-Ref: 213/1
- Egypt

CURRICULUM VITAE MAI MOHAMEDABD EL-AZIZ

Personal Details

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Academic and Professional Qualifications:

2007 **PhD** University of London, UK.

1999 **M.Sc.** Clinical and Chemical Pathology, Tanta University, Egypt

1994 **MB ChB** with honours, Faculty of Medicine, Tanta University, Egypt.

Posts held

Present Post

Assistant Lecturer: Department of Clinical and Chemical Pathology-Tanta University Hospitals- Tanta- Egypt.

Previous Posts

PhD student: Institute of Ophthalmology, Department of Molecular Genetics, London, UK. March 2004 – March 2007.

Assistant Lecturer: Department of Clinical and Chemical Pathology, Tanta University, Egypt. December 2001 – March 2004.

Research assistant: Institute of Ophthalmology, Department of Molecular Genetics, London, UK. December 1999 – December 2001.

A Demonstrator in Clinical Pathology department- Faculty of Medicine- Tanta University, Egypt. September 1999 – December 1999.

Resident: Clinical Pathology Department, Tanta University Hospital, Egypt. September 1996 – September 1999.

Physician (House physician): Ministry of Health, Tanta, Egypt. March 1996 – September 1996.

House Officer: Tanta University Hospital Tanta, Egypt. March 1995 – February 1996.

Professional Experience

I have an overall experience of seven years in the field of molecular genetics in the Institute of Ophthalmology in the UK. This is in addition to my five years of experience working as a clinical scientist and assistant lecturer in the department of clinical chemistry and haematology in Tanta University Hospital in Egypt.

I have been graduated from the University of Medicine in 1994. After a year and half of post graduation training on different specialities, I felt particularly drawn towards the clinical chemistry and haematology branch of Medicine. Hence, I started working as a resident in the Department of Clinical Pathology for 3 years and got specialised as a clinical pathologist in 1999. During my time as a resident I also carried out my MSc on measuring the level of BCL2 in the serum of patients with non-Hodgkin lymphoma as well as relating this to the response to treatment.

I then wanted to expand my knowledge and get experience in the field of human genetics. Therefore, I moved to the UK to work as a research assistant in the Department of Molecular Genetics- Institute of Ophthalmology between December 1999 and October 2001. During this time, I have been able to rapidly learn many techniques in this field including mapping and screening genes for mutations. Significantly I have contributed to the work of two PhD students which involved studying the molecular genetics of corneal dystrophy.

Afterwards, I have been nominated a grant for the PhD in 2004 from the Islamic Development Bank and decided to carry out my PhD in molecular genetics. I carried out my PhD titled "Molecular genetics of autosomal recessive retinitis pigmentosa" under the supervision of Prof. Bhattacharya between April 2004 and April 2007. This study aimed at identifying the causative gene for one of the major loci for autosomal recessive retinitis pigmentosa (RP25). During this study I carried out extensive bioinformatics analysis together with mutation screening of large number of genes from the RP25 interval. I was also employing other methods in order to refine the genetic interval such as recruiting additional families in order to identify novel crossovers. Additionally, I utilised high throughput techniques such as the Genechip mapping 10K array as well as the CGH array analyses. Towards the end of my PhD, I have managed to critically refine the genetic interval I was working on from the original 34 Mb to only 100 Kb. This had a significant

impact on the success of cloning of the RP25 gene. Myself and other colleagues have successfully cloned a novel gene (*EYS*) for recessive RP. This work involved full characterisation of the gene in addition to extensive bioinformatics analysis. Also, my colleagues and I have designed an antibody specific for this novel protein which allowed us to study the cellular localisation of this protein. Subsequently, this will shed the light on the function of this novel gene.

