CURRICULUM VITAE

Name **MARTA CORTON**

Contact details Department of Genetics & Genomics
Institute of Health Research-Fundación Jiménez Díaz Hospital (IIS-FJD)
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CURRENT POSITION

2013 to present **Principal Investigator, Group of Congenital Eye Disorders**
Researcher Miguel Servet, Spanish National Health Institute (from 2013)

Molecular study of several congenital ocular malformations through high-throughput genomic strategies, such as aCGH and Next-generation sequencing (NGS).

EDUCATIONAL BACKGROUND

December 2006 **PhD in Biomedicine** (Cum laude Distinction**)**
Genomic and Proteomic approximation to Polycystic Ovary Syndrome: Search of alterations in adipose omental tissue.
Supervisor: Dr. Belén Peral
Faculty of Medicine, University Autonoma of Madrid

Sept 2003 **Master in Applied Statistical Methods**
UNED University, Spain (Open University)

July 2000 **MSc in Biotechnology**. University Complutense of Madrid
Second year spent at University Paul Sabatier, Toulouse (France), supported by an Erasmus European grant.

July 1997 **BSc in Biology.** University Complutense of Madrid.

PREVIOUS POSITIONS

Feb 2010- Feb 2013 **Senior Postdoctoral Researcher**
Centre for Biomedical Research on Rare Diseases (CIBERER).
Genetics and Genomics Department, IIS-FJD, Madrid, Spain

Molecular study of retinal dystrophies (RD). Searching of novel mutations and loci/genes involved in RD through high-throughput genomic strategies, such as whole genome SNP arrays, aCGH and Next-generation sequencing (NGS).

2007- Jan 2010	**Junior Postdoctoral Researcher** Spanish Centre for Biomedical Research on Rare Diseases (CIBERER). Genomics Medicine Group University of Santiago de Compostela-Galician Foundation of Genomics Centro Hospitalario Universitario de Santiago.

Molecular analysis of Mendelian and complex ophthalmic diseases, including Retinitis pigmentosa (RP), age-related macular degeneration (ADM) and open-angle glaucoma. Searching of novel mutations and loci/genes involved in RP using high-throughput genotyping strategies, such as whole-genome SNP arrays, and Next-generation sequencing (NGS). Identification of novel risk alleles to AMD using Genome-wide association studies (GWAS).

2006	Research Assistant at Tumor Markers Group. Spanish National Cancer Research Centre (CNIO), Madrid, Spain
Sept 2001 - Dec 2005	**PhD student**. "Alberto Sols" - Biomedical Research Institute (IIB) Spanish National Research Council, CSIC), Madrid
Sept 1999 - Sept 2000	**Pre-graduate student** Departament of Genetics, Faculty of Biology University Complutense of Madrid.
1999	**Pre-graduate student** Plant Biotechnology Institute French National Research Council, CNRS. Toulouse, France.

GRANT FUNDING AS PRINCIPAL INVESTIGATOR

"Genetic characterization of aniridia in Spain through genomic and experimental studies".
Grant funded by Spanish Federation of Rare Diseases. 2018 (1 year).

"Genetic diagnosis and possible treatment of albinism".
Intramural Grant from the Spanish Centre for Biomedical Research on Rare Diseases (CIBERER). 2018-2019.

"Clinical, genomic and experimental studies for the characterization of the molecular bases of aniridia and microphthalmia".
Grant from the Spanish National Institute of Health (ISCIII) (PI17/01164)
2018- 2020

"Genetic diagnosis and possible treatment of albinism".
Intramural Grant from the Spanish Centre for Biomedical Research on Rare Diseases (CIBERER-ISCIII). Nov. 2015-Nov. 2016.

"Molecular study of Aniridia and other congenital eye malformations: Searching of new genetic mechanisms by massive sequencing".
Grant from the Spanish Ministry of Economy and Competitiveness (MINECO, SAF2013-46943-R).
2015-2017.

"Application of new techniques of massive sequencing to the genetic study of Aniridia and related anterior segment dysgenesis."
Grant from Mutua Madrileña Foundation.
July 2014 - July 2017.

"Towards a better understanding of the genetic basis of congenital ocular malformations using high-throughput genomic technologies".
Miguel Servet program to incorporate researchers into the Spanish National Health System (ISCIII - CP12/03256)

Participation as associated Investigator in additional 26 funded projects (Spanish and European grants) from 2002 to current.

