Sandra D'Alfonso

Curriculum vitae

PERSONAL DATA

Sandra D'Alfonso

Born at Torino, 17-05-1965

Associate Professor of Human Genetics

Department of Health Sciences and IRCAD (Interdisciplinary Research Center of Autoimmune Diseases)

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BIO AND EDUCATION

-1988: degree in Biological Sciences with the score 110/110, Torino University, Italy.
-1989-1991 fellow of different Italian Associations ("Associazione per la Cura e la Prevenzione dei tumori in Piemonte", Comitato Regionale Piemontese Gigi Ghirotti", Regione Piemonte U.S.S.L. Torino VIII
- November 1989 - February 1990 Attended the laboratory of Cellular Biochemistry directed by Prof. H. Ploegh at "The Netherlands Cancer Institute" Amsterdam (NL)
- 1991- 1995 PhD student of Human Genetics, Torino University, Italy
- Author of 144 papers on International indexed (IF) in the field of HLA immunogenetics and of the genetics of autoimmune and neurodegerative diseases. H Index (Scopus)=34 . Total citations:5499, Total Impact Factor: 810,049

Coordinator of PROGEMUS (PROgnostic GEnetic factors in MUltiple Sclerosis), a cross-sectional multicentric study including all patients affected by MS (Multiple Sclerosis) consecutively examined in 10 Italian MS Centres
Coordinator of GENLES, a network of 6 Italian Centers for the genetic study of Systemic Lupus Erythematosus (SLE)

Co-founder of the SLAGEN Consortium, an Italian multicentric Consortium for the genetics of Amythophic Lateral Sclerosis (ALS)

Member of Member of the IMSGC (International Multiple Sclerosis Genetics) strategic Group, and the International network "Biolupus", for the genetic study of Systemic Lupus Erythematosus (SLE)

UNIVERSITY CAREER

2004-	Associate Professor of Medical Genetics (MED/03), Università del Piemonte Orientale
1996-2004	Assistant Professor of Medical Genetics (MED/03), Università del Piemonte Orientale
1992-1995	PhD Student, Università di Torino
2014	"Abilitation" to full Professor of Medical Genetics (MED/03)

UNIVERSITY POSITIONS

2010-	Member of "Giunta di Dipartimento" (Dipartimento di Scienze della Salute former Dipartimento di Scienze Mediche), Università del Piemonte Orientale
2014-	Component of "Gruppo del Riesame" for the School of Medicine Degree , Università del Piemonte Orientale
2014-	Component of the Library Commission for the School of Medicine, Università del Piemonte Orientale
2014-	Delegate of the School of Medicine for the Job Placement Commission, Università del Piemonte Orientale
2016-	Delegate of the Rector for the Piedmont Regional Commission "Alternanza Scuola Lavoro", Università del Piemonte Orientale
2010-	Component of the Technical Pedagogical Commission (Commissione Tecnica di Programmazione didattico pedagogica), School of Medicine Degree, Università del Piemonte Orientale
2015	Delegate of the Dipartimento di Scienze della Salute as " Referente del **Presidio di Qualità delle sedi per la Formazione",** Università del Piemonte Orientale

SCIENTIFIC POSITIONS

2012-	Member of the Techical Scientific Commitee of IRCAD (Interdisciplinary Research Center on Autoimmune Disease), Università del Piemonte Orientale
2014-2016	President of IRCAD (Interdisciplinary Research Center on Autoimmune Disease), Università del Piemonte Orientale
2014-	Member of Scientific of FISM (Italian Foundation for Multiple Sclerosis)
1999-	Member of the Editorial Board of the International Journal (Nature Publishing Group) " Genes and Immunity"
2008-	Member of the IMSGC (International Multiple Sclerosis Genetics) strategic Group

	Member of the Italian Society of Human Genetics “Societa' Italiana Genetica Umana" (SIGU)

MAIN FIELDS OF INTEREST

1. Genetics of Autoimmune diseases
2. Genetics of Neurodegenerative diseases

CURRENT ISSUES OF RESEARCH

1. **Genetics of Autoimmune diseases:: Genetics of Multiple Sclerosis (MS)**

Abstract;. Since 1997 coordinator or co-investigator of national and international studies leading to the publication of 56 papers and focused on a) genome-wide linkage analysis in Italian MS multiplex families: b) fine mapping of the HLA region with the identification of novel risk and protective factors c) Genome Wide Association Studies (GWAS) with the identification of 110 genes associated with MS susceptibility d) identification of rare MS associated variants through Next Generation Sequencing and functional studies; e) association of the genetic factors with MS clinical features with the identification of genes associated with oligoclonal Immunoglobulin in liquor and age at MS onset

