Publicaciones de usuarios con mención al biobanco (2014-2017)

Autores	Título	Año	Revista
M. Corton, A. Avila-Fernández, L. Campello, M. Sánchez , B. Benavides, M. I. López-Molina, L. Fernández-Sánchez, R. Sánchez-Alcudia, L. R. J. da Silva, N. Reyes, E. Martín-Garrido, O. Zurita, P. Fernández-San José, R. Pérez-Carro, F. García-García, J. Dopazo, B. García-Sandoval, N. Cuenca & C. Ayuso	Identification of the Photoreceptor Transcriptional Co-Repressor *SAMD11* as Novel Cause of Autosomal Recessive Retinitis Pigmentosa.	2016	Scientific Reports - Nature
Radulfus WN Slijkerman, Christel Vaché, Margo Dona, Gema García-García , Mireille Claustres, Lisette Hetterschijt, Theo A Peters, Bas P Hartel, Ronald JE Pennings, José M Millan , Elena Aller , Alejandro Garanto, Rob WJ Collin, Hannie Kremer, Anne-Françoise Roux and Erwin Van Wijk.	Antisense Oligonucleotide-based Splice Correction for USH2A-associated Retinal Degeneration Caused by a Frequent Deep-intronic Mutation.	2016	Molecular Therapy - Nucleic Acids
Consuegra Irene, Rodríguez-Aierbe Clara, Santiuste Inés, Bosch Anna, Martínez-Marín Rosario, Fortuto M Antonia, Díaz Tatiana, Martí Salvador, and Muñoz-Fernández M Ángeles.	Isolation methods of peripheral blood mononuclear cells in Spanish biobanks: an overview.	Aceptado en 2016	Biopreservation and Biobanking
Salvador Martí, Marian León, Carmen Orellana, Javier Prieto, Xavier Ponsoda, Carlos López-García, Juan Jesús Vílchez, Teresa Sevilla, Josema Torres.	Generation of a disease-specific iPS cell line derived from a patient with Charcot-Marie-Tooth type 2K lacking functional *GDAP1* gene.	2016	Lab Resource - Stem Cell Research

Publicaciones de usuarios con muestras del biobanco (2014-2017)

Autores	Título	Año	Revista
Nishiguchi KM, Avila-Fernandez A, van Huet RA, Corton M, Pérez-Carro R, Martín-Garrido E, López-Molina MI, Blanco-Kelly F, Hoefsloot LH, van Zelst-Stams WA, García-Ruiz PJ, Del Val J, Di Gioia SA, Klevering BJ, van de Warrenburg BP, Vazquez C, Cremers FP, García-Sandoval B, Hoyng CB, Collin RW, Rivolta C, Ayuso C.	Exome sequencing extends the phenotypic spectrum for ABHD12 mutations: from syndromic to nonsyndromic retinal degeneration.	2014	Ophthalmology
Koji M. Nishiguchi, Almudena Avila-Fernandez, Ramon A. C. van Huet, MD, Marta Corton, Raquel Pérez-Carro, Esther Martín-Garrido, María Isabel López-Molina, Fiona Blanco-Kelly, Lies H. Hoefsloot, Wendy A. van Zelst-Stams, Pedro J. García-Ruiz Javier del Val, Silvio Alessandro Di Gioia, B. Jeroen Klevering, Bart P. C. van de Warrenburg, Carlos Vazquez, Frans P. M. Cremers, Blanca García-Sandoval, Carel B. Hoyng, Rob W. J. Collin, Carlo Rivolta, Carmen Ayuso.	Exome Sequencing Extends the Phenotypic Spectrum for ABHD12 Mutations.	2014	Ophthalmology
Eduardo Calpena, Dolores Martínez-Rubio, Javier Arpa, Juan J García-Peñas, David Montaner D, Joaquín Dopazo, Francesc Palau, Carmen Espinós.	A novel locus for a hereditary recurrent neuropathy on chromosome 21q21.	2014	Neuromuscular Disorders

