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# Professor Eamonn R. Maher BSc MB ChB MD MA FRCP FMedSci

Professor of Medical Genetics

Reproduction, Genes and Development

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## About

Eamonn Maher is Professor of Medical Genetics and Academic Lead for the Centre for Rare Diseases and Personalised Medicine.

Eamonn has published over 300 research papers in scientific journals as well as reviews and book chapters in the fields of cancer genetics, human developmental genetics, epigenetics and genomic imprinting. He has received major grants from Cancer Research UK, Myrovlytis Trust, Wellcome Trust and other funding agencies.

He has a particular interest in research training for clinician and non-clinician scientists and had supervised more than 25 PhD or MD students to completion.

## Qualifications

- Fellowship of the Academy of Medical Sciences 2006
- Fellow of Royal College of Physicians 1996
- MD 1988
- MB ChB (with Honours) 1980
- BSc (1st Hons) Physiology 1977

## Biography

Eamonn Maher qualified with MB ChB (Hons) in Medicine from the University of Manchester after taking an intercalated BSc (1st Hons) in Physiology. After clinical and research training in Manchester, Cambridge Leeds and London, he was appointed Clinical Lecturer in Medical Genetics in Cambridge University Department of Pathology and Senior Registrar in Clinical Genetics in the East Anglian Region Genetics Service at Addenbrooke's Hospital, Cambridge. After being promoted to University Lecturer (Honorary Consultant) in 1991, he moved to Birmingham to take up the Chair of Medical Genetics in 1996.

Since his appointment Eamonn has built a thriving Medical and Molecular Genetics research theme and is the lead academic for the recently established Centre for Rare Diseases and Personalised Medicine which focuses on research into monogenic inherited disorders.

Eamonn has published widely on the clinical and molecular genetics of inherited disorders, epigenetics and the genetic basis of kidney cancer. His H-index is 67

## Teaching

**Teaching Programmes**

- BMedSci
- MBChB
- MRes

## Postgraduate supervision

Eamonn is interested in supervising doctoral research students in the following areas:

- Molecular analysis of familial and sporadic renal cell carcinoma
- Molecular analysis of familial and sporadic phaeochromocytoma
- The identification of human disease genes
- Role of genomic imprinting and epigenetics in human disease

If you are interesting in studying any of these subject areas please contact Eamonn on the contact details above, or for any general doctoral research enquiries, please email: dr@contacts.bham.ac.uk (mailto:dr@contacts.bham.ac.uk) or call +44 (0)121 414 5005.

For a full list of available Doctoral Research opportunities, please visit our Doctoral Research programme listings. (http://www.bham.findaphd.com/?

es=y&apl=y&aplt=&show) .

## Research

Eamonn's major research interests are centred on using human disease models to gain insights into the molecular basis of disease and basic biological processes. As a clinically active Clinical Geneticist with his own laboratory he supervises both clinical and non-clinical research fellows and PhD students. His research investigations have had a direct effect on clinical practice, most notably the identification of human disease genes has led to the introduction of national and supraregional molecular genetics diagnostic services for conditions such as von Hippel-Lindau disease, familial phaeochromocytoma, Beckwith-Wiedemann syndrome, multiple pterygium syndrome and foetal akinesia sequence, infantile neuraxonal dystrophy, ARC and Micro syndromes.

His research projects range from clinical aspects of human genetic disease, molecular genetic studies and functional analysis of gene products he has achieved international recognition for three research themes:

**Molecular Pathology of Renal Cell Carcinoma and related tumours**

Following his appointment as Clinical Lecturer in Medical Genetics with Malcolm Ferguson-Smith and John Yates he ascertained a large series of families with von Hippel-Lindau disease and pursued genetic linkage studies, culminating, in collaboration with Farida, Latif, Michael Lerman, Marston Linehan and Berton Zbar, in the identification of the VHL tumour suppressor gene (Science 1993). Subsequently his group demonstrated that somatic inactivation of the VHL tumour suppressor gene occurs in most sporadic renal cell carcinoma, identified genotype-phenotype correlations that have provided important insights into structure-function relationships of the VHL protein and, with Patrick Maxwell and Peter Ratcliffe reported the seminal finding that the VHL protein is a major regulator of HIF-1 and HIF-2 protein levels (Nature 1999). VHL disease provides a paradigm for how molecular studies of rare diseases can provide basic insights into human biology ultimately leading to the rational use of novel therapeutic agents (e.g. sorafenib and sunitinib in advanced renal cell carcinoma). Currently his group is pursuing the identification of novel RCC susceptibility genes and characterising the genetics and function of folliculin which is encoded by the gene for another familial renal cancer syndrome (Birt-Hogg-Dube syndrome). A offshoot of his VHL research was an interest in the genetics of phaeochromocytoma and this led to the first identification of germline mutations in *SDHB* and *SDHD* as causes of familial phaeochromocytoma. Mutations in SDH subunit genes are now recognised as an important cause of phaeochromocytoma susceptibility. Eamonn also has a longstanding interest in epigenetics and his group have highlighted the role of epigenetic inactivation of tumour suppressor genes in renal cancers.

**Epigenetics and Imprinting**

Eamonn has pursued clinical and molecular research into the model imprinting disorder Beckwith-Wiedemann syndrome. These studies have led to major advances in our knowledge of the molecular pathology of BWS and the identification of genotype-phenotype correlations that have influenced clinical practice. In addition these studies have provided unique insights into mechanisms of imprinting that had not been apparent from animal studies. In addition to molecular investigations, the clinical insight that there is an increased risk of imprinting disorders after assisted reproductive technologies has proven to be a important finding.

**Identification of human disease genes**

After moving to Birmingham Eamonn initiated a research programme to identify autosomal recessive disease genes by autozygosity mapping and established a National Aytozygosity Mapping Resource with Richard Trembath, Bob Mueller and Geoff Woods. His research programme identified novel disease genes for >20 recessively inherited disorders including achromatopsia (*GNAT2*), ARC syndrome (*VPS33B*), Warburg MICRO syndrome (*RAB3GAP1*), Martsolf syndrome (*RAB3GAP2*), Hermansky-Pudlak syndrome (*BLOC1S3*), infantile neuraxonal dystrophy (*PLA2G6*), multiple pterygium syndrome (*CHRNG, RAPSN*, *DOK7*), familial histiocytosis (*SLC29A3*), Fowler syndrome, primary immunodeficiency (*TRAC1*), Beckwith-Wiedemann syndrome-like imprinting disorder (*NALP2*), infantile parkinsonism-dystonia syndrome (*DAT1*) and trichoheptoenteric syndrome (*TTC37*). These gene discovery projects have not only provided novel insights into human disease processes but have offered excellent training projects for clinical fellows and PhD students who have gone on to establish themselves as independent investigators.

The ultimate aim of Eamonn's research is to not only to improve the diagnosis and management of human genetic disease but also to develop novel targeted therapies and this was the rationale behind the establishment of the *Centre for Rare Diseases and Personalised Medicine*.

## Other activities

- Honorary Consultant in Clinical Genetics, West Midlands Region Genetics Service, Birmingham Women's Hospital, B15 2TG,
- Member Journal Editorial Board: *Clinical Epigenetics*
- Editor in Chief *Journal of Medical Genetics* 1988-2009

## Publications


Morgan NV, Goddard S, Cardno TS, McDonald D, Rahman F, Barge D, Ciupek A,Straatman-Iwanowska A, Pasha S, Guckian M, Anderson G, Huissoon A, Cant A, Tate WP, Hambleton S, Maher ER. Mutation in the TCRα subunit constant gene (TRAC)leads to a human immunodeficiency disorder characterized by a lack of TCRαβ+ T cells. **J Clin Invest**. 2011 Feb 1;121(2):695-702. doi: 10.1172/JCI41931. Epub 2011 Jan 4.