2. **Genetics of Autoimmune diseases: Systemic Lupus Erythematosus (SLE)**

Abstract – 500 ch. max. Since 1995 coordinator or co-investigator of national and international studies leading to the identification of 80 SLE associated genes and the functional characterization of a subset of them including Osteopontin, BANK1, kallikreins. These studied leaded to publication of 38 papers

3. **Genetics of Amyotrophic Lateral Sclerosis (ALS)**

Abstract – 500 ch. max. Since 2006 coordinator or co-investigator of national and international studies, which a) defined the mutational spectrum of ALS patients in Italy; b) identified new ALS causing genes, c) defined the role of common variants in the genetic susceptibility of ALS sporadic cases, d) investigated the role of tandem repeats through the coordination of a multicentric project focused on a genome wide approach and whole genome sequencing. A total of 23 papers have been published

CURRENT FUNDED PROJECTS

BANDO: FISM (Fondazione Italiana per la Sclerosi Multipla)

TITOLO: Genomic and functional evaluation of the role of the TNFSF14-TNFRSF14 pathway in susceptibility to multiple sclerosis
ROLE: Coordinator of the project

DURATA: 24 mesi (2016)

QUOTA:185K

BANDO :Ministero della Salute Progetto di Ricerca Finalizzata

TITOLO: An integrated genomic, transcriptomic and epigenomic approach in multiplex multiple sclerosis families. RF-2011-02350347

Role: Research Unit responsible

DURATA: 36 MESI (2014)

QUOTA: 90K

PROGETTO: AriSLA (Fondazione Italiana di ricerca per la SLA – Sclerosi Laterale Amiotrofica)

TITOLO: "Genome-wide analysis of DNA tandem repeats in ALS: the role of Repeat-ome"

ROLE: Coordinator of the project

DURATA: 24 mesi (2014)

QUOTA: 170K

PROGETTO: Fondazione CARIPLO-2010 (N. 2010-0728)

TITOLO: Search for genes involved in the susceptibility to Multiple Sclerosis (MS): a replication and follow-up study of genetic markers identified in an Italian case-control whole genome association study).

ROLE: Coordinator of the project

2011-2016

QUOTA:300K

RUOLO: PI del progetto

TOP FIVE PAPERS

International Multiple Sclerosis Genetics Consortium (IMSGC), Beecham AH, Patsopoulos NA, Xifara DK, Davis MF, Kemppinen A, Cotsapas C, Shah TS, Spencer C, Booth D, Goris A, Oturai A, Saarela J, Fontaine B, Hemmer B, Martin C, Zipp F, **D'Alfonso** S, Martinelli-Boneschi F, Taylor B, Harbo HF, Kockum I, Hillert J, Olsson T, Ban M, Oksenberg JR, Hintzen R, Barcellos LF;