SUPERVISION OF DOCTORAL / GRADUATE THESIS

- Maria Tarilonte, PhD candidate. Granted by Fundación Conchita Rábago. From 2016-
- Mª Inmaculada Martin, PhD. Universidad Autónoma de Madrid. 2018. Cum laude distinction.
- Patricia Fernandez San Jose, PhD. Universidad Autónoma de Madrid. 2017. Cum laude distinction.
- Sorina Daniela Tatu, PhD. Universidad Autónoma de Madrid. 2016. Cum laude distinction
- Marcos Elizalde. Faculty of Biology. Universidad Autónoma de Madrid. 2015

PUBLICATION LIST

1. N Ragge, B Isidor, P Bitoun, S Odent, I Giurgea, B Cogné, W Deb, M Vincent, J Le Gall, J Morton, D Lim, DDD study, G Le Meur, C Zazo Seco, D Zafeiropoulou, P Zwijnenburg, A Arteche, ST Swafiri, W Newman, C Ayuso, **M Corton**, P Calvas, N Chassaing. Expanding the Phenotype of the X-linked BCOR Microphthalmia syndromes. Hum Genet. 2018 Jul 4.
2. R Pérez-Carro, F Blanco-Kelly, L Galbis-Martínez, G García-García, E Aller, B García-Sandoval, P Mínguez, **M Corton**, I Mahillo, I Martín-Mérida, A Avila-Fernández, JM. Millán, C Ayuso. Unravelling the pathogenic role and genotype-phenotype correlation of the USH2A p.(Cys759Phe) variant among Spanish families. PLoS One. 2018 Jun 18;13(6)
3. I Martin-Merida, D Aguilera-García, P Fernandez-San Jose, F Blanco-Kelly, O Zurita, B Almoguera, B Garcia-Sandoval, A Avila-Fernandez, A Arteche, M Carballo, **M Corton***, C Ayuso*. Mutational spectrum in a Spanish cohort of 253 adRP families. * Equal contribution. Toward the Mutational Landscape of Autosomal Dominant Retinitis Pigmentosa: A Comprehensive Analysis of 258 Spanish Families**.** Invest Ophthalmol Vis Sci. 2018 May 1;59(6):2345-2354.
4. I Sanchez-Navarro*, LR Jacy da Silva*, O Zurita, N Sanchez-Bolivar, F Blanco-Kelly, MI Lopez-Molina, B Garcia-Sandoval, ST Swafiri, P Mínguez, R Riveiro-Alvarez, I Lorda, R Sanchez-Alcudia, R Perez-Carro, D Valverde, A Avila-Fernandez, **M Corton***, C Ayuso*. * Equal contribution. Molecular diagnosis of retinal syndromes ans associated ciliopathies: A targeted bases resequencing combined by copy-number variation analysis. Sci Rep. 2018 Mar 27;8(1):5285
5. F. Ceroni, DA. Bax, D. Aguilera-Garcia, R. Holt, J. Bruty, MJ. Ballesta, MJ. Sanchez-Soler, Y. Wallis, D.

McMullan, M. Griffiths, F. Blanco-Kelly, P. Ramos, A, Stewart, J. Hoffman, D. Bunyan, K. Lachlan, A. Fryer, V. McKay, A. Saggar, P. Calvas, N. Chassaing, DDD Study, C. Ayuso, **M. Corton***, N. Ragge*. Novel variants in GJA8: expanding the phenotypic spectrum to include microphthalmia and coloboma. *Equal contribution. Hum Genet. 2018 Feb 20.

6. C Zazo Seco, J Plaisancié, T Lupasco, C Michot, J Pechmeja, J Delanne, E Cottereau, C Ayuso, **M Corton**, P Calvas, N Ragge, N Chassaing. Expanding the phenotype associated with novel and known mutations in PITX3. Ophthalmic Genet. 2018 Feb 6:1-7.

7. - A Bolinches-Amorós, D Lukovic, A Artero Castro, M León Rodriguez, K Kamenarova, R Kaneva, P Jendelova, F Blanco-Kelly, C Ayuso, **M Cortón**, S Erceg. Generation of a human iPSC line from a patient with congenital glaucoma caused by mutation in CYP1B1 gene. Stem Cell Research. https://doi.org/10.1016/j.scr.2018.01.004.

