with sacral bony abnormalities. Archives of Disease in Childhood, 1989, 64: 1618-1620
  22. **Kumar D** Late onset Spinocerebellar Ataxia of possible X-linked recessive inheritance Brain dysfunction, 1989,2(5): 268-272
  23. **Kumar D**, Blank C E. Early onset Spinocerebellar Ataxia with retained tendon reflexes: an autosomal recessive disease distinct from Friedreich's Ataxia. Indian Pediatrics, 1989, 26: 1014-1019
  24. Priestley L, **Kumar D**, Sykes B Amplification of COL2A1 variable region used for segregation analysis in a family with the Stickler syndrome Hum Genet, 1990,85: 525-526
  25. **Kumar D**, Mueller RF, Duggan MB, Brocklebank T. Familial aplasia/ hypoplasia of pelvis, femur, and ulna with abnormal digits in an inbred Muslim family: an example of autosomal recessive disorder with overlapping features of syndromes of Fuhrmann, Al-Awadi, and Raas-Rothschild. Am. J.Medical Genetics 1997; 70:107-113
  26. **Kumar D**, Garcia-Minaur S, Quarrell OWJ, Landa G and Garazar C. A distinct syndrome of familial cleft lip and palate with prominent eyes and characteristic facies in two unrelated families. Clinical Dysmorphology, 1996;5:295-301.
  27. **Kumar D**, Moss G, Primhak R, Coombs R: Congenital renal tubular dysplasia and skull ossification defects similar to teratogenic effects of the angiotensin-1 converting enzyme [ACE] inhibitors J Med Genet 1997;34:541-545
  28. **Kumar D**, Masel J A new multiple malformation syndrome of Mullerian Dysgenesis, facial hypoplasia, bilateral limb deformity brachydactyly, spinal stenosis and scoliosis. Clinical Genetics July 1997; 52: 30-36.
  29. **Kumar D**, Rittey C, Cameron A, Variend S: A recognizable inherited syndrome of progressive central nervous system degeneration and intra cranial calcification with overlapping features of Aicardi-Goutieres syndrome. Am J Medical Genetics 1998;75:508-515.
  30. **Kumar D**, Primhak, R, Kumar, A: Variable phenotype in Kaufman-McKusick syndrome: Report of Inbred Muslim Family and Review of Literature. Clinical