Wellcome Trust Case Control Consortium 2 (WTCCC2); International IBD Genetics Consortium (IIBDGC), Agliardi C, Alfredsson L, Alizadeh M, Anderson C, Andrews R, Søndergaard HB, Baker A, Band G, Baranzini SE, Barizzone N, Barrett J, Bellenguez C, Bergamaschi L, Bernardinelli L, Berthele A, Biberacher V, Binder TM, Blackburn H, Bomfim IL, Brambilla P, Broadley S, Brochet B, Brundin L, Buck D, Butzkueven H, Caillier SJ, Camu W, Carpentier W, Cavalla P, Celius EG, Coman I, Comi G, Corrado L, Cosemans L, Cournu-Rebeix I, Cree BA, Cusi D, Damotte V, Defer G, Delgado SR, Deloukas P, di Sapio A, Dilthey AT, Donnelly P, Dubois B, Duddy M, Edkins S, Elovaara I, Esposito F, Evangelou N, Fiddes B, Field J, Franke A, Freeman C, Frohlich IY, Galimberti D, Gieger C, Gourraud PA, Graetz C, Graham A, Grummel V, Guaschino C, Hadjixenofontos A, Hakonarson H, Halfpenny C, Hall G, Hall P, Hamsten A, Harley J, Harrower T, Hawkins C, Hellenthal G, Hillier C, Hobart J, Hoshi M, Hunt SE, Jagodic M, Jelčić I, Jochim A, Kendall B, Kermode A, Kilpatrick T, Koivisto K, Konidari I, Korn T, Kronsbein H, Langford C, Larsson M, Lathrop M, Lebrun-Frenay C, Lechner-Scott J, Lee MH, Leone MA, Leppä V, Liberatore G, Lie BA, Lill CM, Lindén M, Link J, Luessi F, Lycke J, Macciardi F, Männistö S, Manrique CP, Martin R, Martinelli V, Mason D, Mazibrada G, McCabe C, Mero IL, Mescheriakova J, Moutsianas L, Myhr KM, Nagels G, Nicholas R, Nilsson P, Piehl F, Pirinen M, Price SE, Quach H, Reunanen M, Robberecht W, Robertson NP, Rodegher M, Rog D, Salvetti M, Schnetz-Boutaud NC, Sellebjerg F, Selter RC, Schaefer C, Shaunak S, Shen L, Shields S, Siffrin V, Slee M, Sorensen PS, Sorosina M, Sospedra M, Spurkland A, Strange A, Sundqvist E, Thijs V, Thorpe J, Ticca A, Tienari P, van Duijn C, Visser EM, Vucic S, Westerlind H, Wiley JS, Wilkins A, Wilson JF, Winkelmann J, Zajicek J, Zindler E, Haines JL, Pericak-Vance MA, Ivinson AJ, Stewart G, Hafler D, Hauser SL, Compston A, McVean G, De Jager P, Sawcer SJ, McCauley JL. Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis. Nat Genet. 2013 Nov;45(11):1353-60. doi: 10.1038/ng.2770. Epub 2013 Sep 29. PubMed PMID: 24076602; PubMed Central PMCID: PMC3832895.

International Multiple Sclerosis Genetics Consortium; Wellcome Trust Case Control Consortium 2, Sawcer S, Hellenthal G, Pirinen M, Spencer CC, Patsopoulos NA, Moutsianas L, Dilthey A, Su Z, Freeman C, Hunt SE, Edkins S, Gray E, Booth DR, Potter SC, Goris A, Band G, Oturai AB, Strange A, Saarela J, Bellenguez C, Fontaine B, Gillman M, Hemmer B, Gwilliam R, Zipp F, Jayakumar A, Martin R, Leslie S, Hawkins S, Giannoulatou E, **D'Alfonso** S, Blackburn H, Martinelli Boneschi F, Liddle J, Harbo HF, Perez ML, Spurkland A, Waller MJ, Mycko MP, Ricketts M, Comabella M, Hammond N, Kockum I, McCann OT, Ban M, Whittaker P, Kemppinen A, Weston P, Hawkins C, Widaa S, Zajicek J, Dronov S, Robertson N, Bumpstead SJ, Barcellos LF, Ravindrarajah R, Abraham R, Alfredsson L, Ardlie K, Aubin C, Baker A, Baker K, Baranzini SE, Bergamaschi L, Bergamaschi R, Bernstein A, Berthele A, Boggild M, Bradfield JP, Brassat D, Broadley SA, Buck D, Butzkueven H, Capra R, Carroll WM, Cavalla P, Celius EG, Cepok S, Chiavacci R, Clerget-Darpoux F, Clysters K, Comi G, Cossburn M, Cournu-Rebeix I, Cox MB, Cozen W, Cree BA, Cross AH, Cusi D, Daly MJ, Davis E, de Bakker PI, Debouverie M, D'hooghe MB, Dixon K, Dobosi R, Dubois B, Ellinghaus D, Elovaara I, Esposito F, Fontenille C, Foote S, Franke A, Galimberti D, Ghezzi A, Glessner J, Gomez R, Gout O, Graham C, Grant SF, Guerini FR, Hakonarson H, Hall P, Hamsten A, Hartung HP, Heard RN, Heath S, Hobart J, Hoshi M, Infante-Duarte C, Ingram G, Ingram W, Islam T, Jagodic M, Kabesch M, Kermode AG, Kilpatrick

