

EUROPEAN
CURRICULUM VITAE
FORMAT



PERSONAL INFORMATION

Name Sonia Abdelhak
Address BP74; 13 Place Pasteur 1002, Tunis-Tunisia
Telephone +21671849110 (direct line) +21698364000 (mobile)
Fax +21671791833
E-mail sonia.abdelhak@pasteur.rns.tn, sonia.abdelhak@hexabyte.tn

Nationality Tunisian

Date of birth July 30th 1964

WORK EXPERIENCE

- Dates (from – to) **September 1997-September 2004**
• Name and address of employer Ministry of Health, Institut Pasteur de Tunis
• Type of business or sector Academic Research
• Occupation or position held Biologist, Head of Research Group
• Main activities and responsibilities Head of the Sequencing Core Facility.
Research activity, Field: Human Molecular Genetics, Principal Investigator.
- Dates (from – to) **September 2004-**
• Name and address of employer Ministry of Health, Institut Pasteur de Tunis
• Type of business or sector Academic Research
• Occupation or position held Principal Biologist, Head of Research Unit "Molecular Investigation of Genetic Orphan Diseases".
Head of the Sequencing Core Facility.
Head of the Genetic Testing Unit.
• Main activities and responsibilities - Research activity, Field: Human Molecular Genetics, Supervisor of 15 PhD and supervised 20 Master students .
- Contribution to postgraduate teaching programs (Human Molecular Genetics and Introductory Courses of Bioinformatics Faculty of Science of Tunis, Faculty of Medicine of Tunis and Faculty of Medicine of Sfax, Tunisia). Contribution to undergraduate teaching program (Molecular Biology, Faculty of Science of Nouakchott, Mauritania)
- Genetic Testing (Paternity Testing)
- Independent Expert for CNEAR (National Committee for Research Activity Evaluation) and E.C. (FP6-INCO projects evaluation *expert N: EX2002B045663*)

EDUCATION AND TRAINING

- Dates (from – to) **September 1987 to February 1989**

<ul style="list-style-type: none"> • Name and type of organisation providing education and training • Principal subjects/occupational skills covered • Title of qualification awarded 	<p>Faculte des Sciences de Tunis, Tunisia</p> <p>Production and characterisation of monoclonal antibodies against Leishmania infantum. Immunology</p> <p>Master in Science</p>
<ul style="list-style-type: none"> • Dates (from – to) • Name and type of organisation providing education and training • Principal subjects/occupational skills covered • Title of qualification awarded 	<p>1990-1991</p> <p>University of Paris VII, France</p> <p>Generation of DNA probes in the vicinity of the Spinal Muscular Atrophies locus by Alu-PCR amplification on somatic hybrid cells. Human Genetics</p> <p>Master in Science</p>
<ul style="list-style-type: none"> • Dates (from – to) • Name and type of organisation providing education and training • Principal subjects/occupational skills covered • Title of qualification awarded 	<p>1989-1992</p> <p>Faculte des Sciences de Tunis, Tunisia</p> <p>Genetic mapping and prenatal diagnosis of infantile spinal muscular atrophies. Human Molecular Genetics</p> <p>Thesis in Science</p>
<ul style="list-style-type: none"> • Dates (from – to) • Name and type of organisation providing education and training • Principal subjects/occupational skills covered • Title of qualification awarded 	<p>1992-1997</p> <p>Faculte des Sciences de Tunis, Tunisia</p> <p>Refined mapping and cloning of the gene responsible for branchio-oto-renal syndrome. Human Molecular Genetics</p> <p>Sate Doctorate in Science</p>
<ul style="list-style-type: none"> • Dates (from – to) • Name and type of organisation providing education and training • Principal subjects/occupational skills covered • Title of qualification awarded 	<p>1992-1997</p> <p>University of Paris VII, France</p> <p>Refined mapping and cloning of the gene responsible for branchio-oto-renal syndrome. PhD</p>

PERSONAL SKILLS AND COMPETENCES

Acquired in the course of life and career but not necessarily covered by formal certificates and diplomas.

- .Genomics, Bioinformatics, Biotechnology, Genetic counseling, Training through research activity for undergraduate and graduate students, capacity building, management of research activity, management of a core facility, management of a genetic testing service, evaluation of research projects, ethical issues related to genetic testing in developing countries..