8. - F Zurita-Díaz, T Galera-Monge, A Moreno-Izquierdo, **M Cortón**, C Ayuso, R Garesse, ME Gallardo. Establishment of a human DOA 'plus' iPSC line, IISHDOi003-A, with the mutation in the OPA1 gene: c.1635C>A; p.Ser545Arg. Stem Cell Research. doi: 10.1016/j.scr.2017.08.017

9. - JJ Ferre-Fernández, JD Aroca-Aguilar, C Medina-Trillo, JM Bonet-Fernández, CD Méndez-Hernández, L Morales-Fernández, **M Corton**, MJ Cabañero-Valera, M Gut, R Tonda, C Ayuso, M Coca-Prados, J García-Feijoo, J Escribano. Whole-exome sequencing of congenital glaucoma patients reveals hypermorphic variants in GPATCH3, a new gene involved in ocular and craniofacial development. Sci Rep. 2017 Apr 11;7:46175.

10. - I Prieto*, L del Puerto-Nevado*, N González, S Portal-Núñez, S Zazo, **M Cortón**, P Mínguez, C Gómez-Guerrero, JM Arce, AB Sanz, S Mas, O Aguilera, G Álvarez-Llamas, P Esbrit, A Ortiz, C Ayuso, J Egido, F Rojo, J García-Foncillas. Colon cancer modulation by a diabetic environment: a single institutional experience. PLoS ONE 12(3).

11. - Fiona Blanco-Kelly, María Palomares, Elena Vallespin, Cristina Villaverde, Rubén Martín-Arenas, Camilo Velez, Isabel Lorda-Sánchez, Julián Nevado, María José Trujillo-Tiebas, Pablo Lapunzina, Carmen Ayuso, **Marta Corton**. Improving Molecular Diagnosis of Aniridia and WAGR Syndrome Using Customized Targeted Array-Based CGH. PLoS One. 2017 Feb 23;12(2):

12. - Inmaculada Martin-Merida, Rocio Sanchez-Alcudia, Patricia Fernandez-San Jose, Fiona Blanco-Kelly, Raquel Perez-Carro, Luciana Rodriguez J. da Silva, Berta Almoguera, Blanca Garcia-Sandoval, Maria Isabel Lopez-Molina, Almudena Avila-Fernandez, Miguel Carballo, **Marta Corton***, Carmen Ayuso*. Analysis of the PRPF31 gene in Spanish autosomal dominant Retinitis Pigmentosa patients: a novel genomic rearrangement**.** Invest Ophthalmol Vis Sci. 2017 Feb 1;58(2):1045-1053. * Equally contributors.

13. - Blanco-Kelly F, Rodrigues-Jacy da Silva L, Sanchez-Navarro I, Riveiro-Alvarez R, Lopez-Martinez MA, **Corton M**, Ayuso C. New CDH3 mutation in the first Spanish case of hypotrichosis with juvenile macular dystrophy, a case report. BMC Med Genet. 2017 Jan 7;18(1):1.

14. - González N*, Prieto I*, Del Puerto-Nevado L*, Portal-Nuñez S*, Ardura JA, **Corton M**, Fernández-Fernández B, Aguilera O, Gomez-Guerrero C, Mas S, Moreno JA, Ruiz-Ortega M, Sanz AB, Sanchez-Niño MD, Rojo F, Vivanco F, Esbrit P, Ayuso C, Alvarez-Llamas G, Egido J, García-Foncillas J, Ortiz A, Consortium DC. 2017 update on the relationship between diabetes and colorectal cancer: epidemiology, potential molecular mechanisms and therapeutic implications * Contributed equally. Oncotarget. 2017 Jan 3.

15. - **Corton M**, Avila-Fernandez A, Campello L, Sanchez M, Benavides B, Lopez-Molina MI, Fernandez-Sanchez L, Sanchez-Alcudia R, da Silva LRJ, European Retinal Disease Consortium, Reyes N, Martin-Garrido E, Zurita O, Fernandez-San Jose P, Perez-Carro R, Garcia-Garcia F, Dopazo J, Garcia-Sandoval B, Cuenca N, Ayuso C. Identification of the Photoreceptor Transcriptional Co-Repressor SAMD11 as Novel Cause of Autosomal Recessive Retinitis Pigmentosa. Sci Rep. 2016 Oct 13;6:35370.

