

Short CV Nadem Soufir

Civil Status

Soufir Nadem, born 02.16.1964, MD, PhD

Professional addresses:

Genetic Department, Bichat Hospital, AP-HP, 46 rue Henri Huchard, 75018, Paris, France

Skin Research Center. INSERM U976 Hopital Saint-Louis Pavillon Bazin

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Main Diplomas

1982-1989 Medical School

1994-1995 Master in Human Genetics, University Paris 6

1996-1997 MD of Dermatology, PhD of Medicine, University Paris 6

1999-2000 PhD Human Genetics, University Paris 6

2000-2001: Post Doctoral Training at the Imperial Cancer Research Foundation, London

2004 Habilitation to Direct Research

2004 Associate Professor in Genetics, Bichat Hospital, AP-HP, University Paris 7

2007 Pedagogy Diploma

Teaching responsibilities at the University

2002-2007 Biochemistry, University Paris 7

2004-2013 Master of Genetics, University Paris 7

Since 2004 Human Genetics at Medical School, University Paris 7

Since 2010 Creation of inter university diploma dedicated to predisposition to cancer

Hospital activities

- 2001-2004 Assistant professor of Genetics

- Since 2004 -2011 Associate Professor first class in Genetics

- Since 2011: Professor of Genetics

- **Since 2011:** Coordination of the organization of oncogenetics consultations of North Hospital Group of Paris (Bichat, Beaujon, Saint- Louis). Genetic predisposition to skin cancers, gastrointestinal cancers (colon, pancreas) and urological cancers.

- Molecular Diagnosis in oncogenetics (predisposition to skin cancer and digestive cancer) in the Genetic Department of Bichat Hospital, Paris

Member of scientific societies and networks

Since 2002 National Network of predisposition to skin cancer

Since 2003 Scientific coordination of MelanCohort, a melanoma network of Paris region

Since 2004 French Society of Human Genetics

Since 2004 European Society of Human Genetics

Since 2007 Scientific co-coordinator of National Network Basal Cell Carcinoma Gorlin

Since 2010 International Melanoma Networks: GenoMel and BioGenoMel (Pr Julia Newton Bishop)

Research Activities

- Major Research projects:

- Identification of susceptibility genetic markers to melanoma and basal cell carcinomas.
- Study of prognostic genetic factors in melanoma
- Finding genetic variants associated with susceptibility to vitiligo and alopecia areata
- Identification of genetic markers associated with susceptibility to Verneuil's disease (hidradenitis suppurativa)
- 2002-2010 numerous grants funding APHP (2 CIRCS, one PHRC), the ARC, the French Society of Dermatology
- 2007: Patent "Method for in vitro diagnosis of skin cancer" (Reference: EPR/SA/07-288)
- **Since 2009 Director of the Melanoma Team** integrated within the Research Skin Center, Inserm U976 (Dr A Bensussan), Saint Louis. Attachment to the graduate school B2T.

Scientific awards and grants

1998 Price "René Fauvert" Fund Study and Research Medical Corps Hospital of Paris
2000 Fondation René Touraine, Post doctoral fellowship
2000, 2003 Grants from Assistance Publique des Hopitaux de Paris, Contract Research and Innovation Clinic 200 KE
2005 and 2010 Grant from the Association for Research against Cancer 100 KE
2006-2007 Grant Laboratories Schering Plough Foundation 50 KE
2006 Grant from APHP, MelanCohort PHRC, as a co-investigator 30 KE
2001, 2004, 2005, 2009, 2011 Grant from the French Society of Dermatology 150 KE

Publications

61 original articles in English in peer review journals including 30 in the first or last author, 14 general reviews, articles three books. 62 oral presentations or posters.