Morris MR, Ricketts CJ, Gentle D, McRonald F, Carli N, Khalili H, Brown M,Kishida T, Yao M, Banks RE, Clarke N, Latif F, Maher ER. Genome-wide methylation analysis identifies epigenetically inactivated candidate tumour suppressor genes in renal cell carcinoma. **Oncogene**. 2011 Mar 24;30(12):1390-401. Epub 2010 Dec 6.

Kurian MA, Li Y, Zhen J, Meyer E, Hai N, Christen HJ, Hoffmann GF, Jardine P, von Moers A, Mordekar SR, O'Callaghan F, Wassmer E, Wraige E, Dietrich C, Lewis T, Hyland K, Heales S Jr, Sanger T, Gissen P, Assmann BE, Reith ME, Maher ER. Clinical and molecular characterisation of hereditary dopamine transporter deficiency syndrome: an observational cohort and experimental study. **Lancet Neurol**. 2011 Jan;10(1):54-62. Epub 2010 Nov 25.

Maher ER. Genetics of familial renal cancers. **Nephron Exp Nephrol**. 2011;118(1):e21-6. Epub 2010 Nov 11.

Morris MR, Maher ER. Epigenetics of renal cell carcinoma: the path towards new diagnostics and therapeutics. **Genome Med**. 2010 Sep 3;2(9):59

Meyer E, Ricketts C, Morgan NV, Morris MR, Pasha S, Tee LJ, Rahman F, Bazin A, Bessières B, Déchelotte P, Yacoubi MT, Al-Adnani M, Marton T, Tannahill D, Trembath RC, Fallet-Bianco C, Cox P, Williams D, Maher ER. Mutations in FLVCR2 are associated with proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome (Fowler syndrome). **Am J Hum Genet**. 2010 Mar 12;86(3):471-8. Epub 2010 Mar 4.

Cullinane AR, Straatman-Iwanowska A, Zaucker A, Wakabayashi Y, Bruce CK, Luo G, Rahman F, Gürakan F, Utine E, Ozkan TB, Denecke J, Vukovic J, Di Rocco M, Mandel H, Cangul H, Matthews RP, Thomas SG, Rappoport JZ, Arias IM, Wolburg H, Knisely AS, Kelly DA, Müller F, Maher ER, Gissen P. Mutations in VIPAR cause an arthrogryposis, renal dysfunction and cholestasis syndrome phenotype with defects in epithelial polarization. **Nat Genet**. 2010 Apr;42(4):303-12.

Hartley JL, Zachos NC, Dawood B, Donowitz M, Forman J, Pollitt RJ, Morgan NV,Tee L, Gissen P, Kahr WH, Knisely AS, Watson S, Chitayat D, Booth IW, Protheroe S, Murphy S, de Vries E, Kelly DA, Maher ER. Mutations in TTC37 cause trichohepatoenteric syndrome (phenotypic diarrhea of infancy). **Gastroenterology**. 2010 Jun;138(7):2388-98, 2398.e1-2. Epub 2010 Feb 20. PubMed PMID: 20176027.


## Expertise

Medical and molecular genetics; determining genetic basis of rare single gene disorders to develop targeted therapeutic interventions; familial cancer syndromes (von Hippel-Lindau disease, Birt-Hogg-Dube syndrome, familial renal cell carcinoma); genomic imprinting and epigenetics (Beckwith-Widemann syndrome and epigenetics of human cancer); identification of autosomal recessive disease genes

## Media experience

As a leading expert on medical genetics Eamonn is regularly called upon to comment on the rare conditions he studies. Daily Telgraph feature on **Alstrom Syndrome (http://www.telegraph.co.uk/health/8347660/Rare-diseases-Alstrom-Syndrome.html)**, BBC News Online feature on **Beckwith-Wiedemann syndrome (http://news.bbc.co.uk/1/hi/health/3555078.stm)**.

Alternative contact number available for this expert: **contact the press office (http://www.birmingham.ac.uk/news/contacts/index.aspx)**

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