- Dysmorphology 1998;7:163-170.
31. **Kumar D:** A case of lateral facial clefts with Fallot's tetralogy, duodenal stenosis and intestinal malrotation- a new multiple congenital anomaly syndrome? *Clinical Dysmorphology* 1999; 8(1): 19-22.
  32. William Reardon, Anne Smith, John W Honour, Peter Hindmarsh, Debipriya Das, GillRumsby, Isabelle Nelson, Sue Malcom, Lesley Ades, David Sillence, **Dhavendra Kumar**, Celia DeLozier-Blanchet, Shane McKee, Thaddeus Kelly, Wallace L McKeehan, Michael Baraitser, Robin M Winter: Evidence for digenic inheritance in some cases of Antley-Bixler syndrome? *J Med Genet* 2000, 37(1):26-32.
  33. Bhola RM, Horne GV, Squirrell DM, Chan TK, **Kumar D:** Autosomal dominant congenital superior oblique palsy. *Eye* 2001, 15: 479-484
  34. Slovtinek AM, Searby C, Al-Gazali L, Hennekam RC, Schrandt Stumpel C, Orcana-Losa M, Pardo-Reoyo S, Canani A, **Kumar D**, Capellini Q, Neri G, Zackai E, Biesecker LG: Mutation analysis of the MKKS gene in Mckusick-Kaufman syndrome and selected Bardet-Biedl syndrome patients. *Human Genetics*, 2002, 110(6):561-7.
  35. Manir Ali, Lorna J Hight, Didier Lacombe, Cyril Goizet, Mary D King, Uta Tacke, Marjo S van der Knaap, Lieven Lagae, Chris Rittey, Han G Brunner, Hans von Bokhoven, Ben Hamel, Yvette A Oade, A Sanchis, I Desguerre, D Cau, N Mathieu, M L Moutard, Pierre Lebon, **D. Kumar**, Andrew P Jackson, and Yanick Joseph Crow. **A second locus for Aicardi-Goutières syndrome at chromosome 13q14-21.** *J Med Genet* (Published Online First: 20 May 2005.) 2006, 43(5):444-450.
  36. Woods CG, Stricker S, Seemann P, Stern R, Cox J, Sherridan E, Roberts E, Springell K, Scott S, Karbani G, Sharif SM, Toomes C, Bond J, **Kumar D**, Al-Gazali L, Mundlos S. Mutations in WNT7A cause a range of limb malformations, including Fuhrmann syndrome and Al-Awadi/Raas-Rothschild/Schinzel phocomelia syndrome. *Am J Hum Genet.* 2006 79(2):402-8.
  37. **Dhavendra Kumar.** Genomic medicine: a new frontier of medicine in the twenty first century. *Genomic Med.* 2007; 1:3-7.
  38. **Dhavendra Kumar.** Genome Mirror-2006: 1) The human variome project 2) The nobel worthiness of RNA interference 3) Metabolomics and beyond. *Genomic Med.* 2007; 1:87-90.
  39. **Dhavendra Kumar.** From evidence based medicine to genomic medicine. *Genomic Med.* Published on-line doi. 10.1007/s11568-007-9013-6; *Genomic Med.* 2007; 1: 95-104
  40. **Dhavendra Kumar.** Genome Mirror-2007: Published on-line *Genomic Med.* 28 Dec. 2007. doi 10.1007/s11568-007-9013-6; *Genomic Med.* 2007; 1:147-148.

#### 10.2.4.2 Book reviews

1. **Kumar D:** "Genetics and You" by John F Jackson. *Am J Medical Genetics* 1997 73:235.
2. **Kumar D:** "Genetic Diseases in Arab Populations" Farag and Teebi, Oxford University Press, New York, 1996. *Am J Med Genetics* 1998 77:251-252.
3. **Kumar D:** "Genetic Counseling in the Dawn of the 21<sup>st</sup> Century" Eds. Bartsocas and Beighten. *Am J Med Genetics* 2000; 94(2):177
4. **Kumar D:** "Neurogenetics" Ed. S Pulst, Oxford University Press *Am J Med Genetics* 2001 104(1):85-85
5. **Kumar D:** "Molecular mechanisms of neuro-degenerative disorders". *Am J Med Genetics*, 2002

### 11. Presentations at professional meetings

1982

- Segregation of characteristic facies and pigmentary changes in a family with deafness: Waardenburg's syndrome. Clinical Genetics Society meeting

Glasgow, April 1982,

**1983**

- Familial atypical acrocephalosyndactyly and association of a fragile site on long arm of no.12 chromosome (fra12q1.3). Clinical Genetics Society meeting, November 1983, Guy's Hospital, London.
- Spinocerebellar Ataxia and genetic linkage with HLA. Clinical Genetics Society meeting, Salford, Sept. 1983.
- HLA typing in hereditary spinocerebellar Ataxia. XV International Congress of Genetics, New Delhi, Dec. 1993.
- Autosomal fragile site at 2q1.3 and acrocephalosyndactyly. Symposium on Recent Trends in Medical Genetics, Madras, 1983.

**1984**

- Genetic aspects of Spinocerebellar Ataxia. International College of Pediatrics Conference. Washington, DC, USA, March 1984.

**1986**

- Phenotypic features of a possible trisomy 8p Clinical Genetics Society meeting, Cardiff, Sept. 1986.
- The Kauffman-McKusick Syndrome: A rare autosomal recessive disorder with hydrometrocolpos. Postaxial polydactyly, congenital heart disease and anomalies of the gastro-intestinal and genito-urinary tracts. Second Birth Defects Conference, Manchester, 1986

**1987**

- Genetic linkage studies in Charcot Marie Tooth disease. Clinical Genetics Society meeting, London, April, 1987.

