

## SAQIB MAHMOOD

MBBS, MSc., PhD

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### Employment

#### **University of Health Sciences Lahore [July 2008 to date].**

*Assistant Professor, Department of Human Genetics & Molecular Biology*

- Teach MPhil and PhD students in Human Genetics and other Basic Medical Sciences.
- Supervise research projects of MPhil and PhD students
- Offer Cytogenetics and Molecular Genetics diagnostic services
- Conduct Genetic Counseling Clinic
- Academic supervision of Allied Health Sciences Department

#### **The Children's Hospital & The Institute of Child Health, Ferozpur Road Lahore [February 1999 to June 2008].**

*Assistant Professor, Paediatric Genetics 17-06-2003 to 30-06-2008*

*Medical Officer, Paediatric Genetics 17-02-1999 to 16-06-2003*

- Established Paediatric Genetics department in this hospital which is the first of its kind in the public sector hospitals in Pakistan. This department consists of Cytogenetics laboratory, Molecular Genetics laboratory and Genetic Counseling clinic.
- Cytogenetics laboratory provides facilities for chromosome analysis to patients with dysmorphic features, ambiguous genitalia, suspected X chromosome defects, mental retardation, couples with infertility and women with history of multiple abortions etc
- Molecular Genetics laboratory provides facilities for the DNA based screening of Thalassaemia and Duchenne Muscular Dystrophy.
- Genetic Counseling Clinic provides counseling to the patients and the families explaining the genetic disorder, implications of the test results, options available, prognosis of the disorder, its management and the risk of having another affected child in the future pregnancies.

#### **Armed Forces Institute of Pathology Rawalpindi [July 1997 to December 1998].**

*Medical Officer on training*

- In the Molecular Genetics Laboratory learned the techniques of DNA extraction from peripheral blood and chorionic villi samples, basic techniques of PCR and procedure of mutation detection in thalassaemia gene by ARMS method for molecular screening and prenatal diagnosis.
- In Cytogenetics laboratory learned the techniques of culturing lymphocytes, harvesting cultures, making slides and staining slides for G banding.

#### **The Children's Hospital & The Institute of Child Health, Ferozpur Road Lahore [May 1996 to June 1997].**

*Medical Officer School Health Services*

- Worked in the pathology laboratory providing diagnostic facilities to the indoor, outdoor and emergency departments.

- Visited the assigned school for the health assessment of school going children.

**King Edward Medical College Lahore [April 1993 to October 1994].**

*Demonstrator Anatomy*

- Taught gross anatomy, neuro-anatomy and histology to the undergraduate students of medical, dental and physiotherapy classes.

**Allama Iqbal Medical College Lahore [November 1989 to January 1993].**

*Demonstrator Anatomy*

- Taught gross anatomy, neuro-anatomy and histology to the undergraduate medical students.

**Mayo Hospital Lahore [May 1989 to November 1989].**

*Medical Officer/Casualty Medical Officer*

- In Urology department worked in the indoor and outdoor departments and operation theatre.
- In Casualty and Emergency departments managed medical and surgical emergencies.

**Services Hospital Lahore [September 1988 to March 1989].**

*House Surgeon*

- Worked in the in the surgical indoor, outdoor and emergency departments.
- Assisted in major surgeries and carried out independently minor surgical procedures.

**Services Hospital Lahore [March 1988 to August 1988].**

*House Physician*

- Worked in the in the medical indoor, outdoor, emergency and intensive care departments.

## Education

### Quaid-e-Azam University, Islamabad

#### PhD in Human Genetics from Department of Biochemistry and Molecular Biology (2006)

- Research thesis on "Genetics of Duchenne Muscular Dystrophy and Primary Microcephaly in Pakistani Population"

### Brunel University United Kingdom

#### MSc in Medical Genetics with Immunology (1996)

- Full time course covering the human molecular genetics, molecular genetics of human diseases, biochemical genetics of human diseases, cancer genetics, immunology and immunogenetics courses.

### Allama Iqbal Medical College Lahore (University of the Punjab)

#### Bachelor of Medicine and Bachelor of Surgery (M.B.B.S) (1987)

- Five year full time course in pre-clinical and clinical medical subjects with hospital based training.

