CURRICULUM VITAE

Date: June 2010

Last name: **Otaegui Bichot**

First name: **David**

Date and place of birth: 29 August 1975 San Sebastián, SPAIN

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Nationality: Spanish

- Head of the Multiple sclerosis research Unite at Biodonostia Research Institute.

EDUCATION:

- Bachelor´s Degree in Biochemistry by the Basque Country University (UPV/EHU) in 1999. (1994-1999)
- PhD in Genetics by the Basque Country University (UPV/EHU) in June 2007.

SCIENTIFIC TRAINING :

- 1999: Start working in the Neurogenetic research group in Donostia Hospital with Dr.López de Munain
- Spend one month (2000) in the Dr. J. Montoya´s lab in Zaragoza, learning the molecular tools to work with the mitochondrial DNA.
- Spend 9 months (2001 Sep → 2002 Jun) In the Dr. M. Hirano´s lab in Columbia University, New York city, USA. This lab is focused in molecular research in neuromuscular diseases.
- Spend 3 months (April-June 2005) in the Dr. Oksenberg and Dr. Baranzini´s laboratory in University of California in San Francisco (UCSF), San Francisco, CA, USA. This lab is focused in multiple sclerosis research. During this time I was learning expression-array data analysis.

- Spend 1 month (Aug 2008) in Dr.Sergio Baranzini lab´s (University of California, San Francisco, ***USA***) working in the data analysis of arrays and in the interaction between the data
- Spend 15 days (Nov 2009) in the *Centre for Cellular and Mollecular Biology* located in Hyderabad, ***India*** , with Dr. Satish Kumar.

FELLOWSHIP

- November 1999 → Sep 2001, Research grant by the ILUNDAIN FUNDAZIOA
- Sep 2001 → October 2002: Grant of the Basque government in the project: "Mitochondrial DNA polymorphisms in the multiple sclerosis"
- Oct 2002 → Sep 2006: Pre-Doctoral grant by the Basque Government
- Oct 2006 → Jan 2007: ILUNDAIN FUNDAZIOA. Researcher
- Feb 2007 → Biodonostia Research Institute. Head of the multiple sclerosis Unit.

INVITED REVIEWER OF:

- Multiple Sclerosis Journal	journal
- International journal of Biomedical Sciences.	journal
- Annals of Neurology	journal
- Neuromolecular medicine.	journal
- Diabetes Research and Clinical Practice	journal
- BMC genomics	journal
- Medical Research Council (South Africa).	Funding institution
- Swiss National Science Foundation	Funding institution

PUBLICATIONS

1. Alcina A, Fedetz M, Fernández O, Saiz A, Izquierdo G, Lucas M, Leyva L, García-León JA, Abad-Grau MD, Alloza I, Antigüedad A, Garcia-Barcina MJ, Vandenbroeck K, Varadé J, de la Hera B, Arroyo R,

Comabella M, Montalban X, Petit-Marty N, Navarro A, **Otaegui D**, Olascoaga J, Blanco Y, Urcelay E, Matesanz Identification of a functional variant in the KIF5A-CYP27B1-METTL1-FAM119B locus associated with multiple sclerosis. F. J Med Genet 2012 Nov 17