16. R Sanchez-Alcudia, M Garcia-Hoyos, MA Lopez-Martinez, N Sanchez-Bolivar, O Zurita, L Rodrigues-Jacy,

M Corton, R Perez-Carro, A Avila-Fernandez, C Rivolta, I Lorda, MJ Trujillo-Tiebas, B Garcia-Sandoval, MI Lopez-Molina, F Blanco-Kelly, R Riveiro-Alvarez, C Ayuso. A comprehensive analysis of choroideremia: from genetic characterization to clinical practice. PLoS One. 2016 Apr 12;11(4).

17. - F Blanco-Kelly; M García-Hoyos; MA Lopez Martínez; MI Lopez-Molina; R Riveiro-Alvarez; P Fernandez-San Jose; A Avila-Fernandez; **M Corton**; B Garcia-Sandoval; C Ayuso. Dominant Retinitis Pigmentosa, p.Gly56Arg mutation in NR2E3: Phenotype in a large cohort of 24 cases. PLoS One. 2016 Feb 24;11(2):e0149473.
18. R Perez-Carro, **M Corton**, I Sánchez-Navarro, O Zurita, N Sanchez-Bolivar, R Sánchez-Alcudia, SH. Lelieveld, E Aller, MA Lopez-Martinez, MI López-Molina, P Fernandez-San Jose, F Blanco-Kelly, R Riveiro-Alvarez, C Gilissen, JM Millan, A Avila-Fernandez, C Ayuso. Panel-based NGS Reveals Novel Pathogenic Mutations in Autosomal Recessive Retinitis Pigmentosa. Sci. Reports.
19. D Lukovic, A Artero Castro, AB Garcia Delgado, MA Martín Bernal, N Luna Pelaez, A Díez Lloret, R Perez Espejo, K Kamenarova, N Cuenca, **M Cortón**, C Ayuso, A Sorkio, H Skottman, S Erceg, S Bhattacharya. Human iPS cell disease model of MERTK-associated retinitis pigmentosa. Sci Rep. 2015 Aug 11;5:12910.
20. K Nikopoulos*, A Avila-Fernandez*, **M Corton**, MI Lopez-Molina, R Perez-Carro, L Bontadelli, A Di Gioia, O Zurita, B Garcia-Sandoval, C Rivolta*, C Ayuso*. Identification of two novel mutations in CDHR1 in consanguineous Spanish families with autosomal recessive retinal dystrophy. Sci Rep. 2015 Sep 9;5:13902
21. B Almoguera, J Li, P Fernandez-San Jose; Y Liu, M March, R Pellegrino, R Golhar, **M Corton**, F Blanco-Kelly, MI López-Molina, B García-Sandoval, Y Guo, L Tian, X Liu, L Guan, J Zhang; B Keating, X Xu, H Hakonarson, CAyuso. Identification of a novel deletion in PRPF31 as the genetic cause of Retinitis Pigmentosa using whole exome sequencing. PLoS One. 2015 Jul 21;10(7
22. Castro-Sánchez S, Álvarez-Satta M, **Cortón M,** Guillén E, Ayuso C, Valverde D.Exploring genotype-phenotype relationships in Bardet-Biedl syndrome families. J Med Genet. 2015 Jun 16.
23. -T Chassine*, B Bocquet*, V Daien, A Avila-Fernandez, C Ayuso, R Collin, **M Corton**, F Hejtmancik, I van den Born, J Klevering, A Riazzuddin, N Sendon, A, Isabelle Meunier, CP. Hamel. Autosomal recessive retinitis pigmentosa with RP1 mutations is associated with myopia. British Journal of Ophthalmology. 2015 Apr 16.
24. Avila-Fernandez A*, Perez-Carro R*, **Corton M**, Lopez-Molina MI, Campello L, Garanto A, Fernadez-Sanchez L, Duijkers L, Lopez-Martinez MA, Riveiro-Alvarez, Rodrigues-Jacy da Silva L, Sanchez-Alcudia R, Martin-Garrido E, Garcia-Garcia F, Dopazo J, Garcia-Sandoval B, Collin RW, Cuenca N, Ayuso C.Mutations in the ZNF408 Gene Cause Autosomal Recessive Retinitis Pigmentosa with Vitreal Alterations. Hum. Mol. Genet.
25. P Fernandez-San Jose, **M Cortón**, F Blanco-Kelly, A Ávila-Fernández, MAl Lopez-Martinez, R Sánchez-Alcudia, I Sanchez-Navarro, R Perez-Largo, O Zurita, N Sanchez-Bolivar, MI Lopez-Molina, B García-Sandoval, R Riveiro-Alvarez, C Ayuso. Targeted Next generation sequencing improves the diagnosis of Autosomal Dominant Retinitis Pigmentosa in Spanish patients. IOVS. 2015 Apr;56(4):2173-82.
26. B Almoguera*, J Liang*, **M Corton**, P Fernandez-San Jose, F Blanco-Kelly, MI Lopez-Molina, B Garcia-Sandoval, J del Val, Y Guo, L Tian, X Liu, L Guan, RJ Torres, JG Puig, H Hakonarson, X Xu, B Keating and C Ayuso. Expanding the phenotype of PRPS1 syndromes: Retinitis pigmentosa, neuropathy and hearing loss in females. Orphanet J Rare Dis. 2014 Dec 10;9(1):190.
27. R Sánchez-Alcudia, **M Corton**, A Ávila-Fernández, O Zurita, SD Tatu, R Pérez-Carro, P Fernandez-San Jose, MA Lopez-Martinez, FJ Del Castillo, JM Millan, F Blanco-Kelly, B García-Sandoval, MI Lopez-Molina, R Riveiro-Alvarez, C Ayuso. Retinal Dystrophies Challenge: Intrafamilial Genetic Heterogeneity in Spanish Population. Invest Ophthalmol Vis Sci. 2014 Oct 23;55(11):7562-71.