**1988**

- Turcot Syndrome segregating as autosomal dominant trait. Clinical Genetics Society, Southampton, Sept. 1988.

**1992**

- Heterozygote expression in Kaufman-Mckusick syndrome. Manchester Birth Defects Conference, October, 1992.

**1993**

- A new syndrome of Mullerian dysgenesis, facial hypoplasia, bilateral forearm deformity, brachydactyly, spinal stenosis and scoliosis. Bone Dysplasia Society Meeting, Chicago, June 1993.

**1994**

- Monosomy 22q11, hypoparathyroidism, and phenotypic features of Noonan's syndrome. Spring Conference of the Clinical Genetics Society, London, 1994
- Phenotype of de novo partial 16p trisomy. Manchester Birth Defects meeting, October 1994 An autosomal recessive syndrome similar to the fetal effects of angiotensin-1 converting enzyme (ACE) inhibitors. Manchester Birth Defects meeting, October, 1994

**1995**

- Autosomal recessive syndrome of renal tubular dysgenesis similar to teratogenic effects of angiotensin-1 converting enzyme (ACE) inhibitors. 2<sup>nd</sup> Joint Clinical Genetics Conference of the March of Dimes and American College of Medical Genetics, 6-9 March, 1995, Los Angeles, USA.
- A new syndrome of Mullerian dysgenesis with conductive deafness, facial dysmorphic features, vertebral anomalies, and distal limb

abnormalities. 2<sup>nd</sup> Joint Clinical Genetics Conference of March of Dimes and American College of Medical Genetics, 6-9 March, 1995, Los Angeles, USA.

- The multidisciplinary approach in the management of Neurofibromatosis - the Sheffield Special NF clinic. 11<sup>th</sup> Clinical Care Symposium of the National Neurofibromatosis Foundation, USA, 5 March, 1995, Los Angeles, USA.
- The clinical and pathological features of autosomal recessive Aicardi-Goutierre syndrome of progressive nervous system degeneration and intracranial calcification. Spring Conference of the Clinical Genetics Society, 15 March 1995, London.

**1996**

- Assisted reproduction in male cystic fibrosis patients with congenital absence of the vas deferens. York Meeting of the British Society of Human Genetics, 1996.
- Experience of using donated oocytes in assisted reproduction in females affected or a carrier with a genetic disease. American Society of Human Genetics meeting, San Francisco, 29 Oct- 2 Nov., 1997
- Multiple congenital anomaly (MCA) syndromes associated with abnormal Mullerian development: A clinical and genetic review 7<sup>th</sup> Manchester Birth Defects Conference, 8-11 Oct. 1996
- Multiple anomalies associated with cutis marmorata telangiectatica congenita and hemangiomas: A clinical review 7<sup>th</sup> Manchester Birth Defects conference, 8-11 Oct. 1996.
- Progeroid syndromes: A clinical review. 7<sup>th</sup> Manchester Birth Defects Conference, 8-11 Oct. 1996.
- Focal facial dermal dysplasia angiokeratoma and osteochondromal extension of a phenotype or a new syndrome of mesodermal abnormalities. 7<sup>th</sup> Manchester Birth Defects Conference, 8-11 Oct. 1996.

**1998**

- Abnormal Mullerian duct development in association with multiple developmental anomalies: Review of the clinical spectrum and etiologic considerations. Annual meeting of the American Society of Human Genetics Oct. 1998, Denver, Colorado, USA.

**2001**

- Fetal 'ACE-inhibitor' like syndrome: Spring meeting of the Clinical Genetics Society, 22 March 2001, London.