Publications

1. **Abd El-Aziz MM**, Barragan I, O'Driscoll CA, Goodstadt L, Prigmore E, Borrego S, Mena M, Pieras JI, El-Ashry MF, Safieh LA, Shah A, Cheetham ME, Carter NP, Chakarova C, Ponting CP, Bhattacharya SS, Antinolo G. EYS, encoding an ortholog of Drosophila spacemaker, is mutated in autosomal recessive retinitis pigmentosa. ***Nat Genet.*** (2008) **40**: 1285-7.

2. **Abd El-Aziz MM**, Barragan I, O'Driscoll C, Borrego S, Abu-Safieh L, Pieras JI, El-Ashry MF, Prigmore E, Carter N, Antinolo G, Bhattacharya SS. Large-scale molecular analysis of a 34 Mb interval on chromosome 6q: major refinement of the RP25 interval. ***Ann. Hum. Genet.*** (2008) **72**: 463-77.

3. Barragán I*, **Abd El-Aziz MM***, Borrego S, El-Ashry MF, O'Driscoll C, Bhattacharya SS, Antiñolo G. Linkage validation of RP25 Using the 10K genechip array and further refinement of the locus by new linked families. ***Ann. Hum. Genet.*** (2008) 72: 454-62.
*** Joint authorship**

4. Barragán I, Borrego S, **Abd El-Aziz MM**, El-Ashry MF, Abu-Safieh L, Bhattacharya SS, Antiñolo G. Genetic analysis of FAM46A in Spanish families with autosomal recessive retinitis pigmentosa: characterisation of novel VNTRs. ***Ann Hum Genet.*** (2008) **72**: 26-34.

5. **Abd El-Aziz MM**, El-Ashry MF, Chan WM, Chong KL, Barragan I, Antiñolo G, Pang CP, Bhattacharya SS.A novel genetic study of Chinese families with autosomal recessive retinitis pigmentosa. ***Ann Hum Genet.*** (2007) **71**: 281-94.

6. **Abd El-Aziz MM**, Patel RJ, El-Ashry MF, Barragan I, Marcos I, Borrego S, Antiñolo G, Bhattacharya SS. Exclusion of four candidate genes, KHDRBS2, PTP4A1, KIAA1411 and OGFRL1, as causative of autosomal recessive retinitis pigmentosa. ***Ophthalmic Res.*** (2006) **38**: 19-23.

7. **Abd El-Aziz MM**, El-Ashry MF, Barragan I, Marcos I, Borrego S, Antiñolo G, Bhattacharya SS. Molecular genetic analysis of two functional candidate genes in the autosomal recessive retinitis pigmentosa, RP25, locus. ***Curr Eye Res.*** (2005) **30**: 1081-7.

8. El-Ashry MF, **Abd El-Aziz MM**, Bhattacharya SS. A clinical and molecular genetic study of Egyptian and Saudi Arabian patients with primary congenital glaucoma (PCG). ***J Glaucoma.*** (2007) **16**: 104-11.

9. Chakarova CF, Papaioannou MG, Khanna H, Lopez I, Waseem N, Shah A, Theis T, Friedman J, Maubaret C, Bujakowska K, Veraitch B, **Abd El-Aziz MM**, *et al.* Mutations in TOPORS cause autosomal dominant retinitis pigmentosa with perivascular retinal pigment epithelium atrophy. ***Am J Hum Genet.*** (2007) **81**: 1098-103.

10. El-Ashry MF, **Abd El-Aziz MM**, Hardcastle AJ, Bhattacharya SS, Ebenezer ND. A clinical and molecular genetic study of autosomal-dominant stromal corneal dystrophy in British population. ***Ophthalmic Res.*** (2005) **37**: 310-7.

11. El-Ashry MF, **Abd El-Aziz MM**, Shalaby O, Wilkins S, Poopalasundaram S, Cheetham M, Tuft SJ, Hardcastle AJ, Bhattacharya SS, Ebenezer ND. Novel CHST6 nonsense and missense mutations responsible for macular corneal dystrophy. ***Am J Ophthalmol.*** (2005) **139**: 192-3.

