

1. Genetic fine localization of the arrestin gene 5cM distal from D2S172 on chromosome 2q35-q37. **Valverde D.**, Bayes M., Martínez I., Grinberg D., Vilageliu L., Balcells S., Gonzalez R., Baiget M. *Human Genetics*. (1994) 94: 193-194. IF: 3,66.
2. Homozygous tandem duplication within the gene encoding the beta subunit of rod phosphodiesterase as a cause for autosomal recessive retinitis pigmentosa. Bayés M., Giordano M., Balcells S., Grinberg D., Vilageliu L., Martínez I., Ayuso C., Benitez J., Ramos MA., Chivelet P., Solans T., **Valverde D.**, Amselem S., Goosens M., Baiget M., Gonzalez R., Besmond C. *Human Mutation*. (1995) 5: 228-234. IF: 6,474.
3. Retinitis Pigmentosa in Spain. Ayuso C, García-Sandoval B, Najera C, **Valverde D**, Carballo M, Antiñolo G. *Clinical Genetics*. (1995) 48: 120-122. IF: 3,140.
4. Identificación de la mutación Arg 135-Leu en el gen de la rodopsina en una familia con Retinosis Pigmentaria Autosómica Recesiva. Carballo M., Reig C., Antich C., **Valverde D.**, Baiget M. *Medicina Clínica*. (1996) 106: 219-221. IF: 1,327.
5. Evidence against involvement of recoverin in autosomal recessive retinitis pigmentosa in 42 spanish families. Bayés M., **Valverde D.**, Balcells S., Grinberg D., Vilageliu L., Benitez J., Ayuso C., Beneyto M., Baiget M., Gonzalez-Duarte R. *Human Genetics*. (1995) 96: 89-94. IF: 3,66
6. A novel mutation in exon 17 of the  $\beta$  subunit of rod phosphodiesterase in two RP sisters of a consanguineous family. **Valverde D.**, Solans T., Grinberg D., Balcells S., Vilageliu L., Bayés M., Chivelet P., Besmond C., Goossens M., Gonzalez -Duarte R., Baiget M. *Human Genetics*. (1996) 97: 35-38. IF:3,66
7. Chromosome fragments with alphoid sequences derived from a pseudoisodicentric Y chromosome. Fernández JL., **Valverde D.**, Gosálvez J., Piñeiro C., Pereira S., Goyanes V. *Journal of Medical Genetics*. (1996) 33: 84-88. IF: 5,087
8. Identificaton of a novel Arg552Gln mutation in exon 13 of the  $\beta$  subunit of rod phosphodiesterase gene in a Spanish family with autosomal recessive Retinitis Pigmentosa. **Valverde D.**, Baiget M., Seminago R., del Río E., García-Sandoval B., del Río T., Bayés M., Martínez A., Grinberg D., Ayuso C. *Human Mutation*. (1996): 393-394. IF: 6,474
9. Autosomal recessive retinitis pigmentosa in Spain: evaluation of four genes and two loci involved in the disease. Bayes M, Martínez-Mir A, **Valverde D**, del Rio E, Vilageliu L, Grinberg D, Balcells S, Ayuso C, Baiget M, González-Duarte R. *Clinical Genetics*. (1996) 50: 380-387. IF: 3,140.
10. Alu I in situ digestion of human alphoid and classical satellite DNA regions. High-resolution digital image analysis of FISH signals from condensed and extended chromatin. JL. Fernandez, **D. Valverde**, V. Goyanes, I. Buño, J. Gosálvez. *Cytogenetics Cell Genetics*. (1997) 76: 94-100. IF: 1,993.
11. Putative association of a mutant ROM 1 allele with retinitis pigmentosa. Martínez A., Vilela C., Baiget M., **Valverde D.**, Dain L., Beneyto M., Marco M., Grinberg D., Balcells S., Gonzalez R., Vilageliu L. *Human Genetics*. (1997) 99: 827-830. IF: 3,66.
12. Análisis mutacional en 17 afectos gallegos de Fibrosis Quística. Vazquez-Gundín F., Mosquera A., Bricchette I., Dorado S., Ferreiro B., Fernandez JL, **Valverde D.** *Real Academia Gallega de Ciencias*. (1997) XVI: 43-49.