28. Fernandez-San Jose P, Blanco-Kelly F, **Corton M**, Trujillo-Tiebas MJ, Riveiro-Alvarez R, Lopez-Molina MI, Gimenez A, Avila-Fernandez A, Garcia-Sandoval B, Hernan I, Carballo M, and Ayuso C. RHO mutations in Spanish population and associated phenotypes. Acta Ophthalmologica 2015 Feb;93(1).

29. KM. Nishiguchi*, A Avila-Fernandez*, van Huet RA***, M Corton**, 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 amplifies the phenotypic spectrum for ABHD12 mutations: from PHARC syndrome to non-syndromic retinal degeneration. Ophthalmology. 2014 Aug;121(8):1620-7.

30. **M Corton**; A Avila-Fernandez, E Vallespin; MI Lopez-Molina; B Almoguera; E Martín-Garrido; MI Khan; F Blanco-Kelly; R Riveiro-Alvarez; M Brión; B García-Sandoval; FPM Cremers; A Carracedo; C Ayuso. Involvement of LCA5 in Leber congenital amaurosis and retinitis pigmentosa in the Spanish Population. Ophthalmology. 2014 Jan;121(1):399-407.

31. G Manes*, I Meunier*, A Avila-Fernández, S Banfi, G Le Meur, X Zanlonghi, **M Corton**, F Simonelli, P Brabet, G Labesse, I Audo, S Mohand-Said, C Zeitz, JA Sahel, M Weber, H Dollfus, CM Dhaenens, D Allorge, E De Baere, RK Koenekoop, S Kohl, FPM Cremers, JG Hollyfield, A Sénéchal, M Hebrard, B Bocquet, C Ayuso, and CP Hamel. Mutations in IMPG1 cause vitelliform macular dystrophies. * Equal contribution. Am J Hum Genet. 2013 Sep 5;93(3):571-8

32. Kamenarova K, **Cortón M**, García-Sandoval B, Fernández P, Panchev V, Lopez-Molina MI, Chakarova C, Ayuso C, and BhattacharyaSS. Novel mutations in GUCA1A suggesting different mechanisms of pathogenesis. BioMed Research International (J. Biomed Biotech) 2013;2013:517570.

33. **M Corton,** KM Nishiguchi, A Avila-Fernández, R Riveiro-Alvarez, SD Tatu, C Ayuso, C Rivolta. Exome sequencing of individual patients with retinal dystrophies as a tool for molecular diagnosis. PLoS One. 2013 Jun 14;8(6):

34. R Riveiro-Alvarez, MA Lopez-Martinez, D Cantalapiedra, A Avila-Fernandez, F Blanco-Kelly, A. Gimenez, MI Lopez-Molina, B Garcia-Sandoval, **M Corton**, S Tatu, MJ Trujillo-Tiebas, C Ramos and C Ayuso. Outcome of ABCA4 disease-associated alleles in autosomal recessive Stargardt disease (arSTGD), autosomal recessive Cone-Rod Dystrophies (arCRD) and autosomal recessive Retinitis Pigmentosa (arRP): retrospective analysis in 333 Spanish families. Ophthalmology. 2013 Nov;120(11):2332-7.