12. El-Ashry MF, **Abd El-Aziz MM**, Ficker LA, Hardcastle AJ, Bhattacharya SS, Ebenezer ND. BIGH3 mutation in a Bangladeshi family with a variable phenotype of LCDI. ***Eye*** (2004) **18**: 723-8.

13. El-Ashry MF, **Abd El-Aziz MM**, Larkin DF, Clarke B, Cree IA, Hardcastle AJ, Bhattacharya SS, Ebenezer ND. A clinical, histopathological, and genetic study of Avellino corneal dystrophy in British families. ***Br J Ophthalmol.*** (2003) **87**: 839-42.

14. El-Ashry MF, **Abd El-Aziz MM**, Wilkins S, Cheetham ME, Wilkie SE, Hardcastle AJ, Halford S, Bayoumi AY, Ficker LA, Tuft S, Bhattacharya SS, Ebenezer ND. Identification of novel mutations in the carbohydrate sulfotransferase gene (CHST6) causing macular corneal dystrophy. ***Invest Ophthalmol Vis Sci.*** (2002) **43**: 377-82.

Oral Presentations

1. **Mai Abd El-Aziz**, Isabel Barragán, Ciara O'Driscoll, Salud Borrego, Elena Prigmore, Michael Cheetham, Nigel Carter, Chris Ponting, Shomi Bhattacharya and Guillermo Antinolo Identification of a Major Gene (*RP25*) for Autosomal Recessive Retinitis Pigmentosa. (2008) ARVO, Program 3283.

2. M.F. El-Ashry, **M.M. Abd El-Aziz**, S.S. Bhattacharya. Mutation screening of CYP1B1 in Saudi Arabian and Egyptian families with primary congenital glaucoma: identification of novel mutation. (2005) ARVO, Program 1096.

3. M.F. El-Ashry, **M.M. Abd El-Aziz,** N.D. Ebenezer, A.J. Hardcastle, S.S. Bhattacharya. Identification of novel mutations in a carbohydrate sulfotransferase gene CHST6 causing macular corneal dystrophy. (2001) ARVO, Program 1655.

4. **Abd El-Aziz MM**, El-Ashry MF, Ebenezer N, Lehmann OJ, Ocaka L, Wilkie S, Bhattacharya SS. A novel mutation in keratocan causes autosomal recessive cornea plana and microphthalmia. (2001) ARVO

Poster Presentations

1. **M.M. Abd El-Aziz** , M.F. El-Ashry, L.Abu Safieh, W.-M.Chan, C.P. Pang, S.S. Bhattacharya Linkage of Chinese Families With Autosomal Recessive Retinitis Pigmentosa to the RP25 Locus. (2006) ARVO, Program 3273/B743

2. **M.M. Abd El-Aziz**, R.J. Patel, M.F. El-Ashry, I.Barragan, I.Marcos, G.Antinolo, S.S. Bhattacharya Mutation Screening of Four Candidate Genes, KHDRBS2, PTP4A1, KIAA1411, and OGFRL1, in the Autosomal Recessive Retinitis Pigmentosa (RP25) Critical Interval. (2005) ARVO, Program 1808/B577.

3. M.F. El-Ashry, **M.M. Abd El-Aziz**, N.D. Ebenezer, A.J. Hardcastle, S.S. Bhattacharya. Spectrum of Bigh-3 gene mutations and novel polymorphisms in the British populations. (2003) ARVO, Program 3857/B560

4. M.F. El-Ashry, **M.M. Abd El-Aziz,** N.D. Ebenezer, A.J. Hardcastle, S.S. Bhattacharya. Novel mutations in CHST6 gene causing macular corneal dystrophy. (2002) ARVO.

5. M.F. El-Ashry, **M.M. Abd El-Aziz**, N.D. Ebenezer, A.J. Hardcastle, S.S. Homozygozity mapping of a Bangladeshi family with cornea plana (CNA2) to chromosome 12 (2000) ARVO.