2. Matías Sáenz-Cuesta, Natalia Martínez-Pomar; Javier de Gracia, Pilar Echaniz, Eduardo Villegas, Alvaro Prada, **David Otaegui**, Nuria Matamoros, Emilio Cuadrado. TACI mutation in Good´s Syndrome: in search of a genetic basis. Clinical immunology.
3. Jezabel Varadé*[1], Manuel Comabella*[2], Miguel A. Ortiz[1], Rafael Arroyo[3], Oscar Fernández[4], M. José Pinto-Medel[5], María Fedetz[6], Guillermo Izquierdo[7], Miguel Lucas[8], Carlos López Gómez[4], Antonio Catalá Rabasa[6], Antonio Alcina[6], Fuencisla Matesanz[6,7], Iraide Alloza[9], Alfredo Antigüedad[10], María García-Barcina[11], **David Otaegui[12]**, Javier Olascoaga[13], Albert Saiz[14], Yolanda Blanco[14], Xavier Montalbán[2], Koen Vandenbroeck*[9, 15], Elena Urcelay. Replication study of ten genes showing evidence for association with multiple sclerosis: validation of *TMEM39A, IL12B* and *CLBL* genes. Acepted in Multiple Sclerosis
4. Cantó E[1], Reverter F[2], Morcillo-Suárez C[3,4], Matesanz F[5], Fernández O[6], Vandenbroeck K[7,8], Rodríguez-Antigüedad A[9], Urcelay E[10], **Otaegui D[11]**, Olascoaga J[11], Saiz A[12], Navarro A[4,13], Sanchez A[14], Domínguez C[15,16], Caminero A[1], Horga A[1], Tintoré M[1], Montalban X[1], Comabella M. Chitinase 3-like 1 plasma levels are increased in patients with progressive forms of multiple sclerosis. Acepted in Multiple Sclerosis.
5. Alloza I, **Otaegui D**, de Lapuente AL, Antigüedad A, Varadé J, Núñez C, Arroyo R, Urcelay E, Fernandez O, Leyva L, Fedetz M, Izquierdo G, Lucas M, Oliver-Martos B, Alcina A, Saiz A, Blanco Y, Comabella M, Montalban X, Olascoaga J, Matesanz F, Vandenbroeck K. ANKRD55 and DHCR7 are novel multiple sclerosis risk loci. Genes Immun. 2011 Dec 1. doi: 10.1038/gene.2011.81. [Epub ahead of print
6. Irizar H, Muñoz-Culla M, Zuriarrain O, Goyenechea E, castillo-Triviño T, prada A, Saenz-Cuesta M, De Juan MD, Lopez de Munain A, Olascoaga J, **Otaegui D**. HLA-DRB1*1501 AND MULTIPLE SCLEROSIS, A FEMALE ASSOCIATION? Multiple Sclerosis. In press.2011

7. M. L. Cavanillas, O. Fernández, M. Comabella, A. Alcina, M. Fedetz, G. Izquierdo, M. Lucas, M. C. Cénit, R. Arroyo, K. Vandenbroeck, I. Alloza, M. García-Barcina, A. Antigüedad, L. Leyva, C. López Gómez, J. Olascoaga, **D. Otaegui**, Y. Blanco, A. Saiz, X. Montalbán, F. Matesanz and E. Urcelay. Replication of top markers of a genome-wide association study in multiple sclerosis in Spain. Genes Immun. 2010 August
8. Alcina A, Vandenbroeck K, **Otaegui D**, Saiz A, Gonzalez JR, Fernandez O, Cavanillas ML, Cénit MC, Arroyo R, Alloza I, García-Barcina M, Antigüedad A, Leyva L, Izquierdo G, Lucas M, Fedetz M, Pinto-Medel MJ, Olascoaga J, Blanco Y, Comabella M, Montalban X, Urcelay E, Matesanz F. The autoimmune disease-associated KIF5A, CD226 and SH2B3 gene variants confer susceptibility for multiple sclerosis. Genes Immun. 2010 May 27
9. Koen Vandenbroeck[1,2], Iraide Alloza[1], Bhairavi Swaminathan[1], Alfredo Antigüedad[3], **David Otaegui[4]**, Javier Olascoaga[4], Maria García Barcina[5], Virginia de las Heras[6], Manuel Bartolomé[6], Miguel Fernández-Arquero[7], Rafael Arroyo[6], Roberto Alvarez-Lafuente[6], M. Carmen Cénit[7] and Elena Urcelay. Validation of IRF5 as multiple sclerosis risk gene: putative role in interferon beta therapy and human herpes virus 6 infection. Genes and immunity 2010
10. Swaminathan B, Matesanz F, Cavanillas ML, Alloza I, **Otaegui D**, Olascoaga J, Cénit MC, Heras VD, Barcina MG, Arroyo R, Alcina A, Fernandez O, Antigüedad A, Urcelay E, Vandenbroeck K. Validation of the CD6 and TNFRSF1A loci as risk factors for multiple sclerosis in Spain, J Neuroimmunol. 2010 Apr 27
11. Ruiz-Martínez J. Gorostidi A. Ibañez B, Alzualde A, **Otaegui D,** Moreno F, López de Munain A, bergareche A, Gömez Esteban JC, Martí-Massó F. Penetrance in Parkinson´s disease related to the LRRK2 R1441G mutation in the Basque Country (Spain)" Movement Disorders 2010
12. Alzualde A, Moreno F, Martinez-Lage P, Ferrer I, Gorostidi A, **Otaegui D**, Blázquez L, Atares B, Cardoso S, Martinez de Pancorbo M, Juste R, Rodriguez-Martínez AB, Indakoetxea B, Lopez de Munain A. Somatic mosaicism in a case of apparently sporadic Creutzfeldt-Jakob

disease carrying a de novo D178N mutation in the PRNP gene. Neuropsychyatric genetics 2010, 2010 Oct 5;153B(7):1283-91