TJ, Kim C, Klopp N, Koivisto K, Larsson M, Lathrop M, Lechner-Scott JS, Leone MA, Leppä V, Liljedahl U, Bomfim IL, Lincoln RR, Link J, Liu J, Lorentzen AR, Lupoli S, Macciardi F, Mack T, Marriott M, Martinelli V, Mason D, McCauley JL, Mentch F, Mero IL, Mihalova T, Montalban X, Mottershead J, Myhr KM, Naldi P, Ollier W, Page A, Palotie A, Pelletier J, Piccio L, Pickersgill T, Piehl F, Pobywajlo S, Quach HL, Ramsay PP, Reunanen M, Reynolds R, Rioux JD, Rodegher M, Roesner S, Rubio JP, Rückert IM, Salvetti M, Salvi E, Santaniello A, Schaefer CA, Schreiber S, Schulze C, Scott RJ, Sellebjerg F, Selmaj KW, Sexton D, Shen L, Simms-Acuna B, Skidmore S, Sleiman PM, Smestad C, Sørensen PS, Søndergaard HB, Stankovich J, Strange RC, Sulonen AM, Sundqvist E, Syvänen AC, Taddeo F, Taylor B, Blackwell JM, Tienari P, Bramon E, Tourbah A, Brown MA, Tronczynska E, Casas JP, Tubridy N, Corvin A, Vickery J, Jankowski J, Villoslada P, Markus HS, Wang K, Mathew CG, Wason J, Palmer CN, Wichmann HE, Plomin R, Willoughby E, Rautanen A, Winkelmann J, Wittig M, Trembath RC, Yaouanq J, Viswanathan AC, Zhang H, Wood NW, Zuvich R, Deloukas P, Langford C, Duncanson A, Oksenberg JR, Pericak-Vance MA, Haines JL, Olsson T, Hillert J, Ivinson AJ, De Jager PL, Peltonen L, Stewart GJ, Hafler DA, Hauser SL, McVean G, Donnelly P, Compston A. Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature. 2011 Aug 10;476(7359):214-9. doi: 10.1038/nature10251. PubMed PMID: 21833088; PubMed Central PMCID: PMC3182531.

Corrado L, Ratti A, Gellera C, Buratti E, Castellotti B, Carlomagno Y, Ticozzi N, Mazzini L, Testa L, Taroni F, Baralle FE, Silani V, **D'Alfonso** S. High frequency of TARDBP gene mutations in Italian patients with amyotrophic lateral sclerosis. Hum Mutat. 2009 Apr;30(4):688-94. doi: 10.1002/humu.20950. PubMed PMID: 19224587.

Kozyrev SV, Abelson AK, Wojcik J, Zaghlool A, Linga Reddy MV, Sanchez E, Gunnarsson I, Svenungsson E, Sturfelt G, Jönsen A, Truedsson L, Pons-Estel BA, Witte T, **D'Alfonso** S, Barizzone N, Danieli MG, Gutierrez C, Suarez A, Junker P, Laustrup H, González-Escribano MF, Martin J, Abderrahim H, Alarcón-Riquelme ME. Functional variants in the B-cell gene BANK1 are associated with systemic lupus erythematosus. Nat Genet. 2008 Feb;40(2):211-6. doi: 10.1038/ng.79. Epub 2008 Jan 20. Erratum in: Nat Genet. 2008 Apr;40(4):484. Barrizzone, Nadia [corrected to Barizzone, Nadia]. PubMed PMID: 18204447.

D'Alfonso S, Barizzone N, Giordano M, Chiocchetti A, Magnani C, Castelli L, Indelicato M, Giacopelli F, Marchini M, Scorza R, Danieli MG, Cappelli M, Migliaresi S, Bigliardo B, Sabbadini MG, Baldissera E, Galeazzi M, Sebastiani GD, Minisola G, Ravazzolo R, Dianzani U, Momigliano-Richiardi P. Two single-nucleotide polymorphisms in the 5' and 3' ends of the osteopontin gene contribute to susceptibility to systemic lupus erythematosus. Arthritis Rheum. 2005 Feb;52(2):539-47. PubMed PMID: 15692970

AWARDS

1. 2014 "Best International Scientific Paper", Università del Piemonte Orientale.