Meetings and courses attended

Meetings

1. ARVO, Fort Lauderdale, USA, May 2008
2. ARVO, Fort Lauderdale, USA, May 2006
3. ARVO, Fort Lauderdale, USA, May 2005
4. Tanta University 8th Annual Congress, Egypt, April 2003
5. Mansoura University 6th Annual Congress, Egypt, September 2002
6. Tanta University 7th Annual Congress, Egypt, April 2002
7. ARVO, Fort Lauderdale, USA, May 2001
8. ARVO, Fort Lauderdale, USA, May 2000
9. Tanta University 4th Annual Congress, Egypt, April 1999

Courses

1. Statistics course in the UCL. 2006
2. Ensembl course in the UCL 2005
3. Bioinformatics course in the UCL 2005
4. Postgraduate intensive course on clinical immunology collaboration with immunology department, Saint Louise Hospital, Paris 7 University. 14-24 September, 2002.

5. Workshop on clinical immunology collaboration with immunology department, Saint Louise Hospital, Paris 7 University. HLA typing from serology to sequencing. 28, September- 1 October, 2002

Memberships

1. ARVO
1. The Egyptian medical syndicate: registration No. 118245
2. Clinical pathologists Society of Egypt: registration No. 4055

Special distinctions/ Prizes

I have been nominated a grant for the PhD in 2004.

Personal interests

I am very keen to regularly walk and run to keep fit and for relaxation. Also, I quite enjoy reading novels as well as learning new IT skills. I additionally enjoy working as a volunteer helper and to fulfil my ambition I have been participating as a parent governor in Bentwroth primary school since March 2007.

References

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ABSTRACT

Autosomal recessive retinitis pigmentosa (arRP) is one the commonest forms of monogenic retinal degeneration (RD). To date 24 loci have been implicated in the pathogenesis of arRP. The genes for five of these loci (RP22, RP25, RP28, RP29 and RP30), still remain to be identified. This thesis mainly focused on the cloning of a major gene (RP25); however identifying novel loci for recessive RP constituted a significant objective.

Originally the RP25 locus was mapped to chromosome 6p12.1-q15, a region that spans 34 Mb, by our collaborators in Seville in seven Spanish families. Initially, a whole-genome scan in these families was undertaken using GeneChip 10K array. The data obtained confirmed the initial findings of linkage to the RP25 region. To date, 61 out of 111 genes within the interval (~55%) have been excluded as disease causing by direct sequence analysis. A large number of single nucleotide polymorphisms (SNPs), of which a significant percentage was novel were identified. We have also postulated that both RP25 and Leber congenital amaurosis 5 (LCA5), a severe form of RD, could be due to the same genetic defect since they genetically overlap. Therefore, seventeen LCA families were genotyped to identify new LCA5 families that may further refine the RP25 interval by identifying novel crossovers. However, the gene for LCA5 has been recently cloned and sequence analysis of the RP25 families rules out this gene as causative of RP25.

To investigate if copy number variations (CNVs) exist within the RP25 interval, a comparative genome hybridisation (CGH) was performed on one of the RP25 families (RP5). A clone from the tiling path, chr6tp-19C7, within 6q12 was observed

to be deleted in all affected members of this family indicating that one of the genes within this interval could be responsible for the RP25 phenotype.

A novel approach utilising the 10K GeneChip for identifying the disease locus in three non-consanguineous Chinese families with arRP was implemented. The studied families were probably linked to the RP25 locus; proposing that this approach could be a useful tool for genetic mapping in cases of rare and genetically heterogeneous recessive traits.

Finally, in parallel, a genomewide linkage search in a consanguineous family with arRP was undertaken. Linkage to a 10-cM interval on chromosome 10q23.1-23.3 was observed where a good candidate gene, protocadherin-21 (*PCDH21*), is located. A homozygous 1-bp deletion was identified in this family in addition to two other novel mutations in two different patients raising the possibility that *PCDH21* is likely to be a novel gene for RP.