35. **Corton M**, Tatu SD, Avila-Fernandez A, Vallespín E, Tapias I, Cantalapiedra D, Blanco-Kelly F, Riveiro-Alvarez R, Bernal S, García-Sandoval B, Baiget M and Ayuso C. High frequency of CRB1 mutations as cause of Early-Onset Retinal Dystrophies in the Spanish population. Orphanet J Rare Dis. 2013 Feb 5;8:20.

36. A Avila-Fernandez*, **M Corton***, KM Nishiguchi, N Munoz-Sanz, B Benavides-Mori, F Blanco-Kelly, R Riveiro-Alvarez, B Garcia-Sandoval, C Rivolta and C Ayuso. A RP1 common founder mutation is a major cause of Early-onset Autosomal Recessive Retinitis Pigmentosa in Spanish population. Ophthalmology. 2012 Dec;119(12):2616-21.

37. T Piñeiro-Gallego, **M Cortón**, C Ayuso, M Baiget, D Valverde. Molecular approach in the study of Alstrom syndrome: analysis of ten Spanish families. Mol Vis. 2012;18:1794-802.

38. F Blanco-Kelly, M García-Hoyos, **M Corton**, A Ávila-Fernández, R Riveiro-Álvarez, A Giménez, I Hernan, M Carballo and C Ayuso. Genotyping microarray: mutation screening in spanish families with autosomal dominant retinitis pigmentosa. Molecular Vision. 2012;18:1794-802.

39. A Avila-Fernandez; **M Corton**; MI Lopez-Molina; E Martin-Garrido; D Cantalapiedra; R Fernandez-Sanchez; F Blanco-Kelly; R Riveiro-Alvarez; SD Tatu; MJ Trujillo-Tiebas; B Garcia-Sandoval; FPM Cremers; C Ayuso. Late Onset Retinitis Pigmentosa. Ophthalmology. 2011 Dec;118(12):2523-4.

40. Zabala-Fernández W, Barreiro-de Acosta M, Echarri A, Carpio D, Lorenzo A, Castro J, Martínez-Ares D, Pereira S, Martin-Granizo I, **Corton M**, Carracedo A, Barros F. A pharmacogenetics study of TPMT and

ITPA genes detects a relationship with side effects and clinical response in patients with inflammatory bowel disease receiving Azathioprine. J Gastrointestin Liver Dis. 2011 Sep;20(3):247-53.

41. JM Millán, E Aller, T Jaijo, F Blanco-Kelly, **M Corton**, and C Ayuso Chapter 2: Molecular Epidemiology of Usher Syndrome. In: "Usher syndrome: Pathogenesis, Diagnosis, and Therapy" Editor Satpal Ahuja, Publishers, Nova Science Science Publishers, Inc. USA. ISBN 978-1-61209-227-0

42. Aguirre-Lamban J, Gonzalez-Aguilera JJ, Riveiro-Alvarez R, Cantalapiedra D, Avila-Fernandez A, Villaverde C, **Corton M**, Blanco-Kelly F, Garcia-Sandoval B, Ayuso C. Further associations between mutations and polymorphisms in the ABCA4 gene: clinical implication of allelic variants and their role as protector/risk factors. Invest Ophthalmol Vis Sci. 2011 Aug 5;52(9):6206-12.

43. Avila-Fernandez A, Cantalapiedra D, Aller E, Vallespin E, Aguirre-Lamban J, Blanco-Kelly F, **Cortón M**, Riveiro-Alvarez R, Allikmets R, Trujillo-Tiebas MJ, Millan J, Cremers F, and Ayuso C. Mutation Analysis of 272 Spanish Families Affected by Autosomal Recessive Retinitis Pigmentosa Using a Genotyping Microarray. Mol Vis. 2010 Dec 3;16:2550-8.