Major scientific publications demonstrating his expertise in the project field during the last five years

1. - Guedj M, Bourillon A, Combadières C, Rodero M, Dieudé P, Descamps V, Dupin N, Wolkenstein P, Aegester P, Lebbe C, Basset-Seguin N, Prum B, Saiag P, Grandchamp B, Soufir N; MelanCohort Investigators. Variants of the *MATP/SLC45A2* gene are protective for melanoma in the French population (2008). ***Human Mutation*** 29(9):1154-60
2. - Di Lucca Chrisment J, Guedj M, Lacapere J.J, Bourillon A, Combadieres C, Rodero M, Dieudé P, Descamps V, Dupin N, Wolkenstein P, Aegester P, Lebbé C, Basset-Seguin N, Saiag Ph, Granchamp B, Soufir N Association between variants in the *POLH* gene and melanoma risk (2009). ***Eur J Cancer*** 45(18):3228-36
3. - Soufir N, Ged C, Bourillon A, Austerlitz F, Chemin C, Stary A, Armier J, Pham D, Khadir K,

Roume J, Hadj-Rabia S, Bouadjar B, Taieb A, de Verneuil H, Benchiki H, Grandchamp B, Sarasin A. A prevalent mutation with founder effect in xeroderma pigmentosum group C from north Africa (2010). **J Invest Dermatol.** Jun;130(6):1537-42.

4. - Ibarrola-Villava M, Hu HH, Guedj M, Fernandez LP, Descamps V, Basset-Seguin N, Bagot M, Bensussan A, Saiag P, Farnoli MC, Peris K, Aviles JA, Lluch A, Ribas G, **Soufir N**, MC1R, SLC45A2 and TYR genetic variants involved in melanoma susceptibility in Southern European populations: Results from a Meta-analysis (2012). **Eur J Cancer.** Sep;48(14):2183-91.

5. - Bourillon A, Hu HH, Hetet G, Lacapere JJ, André J, Descamps V, Basset-Seguin N, Ogbah Z, Puig S, Saiag P, Bagot M, Bensussan A, Grandchamp B, Dumaz N, **Soufir N** (2012). Genetic variation at KIT locus may predispose to melanoma. **Pigment Cell Melanoma Res.** Oct.

Major scientific publications in indexed journals and peer-reviewed with international committees or any other significant publications during the last five years (titles and references)

1. Guedj M, Bourillon A, Combadières C, Rodero M, Dieude P, Descamps V, Dupin N, Wolkenstein P, Aegerter P, Lebbe C, Basset-Seguin N, Prum B, Saiag P, Grandchamp B, **Soufir N**; MelanCohort Investigators. Variants of the MATP/SLC45A2 gene are protective for melanoma in the French population. **Hum Mutat.** 2008 Sep;29(9):1154-60

2. **Soufir N**, Grandchamp B, Basset-Seguin N. New trends in the susceptibility to melanoma. **Cancer Treat Res.** 2009;146:213-23. doi: 10.1007/978-0-387-78574-5_19.

3. Di Lucca J, Guedj M, Lacapere JJ, Farnoli MC, Bourillon A, Dieude P, Dupin N, Wolkenstein P, Aegerter P, Saiag P, Descamps V, Lebbe C, Basset-Seguin N, Peris K, Grandchamp B, **Soufir N**. Variants of the xeroderma pigmentosum variant gene (POLH) are associated with melanoma risk. **Eur J Cancer.** 2009 Dec;45(18):3228-36

4. Di Lucca J, Guedj M, Descamps V, Bourillon A, Dieude P, Saiag P, Wolkenstein P, Dupin N, Lebbe C, Basset-Seguin N, Grandchamp B, **Soufir N**. Interactions between ultraviolet light exposure and DNA repair gene polymorphisms may increase melanoma risk. **Br J Dermatol.** 2010 Apr;162(4):891-3

5. **Soufir N**, Ged C, Bourillon A, Austerlitz F, Chemin C, Stary A, Armier J, Pham D, Khadir K, Roume J, Hadj-Rabia S, Bouadjar B, Taieb A, de Verneuil H, Benchiki H, Grandchamp B, Sarasin A. A prevalent mutation with founder effect in xeroderma pigmentosum group C from north Africa. **J Invest Dermatol.** 2010 Jun;130(6):1537-42

6. Jacobelli S, **Soufir N**, Lacapere JJ, Regnier S, Bourillon A, Grandchamp B, Hetet G, Pham D, Palangie A, Avril MF, Dupin N, Sarasin A, Gorin I. Xeroderma pigmentosum group C in a French Caucasian patient with multiple melanoma and unusual long-term survival. **Br J Dermatol.** 2008 Sep;159(4):968-73.