MOTHER TONGUE **Arabic**

OTHER LANGUAGES

Reading skills, Writing skills, Verbal skills	French Very good
Reading skills, Writing skills, Verbal skills	English Very good
Reading skills, Writing skills, Verbal skills	Italian working knowledge
Reading skills, Writing skills, Verbal skills	German working knowledge
• Reading skills, Writing skills, Verbal skills	Spanish working knowledge
ORGANISATIONAL SKILLS AND COMPETENCES <i>Coordination and administration of people, projects and budgets; at work, in voluntary work (for example culture and sports) and at home, etc.</i>	Coordination of research groups, organization of seminars and workshops. Coordinator of the following research projects: - Institutional project on Fanconi Anemia: Contrat IPT-IMM14 (PI, 30.000 TD). - PI and coordinator of European projects: organization of 3 international workshops on health of populations in the Mediterranean (ICA3-2000-50020 60.000 Euros and FP6-510667 152.000 Euros). - PI of a workpackage WHO/TDR ID-A11032 (35.000 \$). - Organization of a workshop on Quantitative Real Time PCR, Direction des Affaires Internationales (DAI) Institut Pasteur de Paris (PI, 15.000 Euros). - Coordination of a research Unit (medium budget per year 35.000 TD/year/3 years).
TECHNICAL SKILLS AND COMPETENCES <i>With computers, specific kinds of equipment, machinery, etc.</i>	Current work with computer environment pc, mac, linux, sequencing and genotyping platform
ARTISTIC SKILLS AND COMPETENCES <i>Music, writing, design, etc.</i>	Gardening, cooking.
OTHER SKILLS AND COMPETENCES <i>Competences not mentioned above.</i>	Participation to International courses European School of Medical Genetics (Third course) April 1-7, 1990; Sestri Levante, Genoa, Italy. Cours de Génétique et Biologie Cellulaires 16-09 to 27-10-1990, Institut Pasteur de Paris, France. Cours de Microbiologie Générale 6-10 to 17-12-1993, Institut Pasteur de Paris, France. Practical Course in Bioinformatics: Computer methods in molecular biology July 3-10, 1998, International Center for Genetic Engineering and Biotechnology (ICGEB), Trieste, Italy. Human Genome Analysis: Genetic analysis of multifactorial diseases, 21st Summer School, July 25-31, 1998, Wellcome Trust Genome Campus, Hinxton, U.K. PROMEDACCESS training held in Tunis, 22-23 May 2007.
DRIVING LICENCE(S)	Yes

A N N E X E
L i s t o f

p u b l i c a t i o n s :