13. **Otaegui D,** Querejeta R, Arrieta A, Lazkano A, Bidaurrazaga A, Arriandiaga JR, Aldazabal P,Garro MA. Phospholipase Cß4 isozyme is expressed in human, rat, and murine heart left ventricles and in HL-1 cardiomyocytes. Mol Cell Biochem. 2009 Oct 24
14. Sistiaga A, Urreta I, Jodar M, Cobo AM, Emparanza J, **Otaegui D**, Poza JJ, Merino JJ, Imaz H, Martí-Massó JF, López de Munain A. Cognitive/personality pattern and triplet expansion size in adult myotonic dystrophy type 1 (DM1): CTG repeats, cognition and personality in DM1.Psychol Med. 2009 Jul 23:1-9.
15. **Otaegui D**, Baranzini SE, Armañanzas R, Calvo B, Muñoz-Culla M, Khankhanian P, Inza I, Lozano JA, Castillo-Triviño T, Asensio A, Olaskoaga J , López de Munain A. Differential micro RNA expression in PBMC from Multiple Sclerosis. PLoS One. 2009 Jul 20;4(7):e6309.
16. A. Sistiaga, P. Camaño, **D. Otaegui**, B. Ibáñez, J. Ruiz-Martinez, J.F. Martí-Massó, and A. López de Munain. Cognitive function in facioscapulohumeral dystrophy correlatos with the molecular defect. Genotype-cognitive phenotype correlation in FSHD. Genes, Brain and behaviour. Genes Brain Behav. 2009 Feb;8(1):53-9. Epub 2008 Sep 22..
17. **Otaegui D**, Zuriarrain O, Castillo-Triviño T, Aransay AM, Ruíz-Martínez J, Olaskoaga J, Martí-Massó JF, López de Munain A. Association between Synapsin III gene promoter SNPs and multiple sclerosis in Basque patients. Multiple sclerosis, 2009;15:124-128
18. Blázquez L, Azpitarte M, Saenz A, Goicoechea M, **Otaegui D**, Ferrer X, Illa I, Gutierrez-Rivas E, Vilchez JJ, López de Munain A. Characterization of novel capn3 isoforms in white blood cells: an alternative approach for limb-girdle muscular dystrophy 2A diagnosis. Neurogenetics 2008 Jul;9(3):173-82, 2008
19. Carcamo-Orive I, Tejados N, Delgado J, Gaztelumendi A, **Otaegui D**, lang V, Trigueros C. ERK2 protein regulates the proliferation of human mesenchymal ítem cells without affecting their mobilization and differentiation potencial. Experimental Cell Research, 2008