## Further Trainings & Workshops Attended

**International Workshop and Symposium on Integrating Genetics in the Medical Curriculum** organized by International Genetics Education Network in association with Human Genetics Unit, Faculty of Medicine University of Colombo  
Sri Lanka 12-14<sup>th</sup> June, 2013

**German Academic Exchange Program (DAAD) fellowship for research at The Institute of Human Genetics & Anthropology, Friedrich Schiller University, Jena, Germany**

1<sup>st</sup> July 2011 to 31<sup>st</sup> August 2011

**4<sup>th</sup> International Conference on Birth Defects and Disabilities in Developing Countries**

New Delhi India from 4<sup>th</sup> - 7<sup>th</sup> October, 2009.

**First International Symposium and Workshop-cum-Training Course on Molecular Medicine and Drug Research.** Jointly organized by Dr Panjwani Center for Molecular Medicine and Drug Research, International Center for Chemical and Biological Sciences, University of Karachi and Organization of Islamic Conference Standing Committee on Scientific and Technological Cooperation Islamabad January 16-30, 2007.

**Molecular Biology Techniques for the genetic diagnosis of Haemophilia.** Hands on training under IHTC fellowship in the Institute of Immunohaematology, Mumbai, India from January 21, 2006 to February 9, 2006.

**International Conference and Workshop on Inborn Errors of Metabolism** at Sir Ganga Ram Hospital, New Delhi from September 28 to October 02, 2005.

**Pre-implantation Genetic Diagnosis Training** at Royal Free Hospital, University College London, UK from January 28 to February 1, 2002.

**Histocompatibility and Immunogenetics Technology Workshop** at All India Institute of Medical Sciences, New Delhi, from March 1 to March 4, 2001.

## Postgraduate

## **Supervision**

### **PhD Research Supervisor**

1. Molecular markers of susceptibility to Prion disease in local Humans and Domesticated Bovids. (**PhD degree awarded**)
2. Expression of Circadian Clock and Cell Cycle Genes in Chronic Lymphocytic Leukemia. (**PhD degree awarded**)
3. Inflammation in Stroke: Correlation of Cytokines TNF-Alpha, IL-10 and Homocysteine in Clinical Stroke (**PhD research in Progress**)
4. Molecular Genetic studies in Pakistani families with Schizophrenia (**PhD research in Progress**)

### **PhD Research Co-Supervisor**

1. Genetic variant in the Leptin, Leptin Receptor and FTO Genes in Obese Pakistani Population (**PhD degree awarded**)
2. Immunochemical and Genetic characterization of Muscular Dystrophies in local patients (**PhD research in Progress**)

### **MPhil/MSc Research Supervisor**

1. Molecular analysis of *GALT* gene in patients with Galactosemia (**MPhil degree awarded**)
2. Serum Leptin levels and Leptin gene mutations in obese children (**MPhil degree awarded**)
3. ITD and D835 mutations of FMS like tyrosine kinase 3 gene in patients with Acute Myeloid Leukemia (**MPhil degree awarded**)
4. Serum Leptin levels and Leptin gene mutations in obese children (**MPhil degree awarded**)
5. Exon Deletion Studies in patients of Duchenne Muscular Dystrophy by Multiple Ligation Probe Amplification (**MSc degree awarded**)
6. Molecular analysis of congenital adrenal hyperplasia secondary to 21-Hydroxylase deficiency in Pakistani population (**MSc degree awarded**)
7. Sequence analysis of Bone Morphogenetic Protein 15 Gene in patients of premature ovarian failure (**MPhil degree awarded**)
8. Biochemical profile of patients with 46, XY Disorder of Sex Development (**MPhil degree awarded**)

### **MPhil/MSc Research Co-Supervisor**

1. Analysis of Intron 13 (CA)<sub>n</sub> Repeats and *HIND III* markers of factor VIII gene and its application for carrier direction (**MPhil degree awarded**)
2. Carrier detection of Haemophilia B by linkage analysis (**MPhil degree awarded**)
3. Nucleophosmin (*NPM1*) gene mutations in local patients with Acute Myeloid Leukemia (**MPhil degree awarded**)
4. Molecular Genetics studies in families with autosomal recessive primary microcephaly (MCPH) from Punjab (**MPhil degree awarded**)

## Research Grants Acquired

1. Grant from Higher Education Commission worth Rs. 1,720,740/- for the project title "Inflammation in Stroke: Correlation of Cytokines TNF-Alpha, IL-10 and Homocysteine in Clinical Stroke."
2. Grant from Higher Education Commission worth Rs. 2,967,550/- for the project title "Molecular Genetic Studies in Pakistani Families with Schizophrenia."
3. Grant from Pakistan Science Foundation worth Rs. 1,076,420/- for the project title "Molecular Genetic Studies in Pakistani Families with Autosomal Recessive Microcephaly"