Zernant J, Xie YA, Ayuso C, Riveiro-Alvarez R, Lopez-Martinez MA, Simonelli F, Testa F, Gorin MB, Strom SP, Bertelsen M, Rosenberg T, Boone PM, Yuan B, Ayyagari R, Nagy PL, Tsang SH, Gouras P, Collison FT, Lupski JR, Fishman GA, Allikmets R.	Analysis of the ABCA4 genomic locus in Stargardt disease.	2014	Hum Mol Genet.
Benaglio P, San Jose PF, Avila-Fernandez A, Ascari G, Harper S, Manes G, Ayuso C, Hamel C, Berson EL, Rivolta C.	Mutational screening of splicing factor genes in cases with autosomal dominant retinitis pigmentosa.	2014	Molecular Vision
Berta Almoguera, Sijie He, Marta Corton, Patricia Fernandez-San Jose, Fiona Blanco-Kelly, Maria Isabel López-Molina, Blanca García-Sandoval, Javier del Val, Yiran Guo, Lifeng Tian, Xuanzhu Liu, Liping Guan, Rosa J Torres, Juan G Puig, Hakon Hakonarson, Xun Xu, Brendan Keating[†] and Carmen Ayuso.	Expanding the phenotype of PRPS1 syndromes in females: neuropathy, hearing loss and retinopathy.	2014	Orphanet Journal of Rare Diseases
Vincenzo Lupo, Samuel I Pascual-Pascual, Manuel Gutiérrez-Molina, Gonzalo Mateo-Martínez, Carmen Espinós, Gema Arriola-Pereda.	Complexity of the phenotypes due to mutations in the *MPZ* gene: Clinical and cellular characterization of the *MPZ* p.D90E mutation.	2014	Pediatric Research

Lukovic D, Artero Castro A, Delgado AB, Bernal Mde L, Luna Pelaez N, Díez Lloret A, Perez Espejo R, Kamenarova K, Fernández Sánchez L, Cuenca N, Cortón M, Avila Fernandez A, Sorkio A, Skottman H, Ayuso C, Erceg S, Bhattacharya SS.	Human iPSC derived disease model of MERTK-associated retinitis pigmentosa.	2015	Scientific Reports - Nature
Almoguera B, Li J, Fernandez-San Jose P, Liu Y, March M, Pellegrino R, Golhar R, Corton M, Blanco-Kelly F, López-Molina MI, García-Sandoval B, Guo Y, Tian L, Liu X, Guan L, Zhang J, Keating B, Xu X, Hakonarson H, Ayuso C.	Application of Whole Exome Sequencing in Six Families with an Initial Diagnosis of Autosomal Dominant Retinitis Pigmentosa: Lessons Learned.	2015	PLoS One
Pla-Martín D, Calpena E, Lupo V, Márquez C, Rivas E, Sivera R, Sevilla T, Palau F, Espinós C.	Junctophilin-1 is a modifier gene of GDAP1-related Charcot-Marie-Tooth disease.	2015	Hum Mol Genet
Sevilla T, Sivera R, Martínez-Rubio D, Lupo V, Chumillas MJ, Calpena E, Dopazo J, Vílchez JJ, Palau F, Espinós C.	The EGR2 gene is involved in axonal Charcot-Marie-Tooth disease.	2015	Eur J Neurol
Avila-Fernandez A, Perez-Carro R, Corton M, Lopez-Molina MI, Campello L, Garanto A, Fernandez-Sanchez L, Duijkers L, Lopez-Martinez MA, Riveiro-Alvarez R, Da Silva LR, Sanchez-Alcudia R, Martin-Garrido E, Reyes N, Garcia-Garcia F, Dopazo J, Garcia-Sandoval B, Collin RW, Cuenca N, Ayuso C.	Whole-exome sequencing reveals ZNF408 as a new gene associated with autosomal recessive retinitis pigmentosa with vitreal alterations.	2015	Hum Mol Genet