**2004**

- 'Genetics of the cardiovascular disorders in childhood': First Asian Genomics Workshop, 12-22 April, Shenzhen, PR China.
- 'Genetic variation among the Indian subcontinent populations', First Asian Genomics Workshop, 12-22 April, PR China
- 'The spectrum and heterogeneity of genetic disorders among people from the Indian subcontinent', First Asian Genomics Workshop, 12-22 April, PR China.

**2006**

- 'The role of Wnt7A in human limb development' Sheridan E, Woods CG, Stricker S, Seeman P, Stern R, Cox J, Roberts E, Springer K, Scott S, Karbani G, Malik S, Bond J, **Kumar D**, Al-Gazali L, Mundlos S. BSHG Annual Conference, 18-20 September 2006 (Spoken presentation; contribution original clinical case report)

- ‘Hypertrophic cardiomyopathy in Fabry disease presenting with sudden cardiac death.’ Adila AlKindy, **D Kumar**, S Davies. BSHG Annual Conference, 18-20 September, York (Poster presentation; contribution- data from Fabry disease clinical audit)

#### 2007

- Notch 3 gene mutation associated with Sneddon’s syndrome- a complex disorder of unknown etiology with variable clinical features overlapping the phenotype of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy [CADASIL]. (Poster) **D. Kumar**, J. Holroyd, I. Frayling, I. Ferguson (ASHG meeting 2007 San Diego, USA).

#### 2008

- The multi-disciplinary approach to delivering the specialist clinical cardiovascular genetic service in Wales. **Dhavendra Kumar**,<sup>1</sup> Sally Davies,<sup>1</sup> Annie Procter,<sup>1</sup> Ian Frayling,<sup>1</sup> Phillip Thomas,<sup>2</sup> Ian McDowell,<sup>3</sup> Julian Sampson,<sup>4</sup> 1. All Wales Medical Genetics Service, Institute of Medical Genetics, University Hospital of Wales, Heath Park, Cardiff, Wales, UK; 2. Department of Cardiology, Morriston Hospital, Swansea and Chair- Cardiac Services Network for Wales, Wales Assembly Government, Cardiff, UK; 3. Department of Biochemistry and Director-Laboratory Medicine, University Hospital of Wales, Cardiff, UK; 4. Academic Head, Institute of Medical Genetics, Cardiff University, UK and Director, Wales Gene Park, Cardiff University, Heath Campus, Cardiff, UK (Poster) BSHG 2008, 15-17 September, York.
- Dealing with sudden unexpected cardiac death- a new challenge. Mrs. Mary Nicol, **D Kumar**, C Owen, P O’Callaghan. (Poster) BSHG 2008, 15-17 September, York

#### 2009

- Genetic Counselling for Inherited Cardiovascular Conditions: A Comparison between Wales (UK), Australian and North American Practice. Nicol, M(1), **Kumar, D** (1), Procter, A (1), Edwards, A (1); Care, M (2) and Macciocca, I (3) (1) All Wales Medical Genetics service, Institute of Medical Genetics, University Hospital of Wales, Cardiff (2) Department of Medical Genetics, The Fred Litwin Family Centre, Mount Sinai Hospital, Toronto Canada; (3) Victoria Genetics Services, Murdoch Children’s Research Institute, Children’s Hospital, Melbourne Australia; British Society of Human Genetics, 2009, Warwick, England, UK and American Society of Human Genetics 2009, Honolulu, Hawaii, USA
- A focus group study of health care professionals involved in the care of families in Wales affected by sudden arrhythmic death syndrome (SADS) *Bethan Handerson*<sup>1</sup>, *Dhavendra Kumar*<sup>2</sup>, *Mary Nicol*<sup>2</sup>, *Kate Brain*<sup>3</sup>, *Rachel Iredale*<sup>4</sup>, *Angus Clarke*<sup>2,3</sup> 1. Bangor University; 2. Institute of Medical Genetics, University Hospital of Wales, Cardiff; 3. Institute of Medical Genetics, School of Medicine, Cardiff University, Cardiff; 4. Genomics Policy Unit, University of Glamorgan, Wales. BSHG 2009, Warwick, England.