## Research Publications

1. Rana S, Shahid A, Ullah H, **Mahmood S**.  
Lack of Association of the NPAS2 Gene Ala394Thr Polymorphism (rs2305160:G>A) with Risk of Chronic Lymphocytic Leukemia.  
Asian Pac J Cancer Prev. 2014;15(17):7169-74.
2. Rana S, Munawar M, Shahid A, Malik M, Ullah H, Fatima W, Mohsin S, **Mahmood S**  
Deregulated expression of circadian clock and clock-controlled cell cycle genes in chronic lymphocytic leukemia.  
Mol Biol Rep. 2014 Jan;41(1):95-103.
3. Koehler K, Malik M, **Mahmood S**, Gießelmann S, Beetz C, Hennings JC, Huebner AK, *et.,al*.  
Mutations in GMPPA Cause a Glycosylation Disorder Characterized by Intellectual Disability and Autonomic Dysfunction.  
Am J Hum Genet. 2013 Oct 3;93(4):727-734.
4. Shahid A, Rana S, Saeed S, Imran M, Afzal N, Mahmood S.  
Common Variant of FTO Gene, rs9939609, and Obesity in Pakistani Females.  
Biomed Res Int. 2013;2013:324093. Epub 2013 Sep 2.
5. Ejaz H, Ikram-ul-Haq, **Mahmood S**, Zafar A, Javed MM.  
Detection of extended-spectrum  $\beta$ -lactamases in Klebsiella pneumoniae: Comparison of phenotypic characterization methods.  
Pak J Med Sci 2013;29(3):768-772.
6. Khokher S, Qureshi MU, Mahmood S, Nagi AH.  
Association of immunohistochemically defined molecular subtypes with clinical response to presurgical chemotherapy in patients with advanced breast cancer.  
Asian Pac J Cancer Prev. 2013;14(5):3223-8.
7. Bijarnia S, Kohli S, Puri RD, Jacob RJ, Saxena R, Jalan A, Siermans EA, **Mahmood S**, Verma IC.  
Molecular Characterisation and Prenatal Diagnosis of Aspartoacylase Deficiency (Canavan Disease)-Report of Two Novel and Two Known Mutations from the Indian Subcontinent.  
Indian J Pediatr. 2013 Jan;80(1):26-31.

8. Mahmood U, Imran M, Naik SI, Cheema HA, Saeed A, Arshad M, **Mahmood S**.  
Detection of common mutations in the GALT gene through ARMS.  
Gene. 2012 Nov 10;509 (2):291-4.
9. Imran M, Saeed MT, **Mahmood S**, Mohsin S, Sohail S, Munir A, Nazir H.  
A strategy for carrier detection in Pakistani haemophilia B families.  
Haemophilia. 2012 Jul;18(4):e348-9.
10. Shahid A, Saeed S, Rana S, **Mahmood S**.  
Family history of diabetes and parental consanguinity: important risk for impaired fasting glucose in south east Asians.  
West Indian Med J. 2012 Jun;61(3):219-23.
11. Imran M, **Mahmood S**, Babar ME, Hussain R, Yousaf MZ, Abid NB, Lone KP  
PRNP gene variation in Pakistani cattle and buffaloes.  
Gene. 2012 Aug 15;505(1):180-5.
12. Bugvi SM, Imran M, **Mahmood S**, Hafeez R, Fatima W, Sohail S.  
Screening of intron 1 inversion and three intragenic factor VIII gene polymorphisms in Pakistani hemophilia A families.  
Blood Coagul Fibrinolysis. 2012 Mar;23(2):132-7.
13. Imran M, **Mahmood S**, Hussain R, Abid NB, Lone KP  
Frequency distribution of PRNP polymorphisms in the Pakistani population.  
Gene. 2012 Jan 15;492(1):186-94.
14. Imran M, **Mahmood S**.  
An overview of human prion diseases.  
Virol J. 2011 Dec 24;8(1):559.
15. Khokher S, Qureshi W, **Mahmood S**, Saleem A, Mahmud S.  
Knowledge, Attitude and Preventive Practices of Women for Breast cancer in the Educational Institutions of Lahore, Pakistan.  
Asian Pac J Cancer Prev. 2011;12(9):2419-24.
16. Imran M, **Mahmood S**.  
An overview of animal prion diseases.  
Virol J. 2011 Nov 1;8 (1):493.
17. Kausar R, Hassan MJ, Khan B, Basit S, **Mahmood S**, Mir A, Ahmad W, Ansar M  
Mutations in WDR62 gene in Pakistani families with autosomal recessive primary microcephaly  
BMC Neurol. 2011 Oct 1;11(1):119
18. Fatima W, Shahid A, Imran M, Manzoor J, Hasnain S, Rana S, **Mahmood S**