Fernandez-San Jose P, Corton M, Blanco-Kelly F, Avila-Fernandez A, Lopez-Martinez MA, Sanchez-Navarro I, Sanchez-Alcudia R, Perez-Carro R, Zurita O, Sanchez-Bolivar N, Lopez-Molina MI, Garcia-Sandoval B, Riveiro-Alvarez R, Ayuso C.	Targeted Next-Generation Sequencing Improves the Diagnosis of Autosomal Dominant Retinitis Pigmentosa in Spanish Patients.	2015	Investigative Ophthalmology and Visual Science
Riveiro-Álvarez R, Xie YA, López-Martínez MÁ, Gambin T, Pérez-Carro R, Ávila-Fernández A, López-Molina MI, Zernant J, Jhangiani S, Muzny D, Yuan B, Boerwinkle E, Gibbs R, Lupski JR, Ayuso C, Allikmets R.	New mutations in the RAB28 gene in 2 Spanish families with cone-rod dystrophy.	2015	JAMA Ophthalmology
Sevilla T, Lupo V, Martínez-Rubio D, Sancho P, Sivera R, Chumillas MJ, García-Romero M, Pascual-Pascual SI, Muelas N, Dopazo J, Vílchez JJ, Palau F, Espinós C.	Mutations in the MORC2 gene cause axonal Charcot-Marie-Tooth disease.	2016	Brain
Lupo, V ; Garcia-Garcia, F; Sancho, P ; Tello, C; Garcia-Romero, M; Villarreal, L; Alberti, A; Sivera, R; Dopazo, J ; Pascual-Pascual, SI; Marquez-Infante, C; Casasnovas, C; Sevilla, T; Espinos, C.	Assessment of Targeted Next-Generation Sequencing as a Tool for the Diagnosis of Charcot-Marie-Tooth Disease and Hereditary Motor Neuropathy	2016	J Mol Diagn
Perez-Carro R, Corton M, Sánchez-Navarro I, Zurita O, Sanchez-Bolivar N, Sánchez-Alcudia R, Lelieveld SH, Aller E, Lopez-Martinez MA, López-Molina MI, Fernandez-San Jose P, Blanco-Kelly F, Riveiro-Alvarez R, Gilissen C, Millan JM, Avila-Fernandez A, Ayuso C.	Panel-based NGS Reveals Novel Pathogenic Mutations in Autosomal Recessive Retinitis Pigmentosa.	2016	Scientific Reports - Nature

Rocio Sanchez-Alcudia, Maria Garcia-Hoyos, Miguel Angel Lopez-Martinez, Noelia Sanchez-Bolivar, Olga Zurita, Ascension Gimenez, Cristina Villaverde, Luciana Rodrigues-Jacy da Silva, Marta Corton, Raquel Perez-Carro, Simona Torriano, Vasiliki Kalatzis, Carlo Rivolta, Almudena Avila-Fernandez, Isabel Lorda, Maria J. Trujillo-Tiebas, Blanca Garcia-Sandoval, Maria Isabel Lopez-Molina, Fiona Blanco-Kelly, Rosa Riveiro-Alvarez, Carmen Ayuso.	A Comprehensive Analysis of Choroideremia: From Genetic Characterization to Clinical Practice.	2016	PLoS One
Corton M, Nishiguchi KM, Avila-Fernández A, Nikopoulos K, Riveiro-Alvarez R, Tatu SD, Ayuso C, Rivolta C.	Exome sequencing of index patients with retinal dystrophies as a tool for molecular diagnosis.	2016	PLoS One
Martí S, León M, Orellana J, Prieto J, Ponsoda X, López-García C, Vílchez JJ, Sevilla T, Torres J.	Generation of a disease-specific iPS cell line derived from a patient with Charcot-Marie-Tooth type 2K lacking functional GDAP1 gene.	2017	Stem Cell Research