20. **Otaegui D**, Irizar H, Goicoechea M, Pérez-Tur J, Belar M and López de Munain A. Molecular characterization of putative modulatory factors in two Spanish families with A1555G deafness. 2008 Audiology and Neurootology. Apr 7;13(5):320-327
21. Quinzi CM, Vu T, Min KC, Tanji K, Barral S, Grewal RP, Kattah A, Camano P, **Otaegui D,** Kunimatsu K, Blake Dm, Wilhelmsen KC, Rowland LP, Hays AP, Bonilla E, Hirano M. X-Linked dominant scapuloperoneal myopathy is due to a mutation in the gene encoding four-and-a-half-LIM protein 1 (FLH1). American Journal of Human Genetics. 2008 Jan;82(1):208-13
22. Adolfo López de Munain, Ainhoa Alzualde (*), Ana Gorostidi, **David Otaegui**, Javier Ruiz-Martínez, Begoña Indakoetxea, Isidro Ferrer, Jordi Pérez-Tur ,Amets Sáenz, Alberto Bergareche, Miriam Barandiarán, Juan José Poza, Ramón Zabalza , Irune Ruiz, Miguel Urtasun, Iñaki Fernández-Manchola, Bixen Olasagasti, Juan Bautista Espinal, Javier Olaskoaga, Marta Ruibal, Fermin Moreno, Nieves Carrera, José Félix Martí Masso. Mutations in progranulin gene:Clinical, Pathological and RNA expresión findings. Biological psychiatry. May 15;63(10):946-52.
23. **Otaegui D**, Mostafavi S, Bernard CCA, Lopez de Munain A, Mousavi P, Oksenberg JR, Baranzini SE. Increased transcriptional activity of milk related genes following the active phase of EAE and MS. Journal of Immunology. 2007. 179(6) : 4047-82.
24. M Goicoechea, F Cía, C San José, A Asensio, JI Emparanza, AG Gil, A López de Cerain, P Aldazabal, M Azpitarte, **D Otaegui**, A López de Munain Minimising Creatine Kinase variability in rats for neuromuscular research purposes. Laboratory Animals 2008. Vol 42(1): 19-25.
25. **Otaegui D**, Ruíz-Martínez J, Olaskoaga J, Emparanza JI, López de Munain A. Influence of CCR5-Δ32 genotype in Spanish population with multiple sclerosis. Neurogenetics. 2007 Aug;8(3):201-5.
26. Blázquez L, De Juan D, Ruíz-Martínez J, Emparanza JI, Sáenz A, **Otaegui D**, Sistiaga A, Martínez-Lage P, Lamet I, Samaranch L, Buiza C, Echeberria I, Arriola E, Cuadrado E, Urdaneta E, Yanguas J, López de Munain A. Genes related to iron metabolism and susceptibility to

Alzheimer´s disease in Basque population. Neurobiology of aging. 2007 Dec;28(12):1941-3

27. **Otaegui D**, Sáenz A, Ruiz-Martinez J, Olaskoaga J, Lopez de Munain A. UCP2 and mitochondrial haplogroups as an MS risk factor. Multiple Sclerosis. 2007; 13 ;454-458
28. Blázquez L, **Otaegui D**, Sáenz A, Paisán-Ruiz C, Emparanza JI, Ruiz-Martinez J, Moreno F, Martí-Masso JF, López de Munain A. Apolipoprotein E in e4 allele in familial and sporadic Parkinson´s disease. Neuroscience letters 2006, 9; 406(3):235-239
29. **Otaegui D**, Sáenz A, Camaño P, Blázquez L, Goicoechea M, Ruíz.Martínez J, Olaskoaga J, Emparanza JA, López de Munain A. CD24 V/V is an allele associated with the risk of developing multiple sclerosis in the Spanish population. Multiple Sclerosis. Multiple Sclerosis. 2006;12:511-514.
30. Sáenz A, Leturcq F, Cobo AM, Poza JJ, Ferrer X, **Otaegui D**, Camaño P, Urtasun M, Vílchez J, Gutierrez-Rivas E, Emparanza J, Merlini L, Paisán C, Goicoechea M, Blázquez L, Eymard B, Lochmuller H, Walter M, Bonnemann C, Figarella-Branger D, Kaplan JC, Urtizberea JA, Martí-Masso JF, López de Munain A. LGMD2A: Genotype-Phenotype correlations based on a large mutational survey on Calpain 3 gene. Brain (2005) 128, 732-742
31. **Otaegui D**, Paisán C, Sáenz A, Martí I, Ribate M, Martí-Massó JF, Pérez-Tur J, López de Munain A. Mitochondrial polymorphisms in parkinson´s disease. Neuroscience letters 2004, 370 pp171-174.
32. **Otaegui D**, Sáenz A, Martínez-Zabaleta M, Villoslada P, Fernández-Manchola, Álvarez de Arcaya A, Emparanza JI, López de Munain A. Mitochondrial Haplogroups in Basque multiple sclerosis patients. multiple sclerosis 2004; 10:532-535
33. Rozen TD, Shanske S, **Otaegui D**, Lu J, Young WB, Bradley K, DiMauro S, Silberstein SD. Study of mitochondrial DNA mutations in patients with migraine with prolonged aura. Headache. 2004 Jul;44(7):674-7.
34. Goertsches R, Villoslada P, Comabella M, Montalban X, Navarro A, G de la Concha E, Arroyo R, López de Munain A, **Otaegui D**, Palacios R,