- Leptin deficiency and leptin gene mutations in obese children from Pakistan  
Int J Pediatr Obes. 2011 Oct;6(5-6):419-27.
19. Khokhar S, **Mahmood S**, Qureshi MU, Khan SA, Chaudhry NA  
"Initial Clinical Response" to neoadjuvant chemotherapy: An In-vivo chemosensitivity test for efficacy in patients with advanced breast cancer  
Asian Pac J Cancer Rev. 2011, 12(4):939-46
  20. **Mahmood S**, Ahmad W, Hassan MJ  
Autosomal Recessive Primary Microcephaly (MCPH): Clinical manifestations, genetic heterogeneity and mutation continuum  
Ophanet J, Rare Dis. 2011, Jun 13;6:39
  21. **Mahmood S**, Beetz C, Tahir MM, Imran M, Mumtaz R, Bassmann I, Jahic A, Malik M, Nürnberg G, Ali Hassan SA, Rana S, Nürnberg P, Huebner CA.  
First HPSE2 missense mutation in Urofacial Syndrome.  
Clin Genet. 2011, Feb 17. doi: 10.1111/j.1399-0004.2011.01649.x.
  22. Khokher S, **Mahmood S**, Khan SA.  
Response to neoadjuvant chemotherapy in patients with advanced breast cancer: a local hospital experience.  
Asian Pac J Cancer Prev. 2010;11(2):303-8.
  23. Sajid N, Ahmed N, **Mahmood S**.  
Clinicopathological features of transient myeloproliferative syndrome and congenital leukaemia.  
J Coll Physicians Surg Pak. 2010 Sep;20(9):576-80.
  24. Rana S, **Mahmood S**  
Circadian rhythm and its role in malignancy  
J Circadian Rhythms. 2010 Mar 31;8:3.
  25. Asim Mumtaz, Nadeem Afzal, Waqas Sami, Romeeza Tahir, Khursheed Javeed, **Saqib Mahmood**  
Evaluation of four ELISA based immunoassays for the detection of IgM antibodies against Dengue virus  
Biomedica Jan – Jun 2010; 26: 54-57
  26. Nadia Rashid, Asim Mumtaz, **Saqib Mahmood**, Nadeem Afzal, Salma Haq, Tanveer Ahmad.  
Assessment of Thyroid Dysfunction in Down Syndrome Patients  
Pak Paed J Dec 2009;33(4):211-7
  27. Hassan MJ, **Mahmood S**, Ali G, Bibi N, Waheed I, Rafiq MA, Ansar M, Ahmad W.  
Intragenic deletions in the *dystrophin* gene in 211 Pakistani Duchenne Muscular Dystrophy Patients  
Pediatrics International (2008) 50, 162–166

28. Mahfoozur Rehman, Abid Ali Qureshi, Quratul Ain, **Saqib Mahmood**  
MRI findings of Pituitary Gland in Beta Thalassaemia Major Patients  
Pak Paed J 2007;31(3):137-41
29. Gul A, Hassan MJ, **Mahmood S**, Chen W, Rahmani S, Naseer MI, Dellefave L, Muhammad N, Rafiq MA, Ansar M, Chishti MS, Ali G, Siddique T, Ahmad W.  
Genetic studies of Autosomal Recessive Primary Microcephaly in 33 Pakistani families: novel sequence variants in ASPM gene. Neurogenetics (2006) May;7(2):105-10
30. Rafiq MA, Ansar M, **Mahmood S**, Haque S, Faiyaz-ul-Haque M, Leal SM, Ahmad W.A  
Recurrent Intragenic Deletion Mutation in DSG4 gene in Three Pakistani Families with Autosomal Recessive Hypotrichosis” J Invest Dermatol. (2004) Jul;123(1):247-8