1. Melki J, **Abdelhak S**, Sheth P, Bachelot MF, Burlet P, Marcadet A, Aicardi J, Barois A, Carriere JP, Fardeau M, et al. Gene for chronic proximal spinal muscular atrophies maps to chromosome 5q. *Nature* 190;344:767-8.
2. Melki J, Sheth P, **Abdelhak S**, Burlet P, Bachelot MF, Lathrop MG, Frezal J, Munnich A. Mapping of acute (type I) spinal muscular atrophy to chromosome 5q12-q14. *Lancet* 1990;336:271-3.
3. **Abdelhak S**, Melki J, Bachelot MF, Burlet P, Sheth P, Frezal J, Munnich A. A PstI polymorphism at the D5S39 locus. *Nucleic Acids* 1990;18:5580.
4. Mattei MG, Melki J, Bachelot MF, **Abdelhak S**, Burlet P, Frezal J, Munnich A. In situ hybridization of two markers closely flanking the spinal muscular atrophy gene to 5q12-q13.3. *Cytogenet Cell Genet* 1991;57:112-3.
5. Sheth P, **Abdelhak S**, Bachelot MF, Burlet P, Masset M, Hillaire D, Clerget-Darpoux F, Frezal J, Lathrop GM, Munnich A, et al. Linkage analysis in spinal muscular atrophy, by six closely flanking markers on chromosome 5. *Am J Hum Genet* 1991;48:764-8.
6. Melki J, **Abdelhak S**, Burlet P, Raclin V, Kalan J, Spiegel R, Gilgenkrantz S, Philip N, Chauvet ML, Dumez Y, et al. Prenatal prediction of Werdnig-Hoffmann disease using linked polymorphic DNA probes. *J Med Genet* 1992;29:171-4.
7. Melki J, Burlet P, Clermont O, Pascal F, Paul B, **Abdelhak S**, Sherrington R, Gurling H, Nakamura Y, Weissenbach J, et al. Refined linkage map of chromosome 5 in the region of the spinal muscular atrophy gene. *Genomics* 1993;15:521-4.
8. Burlet P, **Abdelhak S**, Pascal F, Clermont O, Paul B, Munnich A, Melki J. Trinucleotide repeat polymorphism at the D5S556 locus. *Hum Mol Genet* 1993;2:1328.
9. **Abdelhak S**, Louzir H, Timm J, Blel L, Benlasfar Z, Lagranderie M, Gheorghiu M, Dellagi K, Gicquel B. Recombinant BCG expressing the leishmania surface antigen Gp63 induces protective immunity against *Leishmania major* infection in BALB/c mice. *Microbiol* 1995;141:1585-92.
10. Kalatzis V, **Abdelhak S**, Compain S, Vincent C, Petit C. Characterization of a translocation-associated deletion defines the candidate region for the gene responsible for branchio-oto-renal syndrome. *Genomics* 1996;34:422-5.
11. **Abdelhak S**, Kalatzis V, Heilig R, Compain S, Samson D, Vincent C, Weil D, Cruaud C, Sahly I, et al. A human homologue of the *Drosophila* eyes absent gene underlies branchio-oto-renal (BOR) syndrome and identifies a novel gene family. *Nature Genet* 1997;15:157-64.
12. Vincent C, Kalatzis V, **Abdelhak S**, Chaib H, Compain S, Helias J, Vanecloo F, Petit M. BOR and BO syndromes are allelic defects of EYA1. *Eur J Hum Genet* 1997;5:242-6.
13. **Abdelhak S**, Kalatzis V, Heilig R, Compain S, Samson D, Vincent C, Levi-Acobas F, Cruaud C, et al. Clustering of mutations responsible for branchio-oto-renal (BOR) syndrome in the eyes absent homologous region (eyeHR) of EYA1. *Hum Mol Genet* 1997;6:2247-55.
14. Hardelin, J.P., Soussi-Yanicostas N., Levilliers J., Kalatzis V., **Abdelhak S.**, Cohen-Salmon M. and Petit C. Molecular approach to the pathogenesis of renal anomalies in Kallmann's syndrome and in the branchio-oto-renal syndrome. *Adv. Nephrol. Necker Hosp.* 1998; 28:419-28.
15. El Kares R., **Abdelhak S.**, Dellagi K. Gene structure and characterisation of the promoter region of IL-18. *Archs. Inst. Pasteur Tunis* 2000; 77:57-60.
16. Elloumi-Zghal H, Barbouche MR, Chemli J, Béjaoui M, Harbi A, Snoussi N, **Abdelhak S** and K Dellagi. Clinical and Genetic heterogeneity of inherited autosomal recessive disseminated Bacille Calmette-Guerin infection. *J Infect Dis* 2002; 185:1468-75.
17. Houman MH, Hamzaoui-B'Chir S, Ben Ghorbel I, Lamloum M, Ben Ahmed M, **Abdelhak S**, Miled M. Neurologic manifestations of Behcet's disease: analysis of a series of 27 patients. *Rev Med Interne.* 2002; 23:592-606.
18. Ben Ahmed M, Houman H, **Abdelhak S**, Ben Ghorbel I, Miled M, Dellagi K, Louzir H. MICA transmembrane region polymorphism and HLA B51 in Tunisian Behcet's disease patients. *Adv Exp Med Biol.* 2003;528:225-8.
19. C. Bouchlaka, **S. Abdelhak**, A. Amouri, H. Ben Abid, S. Hadiji, M. Frikha, T. Ben Othman, F. Amri, H. Ayadi, M. Hachicha, A. Rebaï, A. Saad, K. Dellagi & The Tunisian Fanconi Anaemia Study Group. Fanconi anaemia in Tunisia: high Prevalence of group A and identification of new *FANCA* mutations. *J Hum Genet.* 2003;48:352-61.
20. C. Charfeddine, M. Mokni, R. Ben Mousli, R. Elkares, C. Bouchlaka, S. Boubaker, S. Ghedamsi, D. Baccouche, A. Ben Osman, K. Dellagi, **S. Abdelhak**. Novel missense mutation