Perez-Tur J, Jonasdottir A, Benediktsson K, Fossdal R, Sawcer S, Setakis E, Clayton D, Compston A and the Spanish MS Genetic Group. A genomic Screen of Spanish Multiple Sclerosis Patients Reveals Multiple Loci Associated with the disease. J. Neuroimmunol. 2003 Oct;143(1-2):124-8

35. Salviati L, Sacconi S, Mancuso M, **Otaegui D**, Camano P, Marina A, Rabinowitz S, Shiffman R, Thompson K, Wilson CM, Feigenbaum A, Naini AB, Hirano M, Bonilla E, DiMauro S, Vu TH. Mitochondrial DNA depletion and dGK gene mutations. Ann Neurol. 2002 Sep;52(3):311-7.
36. Mancuso M, Salviati L, Sacconi S, **Otaegui D**, Camaño P, Marina A, Bacman S, Moraes C, Carlo J, García M, García-Alvarez M, Monzon L, Naini A, Hirano M, Bonilla E, DiMauro S, Vu T. "Novel mutations in the thymidine kinase 2 (TK2) gene in mitochondrial DNA depletion syndrome" Neurology 2002 Oct 22;59(8):1197-1202
37. A. López de Munain, F. García Bragado, A. Sáenz, **D. Otaegui**, P. Camaño. manifestaciones clínicas y diagnóstico de la distrofia muscular. Jano 12-18 Abril 2002, Vol. LXII.Nº 1.427l

AWARDS:

- Young scientific travel grant in ECTRIMS 2007
- Young scientific travel grant in ECTRIMS 2007
- Award poster in the SEN 2002
- Special Thesis prize by the University of the Basque Country (2009): (extraordinary Thesis prize)

PATENTS:

- **European patent nº EP09382108.0**

Title: METHODS FOR THE DIAGNOSIS OF MULTIPLE SCLEROSIS BASED ON ITS MICRORNA EXPRESSION PROFILING Solicitant: ADMINISTRACIÓN GENERAL DE LA COMUNIDAD AUTÓNOMA DE EUSKADI Inventors: David Otaegui, Javier Olascoaga, Adolfo López de Munain.

COMPETITIVE RESEARCH GRANTS

As Principal Investigator

- Red Española de esclerosis Múltiple. 2012. 22.000€
- SAIO 12. LMP en la esclerosis múltiple. Saiotek. 17500 €. 2012
- ***GV-2011111028***. Rol de las micropartículas como biomarcador en EM. Gobierno Vasco. 35.000 €. 2012-2014
- ***SAIO11-PC11BN003*** Biomarcadores en la Esclerosis Múltiple. SAIOTEK. 45.384 €. 2011-2012
- ***DFG11/005***. Análisis genético y clínico en familias vascas con esclerosis múltiple. DFG11/005. 61.125€ 2011-2012
- ***BIOD11/017***. Mirna identification and quantification in MS patients treat. Financiado por fundación Biogen. 11.500 €. 2010-2012
- ***PS09/02105***. Patrones de expresión génica en el momento del brote. Financiado por el Fondo de investigación Sanitaria (FIS) Cuantía : 102000€. Año 2010-2013
- ***GV-2008111079***. Estudio de los genes implicados en la recuperación del brote en el modelo animal (EAE) de la esclerosis múltiple y en pacientes. Financiado por el Departamento de Sanidad del Gobierno Vasco Cuantía 16300 €. Año 2008-2011.
- ***GV-2007111054***. Estudio en pacientes con Esclerosis Múltiple de los patrones de Expresión génica en el momento del brote. Financiado por el Departamento de Sanidad del Gobierno vasco. Cuantía 20000€, Año 2007-2008 (1 año)
- ***DFG07/006***. Estudio de los patrones de Expresión génica en pacientes con Esclerosis Múltiple durante el momento del brote. Financiado por la Diputación Foral de Gipuzkoa. Cuantía 11500€, Año 2007-2008 (1 año)
- ***DFG08/007.*** Estudio de los patrones de expresión de los microRNA en pacientes con EM. Financiado por la Diputación Foral de Gipuzkoa Cuantía 13.623 €, Año 2008-2009 (1 año)

As Colaborator

- Fundación de Investigación Médica Mutua Madrileña (FMM). 2007-2010
- Fundación de Investigación Médica Mutua Madrileña (FMM). 2007-2008
- Fondo de Investigación Sanitaria (FIS)01/0108-02) : 2001-2003 29000€
- Fundación 2000. 2005-2007. 20000€