## 12. Attendance at Professional courses and workshops

- **Course on Clinical Genetics**, 27-28 May 1983, Department of Human Genetics, University of Edinburgh. This course was an excellent introduction to the general and applied aspects of medical genetics.

- **Medical Genetics, 1984.** Course organised by the Foundation for Advanced Education in the Sciences, National Institutes of Health, Washington DC, 10-12, May 1984. It was a superb workshop focusing on all aspects of medical genetics, accredited by the American Board of Medical Genetics for those studying for the Board certification and as well as for the continued medical education in medical genetics.
- **Short Course in Clinical Genetics, May 1987,** Institute of Child Health, London. A very practical course in medical genetics designed for professionals and scientists from different backgrounds.
- **Diagnosis of Skeletal Dysplasias,** St. Thomas's Hospital, London, Nov. 1987. A very useful and instructive workshop on the clinical and radiological diagnosis of inherited skeletal dysplasias.
- **International Workshop on Neurofibromatoses,** Oxford, Sept. 1992. This workshop was organised jointly by the UK and USA Neurofibromatosis Associations. Plenary lectures were given by renowned clinicians and scientists from all over the world. It was very helpful to me in developing my own professional interest in neurofibromatosis.
- **Management training course for Senior Registrar:** One week residential course for management training aimed at Senior Registrars and Career grade doctors - August 1994, organised by the Trent Institute of Health Management and Administration.
- **'Embryos, Genes and Birth Defects',** 5-7 May 1998, Institute of Child Health, London. This course was aimed at clinicians and scientists interested in developmental biology and dysmorphology. It provided the latest and comprehensive review on various aspects of developmental genetics as applied to dysmorphology.
- **Counselling and communications skills development: 2001-** A four module course designed to develop necessary counselling and communications skills. This postgraduate course is being regularly offered to all categories of Doctors and related health professionals by the Trent Centre for Post Graduate education, University of Nottingham.
- **Teaching the Teachers course 2001:** A professional course to help professionals engaged in teaching and training. Offered by the University of Sheffield.
- **Symposium on "Century of Mendelism in Human Genetics",** 30-31 October 2001, Royal Society of Medicine, London.
- Attendance at an approved programme leading to **'Postgraduate certificate in Medical Education',** 2002-3, Staffordshire University, England.
- **'Genetics of musculoskeletal diseases',** 13 Feb 2004, Royal Society of Medicine, London.
- **'Counselling course for seniors',** 26-28 February 2004, Taunton, Somerset.
- **'Skeletal dysplasia instructional day',** Oxford, 19 March 2004, organised by Skeletal Dysplasia Group (UK)
- **'Manchester cancer genetics course',** Manchester, 22-24 June 2004.
- **'Nuts and Bolts of Health service management',** 7-8 July 2004, organised by Oxford PGMDE.
- **'Manchester Birth Defects Conference',** 9-12 November, 2004
- **'Cardiomyopathy study day',** 26 April, 2006, Guy's Hospital, London
- **'Genetic advances in adult and paediatric cardiology',** 15 May 2006, Royal Society of Medicine, London.

- **‘Paediatric Neurogenetics Club’**, 25 September 2006, Cardiff
- Regular attendance at annual meetings of **British Society of Human Genetics** and its affiliate **Clinical Genetics Society**.

### **13.Membership of Professional Organizations**

1. The Clinical Genetics Society, UK (1980 to 1986- merger with BSHG)
2. British Medical Association from 1985 to date
3. The Skeletal Dysplasia Group (UK) from 1987 to date
4. The British Society of Human Genetics from 1996 to date
7. American Society of Human Genetics from 1997
8. Life Member- Indian Society of Human Genetics, 2002
9. Human Genome Organization (HUGO), 2005
10. Society for Genomics, Policy and Population Health, 2006