- in the gene encoding SLURP-1 in patients with Mal de Meleda from Northern Tunisia. *Br J Dermatol.* 2003 ;149:1108-15.
21. Mokni M, Charfeddine C, Ben Mously R, Baccouche D, Kaabi B, Ben Osman A, Dellagi K, **Abdelhak S**. Heterozygous manifestations in female carriers of Mal de Meleda. *Clin Genet.* 2004;65:244-6.
 22. Bouchlaka C, Ben Othman T, Aissaoui L, Elloumi M, Amouri A, Ben Abid A, Elloumi H, Hadiji S, Slama H, Makni H, Saad A, **Abdelhak S**, Dellagi K & The Tunisian Fanconi Anemia Study Group. Fanconi anemia: Contribution of molecular analysis for the identification of bone marrow graft donor and study of chimerism in grafted patients. *Genet Test.* 2004 ;8:268-75.
 23. Bouchlaka C, **Abdelhak S**, Dellagi K; Le Groupe d'Etude de la Maladie de Fanconi en Tunisie. [Molecular study of Fanconi anemia in Tunisia] *Tunis Med.* 2004 ;82:402-10. Erratum in: *Tunis Med.* 2004; 82:716.
 24. I. Zghal-Mokni, I. Arfa, H. Elloumi-Zghal, A. Abid, C. Amrouche-Rached, B. Kaabi, S. Chakroun, S. Blousa-Chabchoub, S. Gaïgi, S. Ayed, A. Jeddi, K. Dellagi, **S. Abdelhak**. Association study between diabetic retinopathy and aldose reductase gene polymorphism in Tunisians. *J. Fr. Ophtalmol* 2005; 28: 386-390.
 25. Souissi A, Ben Tekaya N, Mourad M, Zribi H, Jerbi E, **Abdelhak S**, Boubaker S, Ben Osman Dahri A. [Darier's disease associated with pemphigus vulgaris: coexistence of two acantholytic mechanisms]. *Ann Dermatol Venereol.* 2006 Jan;133:63-5.
 26. Charfeddine C, Monastiri K, Mokni M, Laadjimi A, Kaabachi N, Perin O, Nilges M, Kassas S, Keirallah M, Guediche MN, Kamoun MR, Tebib N, Ben Dridi MF, Boubaker S, Ben Osman A, **Abdelhak S**. Clinical and mutational investigations of tyrosinemia type II in Northern Tunisia: identification and structural characterization of two novel TAT mutations. *Mol Genet Metab.* 2006 Jun;88:184-91
 27. Charfeddine C, Mokni M, Kassas S, Zribi H, Bouchlaka C, Boubaker S, Rebai A, Ben Osman A, **Abdelhak S**. Further evidence of the clinical and genetic heterogeneity of recessivetransgressive PPK in the Mediterranean region. *J Hum Genet.* 2006;51:841-5.
 28. El Kares R, Barbouche MR, Elloumi-Zghal H, Bejaoui M, Chemli J, Mellouli F, Tebib N, Abdelmoula MS, Boukthir S, Fitouri Z, M'rad S, Bouslama K, Touiri H, **Abdelhak S**, Dellagi MK. Genetic and mutational heterogeneity of autosomal recessive chronic granulomatous disease in Tunisia. *J Hum Genet.* 2006;51:887-95.
 29. Guermazi S, Elloumi-Zghal H, Ben Hassine L, Romani S, Khalfallah N, **Abdelhak S**, Dellagi K. [Homozygous antithrombin type HBS deficiency; a family study.] *Pathol Biol (Paris).* 2006 Nov 28;
 30. Bouchlaka C, Maktouf C, Mahjoub B, Ayadi A, Sfar MT, Sioud M, Gueddich N, Belhadjali Z, Rebai A, **Abdelhak S**, Dellagi K. Genetic heterogeneity of megaloblastic anaemia type 1 in Tunisian patients. *J Hum Genet.* 2007;52:262-70
 31. Arfa I, Abid A, Malouche D, Ben Alaya N, Azegue TR, Mannai I, Zorgati MM, Ben Rayana MC, Ben Ammar S, Blousa-Chabchoub S, Ben Romdhane H, Zouari B, Dellagi MK, **Abdelhak S**. Familial aggregation and excess maternal transmission of Type 2 diabetes in Tunisia. *Post Grad. Med. J.* 2007; 83:348-51.

Reports:

Abdelhak S: Rare diseases and orphan drugs in Northern Africa in "PEOPLE WITH RARE DISEASES – NO LONGER ALONE IN THE WORLD" REPORT OF THE SIXTH EPPOSI-WORKSHOP ON PARTNERING FOR RARE DISEASE THERAPY DEVELOPMENT, page 140-144 edited by Pete Wrobel and published by: European Platform for Patients' Organisations, Science and Industry (EPPOSI) committee