7. Bourillon A, Hu HH, Hetet G, Lacapere JJ, André J, Descamps V, Basset-Seguin N, Ogbah Z, Puig S, Saiag P, Bagot M, Bensussan A, Grandchamp B, Dumaz N, **Soufir N**. Genetic variation at KIT locus may predispose to melanoma. **Pigment Cell Melanoma Res.** 2013 Jan;26(1):88-96

8. Lalou C, Scamuffa N, Mourah S, Plassa F, Podgorniak MP, **Soufir N**, Dumaz N, Calvo F, Basset-Seguin N, Khatib AM. Inhibition of the proprotein convertases represses the invasiveness of human primary melanoma cells with altered p53, CDKN2A and N-Ras genes. **PLoS One.** 2010 Apr 9;5(4):e9992

9. Descamps V, Basset-Seguin N, **Soufir N**. ISET device: a useful tool to answer some questions

- about the biologic behavior of melanocytic nevus cells. *Arch Dermatol.* 2010 Oct;146(10):1156-7
10. Hu HH, Guedj M, Descamps V, Jouary T, Bourillon A, Ezzedine K, Taieb A, Bagot M, Bensusan A, Saiag P, Grandchamp B, Basset-Seguin N, **Soufir N.** Assessment of tyrosinase variants and skin cancer risk in a large cohort of French subjects. *J Dermatol Sci.* 2011 Nov;64(2):127-33
 11. Spica T, Farnoli MC, Hetet G, Bertrand G, Formicone F, Descamps V, Wolkenstein P, Dupin N, Lebbe C, Basset-Seguin N, Saiag P, Cambien F, Grandchamp B, Peris K, **Soufir N.** EDNRB gene variants and melanoma risk in two southern European populations. *Clin Exp Dermatol.* 2011 Oct;36(7):782-7
 12. Davies JR, Randerson-Moor J, Kukalizch K, Harland M, Kumar R, Madhusudan S, Nagore E, Hansson J, Hv̄diom V, Ghiorzo P, Gruis NA, Kanetsky PA, Wendt J, Pjanova D, Puig S, Saiag P, Schadendorf D, **Soufir N.**, Okamoto I, Affleck P, Garcia-Casado Z, Ogbah Z, Ozola A, Queirolo P, Sucker A, Barrett JH, van Doorn R, Bishop DT, Newton-Bishop J. Inherited variants in the MC1R gene and survival from cutaneous melanoma: a BioGenoMEL study. *Pigment Cell Melanoma Res.* 2012 May;25(3):384-94
 13. Hadj-Rabia S, Oriot D, **Soufir N.**, Dufresne H, Bourrat E, Mallet S, Poulhalon N, Taieb A, Catteau B, Sarasin A, Bodemer C. Unexpected extradermatological findings in 31 -Xeroderma Pigmentosum type C patients. *Br J Dermatol.* 2012 Dec 19
 14. Bourillon A, Hu HH, Hetet G, Lacapere JJ, André J, Descamps V, Basset-Seguin N, Ogbah Z, Puig S, Saiag P, Bagot M, Bensusan A, Grandchamp B, Dumaz N, **Soufir N.** Genetic variation at KIT locus may predispose to melanoma. *Pigment Cell Melanoma Res.* 2013 Jan;26(1):88-96
 15. Puntervoll HE, Yang XR, Vetti HH, Bachmann IM, Avril MF, Benfodda M, Catricala C, Dalle S, Duval-Modeste AB, Ghiorzo P, Grammatico P, Harland M, Hayward NK, Hu HH, Jouary T, Martin-Denavit T, Ozola A, Palmer JM, Pastorino L, Pjanova D, **Soufir N.**, Steine SJ, Stratigos AJ, Thomas L, Tinat J, Tsao H, Veinalde R, Tucker MA, Bressac-de Paillerets B, Newton-Bishop JA, Goldstein AM, Akslen LA, Molven A. Melanoma prone families with CDK4 germline mutation: phenotypic profile and associations with MC1R variants. *J Med Genet.* 2013 Feb 5