44. Brión M, Sanchez-Salorio M, **Cortón M**, de la Fuente M, Pazos B, Sobrino B, Carracedo A. New update on the Genetics of Age-Related Macular Degeneration. Acta Ophtalmologica Scandinava. 2010 Nov 25.

45. **Corton M**, Blanco MJ, Torres M, Sanchez-Salorio M, Carracedo A, Brion M. Identification of a novel mutation in the human PDE6A gene in autosomal recessive Retinitis Pigmentosa. Homology with the nmf28/ nmf28 mice model. Clin Genet. 2010 Nov;78(5):495-498.

46. Chen W, Stambolian D, Edwards AO, Branham KE, Othman M, Jakobsdottir J, Tosakulwong N, Pericak-Vance MA, Campochiaro PA, Klein ML, Tan PL, Conley YP, Kanda A, Kopplin L, Li Y, Augustaitis KJ, Karoukis AJ, Scott WK, Agarwal A, Kovach JL, Schwartz SG, Postel EA, Brooks M, Baratz KH, Brown WL; Complications of Age-Related Macular Degeneration Prevention Trial Research Group, Brucker AJ, Orlin A, Brown G, Ho A, Regillo C, Donoso L, Tian L, Kaderli B, Hadley D, Hagstrom SA, Peachey NS, Klein R, Klein BE, Gotoh N, Yamashiro K, Ferris Iii F, Fagerness JA, Reynolds R, Farrer LA, Kim IK, Miller JW, **Cortón M**, Carracedo A, Sanchez-Salorio M, Pugh EW, Doheny KF, Brion M, Deangelis MM, Weeks DE, Zack DJ, Chew EY, Heckenlively JR, Yoshimura N, Iyengar SK, Francis PJ, Katsanis N, Seddon JM, Haines JL, Gorin MB, Abecasis GR, Swaroop A. Genetic Variants near TIMP3 and HDL-Associated Loci Influence Susceptibility to Age-Related Macular Degeneration. Proc Natl Acad Sci U S A. 2010 Apr 20;107(16):7401-6.

47. Escobar-Morreale HF, Insenser M, **Cortón M**, San Millán JL, Peral B. Proteomics and genomics: a hypothesis-free approach to the study of the role of visceral adiposity in the pathogenesis of the polycystic ovary syndrome. Proteomics Clin. Appl. 2008, 2, 444–455.

48. **Cortón M**, Botella-Carretero JI, Villuendas G, López JA, Camafeita E, San Millán JL, Escobar-Morreale HF, Peral B. Proteomic Analysis Of Omental Adipose Tissue In Polycystic Ovary Syndrome And Obesity Using 2-D DIGE And MS. Hum Reprod. 2008 Mar; 23(3):651-61.

49. Sanchez-Carbayo M, Socci ND, Richstone L, **Corton M**, Behrendt N, Wulkfuhle J, Bochner B, Petricoin E, Cordon-Cardo C. Genomic and Proteomic Profiles Reveal the Association of Gelsolin to TP53 Status and Bladder Cancer Progresión. American Journal of Pathology, 2007, 171(5): 1650-1658.

50. Orenes-Piñero E*, **Cortón M***, González-Peramato P, Algaba F, Casal I, Alvaro Serrano O, Sanchez-Carbayo M. Searching Urinary Tumor Markers for Bladder Cancer Using a Two-Dimensional Differential Gel Electrophoresis (2D-DIGE) Approach. Journal of Proteome Research 2007, 6, 4440-4448.

51. **Cortón M**, Botella-Carretero JI, Benguría A, Villuendas G, Zaballos A, San Millán JL, Escobar-Morreale HF, Peral B. Differential Gene Expression Profile in Omental Adipose Tissue in Women with Polycystic Ovary Syndrome. J Clin Endocrinol Metab. 92(1):328-37 (2007).

52. **Cortón, M**, Villuendas G, Botella J, San Millán JL, Escobar-Morreale H, Peral B. Improved resolution of the human adipose tissue proteome at alkaline and wide range pH by the addition of hydroxyethil disulfide. Proteomics, 4: 438-441 (2004).

53. San Millán JL, **Cortón M**, Villuendas G, Sancho J, Peral B, Escobar-Morreale H. Association of the Polycystic Ovary Syndrome (PCOS) with Genomic Variants Related to Insulin Resistance, Diabetes Mellitus and Obesity. J Clin Endocrinol Metab. 89(6): 2640-6 (2004).