13. Identificación de dos mutaciones alélicas en el gen de la subunidad beta de la fosfodiesterasa en una familia española afectada de retinosis pigmentaria autosómica recesiva. Baiget M., Calaf M., **Valverde D.**, del Río E., Reig C., Carballo M., Calvo MT, Gonzalez-Duarte R. *Medicina Clínica.* (1998) 11: 420-423. Índice de Impacto: 1,327.
14. Analysis of the IRBP gene as a cause of RP in 45 ARRP Spanish families. **Valverde D.**, Vazquez-Gundin F., del Río E., Calaf M., Fernandez JL., Baiget M. *Ophthalmic Genetics.* (1998) 19: 203-207.
15. Two novel mutations in the retinitis pigmentosa GTPase regulator (RPGR) gene in Xlinked retinitis pigmentosa (RP3). Miano MG., **Valverde D.**, Solans T., Grammatico B., Migliaccio C., Cirigliano V., DeBernardo C., Ventruto V., Meitinger T., Wright A., Del Porto G., Baiget M., D'Urso M., Ciccodicola A. *Human Mutation.* (1998) 12: 212-213. IF: 6,474.
16. Síndrome de Bardet-Biedl. **Valverde D.** *Visión.* (1998) 13: 12-13.
17. True hermaphroditism and normal male external genitalia: a rare presentation. Montero M., Méndez R., **Valverde D.**, Fernandez JL., Gómez M., Ruiz C. *Acta Pediatrica.* (1999) 88: 909-914. IF: 1,297.
18. Haemochromatosis in Galicia (NW Spain): A celtic influence?. Soto L., Vega A., Goyanes V., **Valverde D.** *Clinical Genetics.* (2000) 57: 454-455. IF: 3,140
19. Mutations in the third exon of the MYOC gene in Spanish patients with primary open-angle glaucoma. Cabana M., Olmedo MV., Baiget M., **Valverde D.** *Ophthalmic Genetics.* (2000) 21:109-115.
20. Evaluation of RLBP1 in 50 Spanish families with autosomal recessive retinitis pigmentosa and 4 retinitis punctata albescens. Bernal S., Calaf M., Adan A., Solans T., **Valverde D.**, Ayuso C., Baiget M. *Ophthalmic Genetics.* (2001) 22: 19-25.
21. Mutations including the promoter region of myocilin/TIGR gene. Saura M., Cabana M., Ayuso C., **Valverde D.** *European Journal Human Genetics* (2005)13, 384-387. IF: 3,697.
22. Clinical and genetic studies in Spanish patients with usher syndrome type II: description of new mutations and evidence for a lack of genotype-phenotype correlation. Bernal S., Medà C., Solans T., Ayuso C, Garcia-Sandoval B, **Valverde D.**, Del Río E., Baiget M. *Clinical Genetics* (2005) 68:204-214. IF: 3,140.
23. Sequence variations in the retinal fascin FSCN2 gene in a Spanish population with autosomal dominant retinitis pigmentosa or macular degeneration. Gamundi MJ., Hernan I., Maseras M., Baiget M., Ayuso C., Borrego S., Antiñolo G., Millán JM., **Valverde D.**, Carballo M. *Molecular Vision* (2005) 11:922-928. IF: 2,377.
25. Mutation profile of the MYO7A gene in Spanish patients with Usher syndrome type I. Jaijo T., Oltra S., Beneyto M., Nájera C., Ayuso C., Baiget M., Carballo M., Antiñolo G., **Valverde D.**, Moreno F., Vilela C., Perez-Garrigues H., Navea A., Millan JM. *Human Mutation* (2006) *Mutation in brief* 875. IF: 6,474.
26. Microarray-based mutation analysis of the ABCA4 gene in Spanish patients with Stargardt disease: evidence of a prevalent mutated allele. **Valverde D.**, Riveiro R, Bernal S, Jaakson K, Baiget M, Navarro R, Ayuso C. *Molecular Vision* (2006); 12:902-908. IF: 2,377.
27. Identification of 14 novel mutations in the long isoform of USH2A in Spanish patients with Usher syndrome type II. Aller E., Jaijo T., Beneyto M., Nájera C., Oltra S., Ayuso C., Baiget

M., Carballo M., Antiñolo G., **Valverde D**, Moreno F., Vilela C., Collado D., Perez-Garrigues H., Navea A., Millan JM. *Journal Medical Genetics* (2006); 43: e55. doi: 10.1136. IF: 5,087.

28. Novel human pathological mutations. Gene symbol: ABCA4. Riveiro-Alvarez R., Trujillo MJ., Cantalapiedra D., Vallespin E., Villaverde C., **Valverde D**, Ayuso C. *Human Genetics* (2006); 119: DOI 10.1007/s00439-006-0179-0. IF: 3,66.

29. Spectrum of the ABCA4 gene mutations implicated in severe retinopathies from Spanish patients. **Valverde D**, Riveiro-Alvarez R., Baiget M., Carballo M., Antiñolo G., Millán JM, Garcia Sandoval B, Ayuso C. *IOVS* (2007) 48: 985-990. IF: 3,766.

30. Partial paternal uniparental disomy (UPD) of chromosome 1 in a one patient with Stargardt disease. R.Riveiro-Alvarez, **D.Valverde**, I.Lorda-Sanchez, MJ Trujillo-Tiebas, D.Cantalapiedra, E.Vallespin, J.Aguirre-Lamban, C.Ramos, C.Ayuso. *Molecular Vision* (2007) 13:96-101. IF: 2,377.

31. Absence of Activating Mutations in the EGFR Kinase Domain in Spanish Head and Neck Cancer Patients. Y. Lemos-González, M. Páez de la Cadena, F.J. Rodríguez-Berrocal, A.M. Rodríguez-Piñeiro, E. Pallas, **D. Valverde**. *Tumour Biology* (2007) 28: 273-279. IF: 2,407.

32. Mutations in the gene encoding bone morphogenetic protein receptor 2 in patients with idiopathic pulmonary arterial hypertension. Baloiira A, Vilariño C, Leiro V, **Valverde D**. *Arch Bronconeumol.* (2008) 44(1):29-34. IF: 1,624.

33. Influence of glutathione S-transferase M1 and T1 homozygous null mutations on the risk of antituberculosis drug-induced hepatotoxicity in a Caucasian population. Leiro V, Fernández-Villar A, **Valverde D**, Constenla L, Vázquez R, Piñeiro L, González-Quintela A. *Liver Int.* (2008) 28(6): 835-839. IF: 2,908.

34. Complexity of phenotype-genotype correlations in Spanish patients with RDH12 mutations. **Valverde D**, Pereiro I., Vallespín E., Ayuso C., Borrego S, Baiget M. *IOVS.* (2009) 50(3):1065-8. IF: 3,766.

35. Glutathione S-transferase M1 and T1 null genotypes increase susceptibility to drug induced liver injury. Leiro V., Fernández-Villar A., Vázquez-Gallardo R., **Valverde D**. *Hepatology* (2009) 49: 1777. IF: 11,355.

36. New mutations in BBS genes in small consanguineous families with Bardet-Biedl syndrome: Detection of candidate regions by homozygosity mapping. Pereiro I, **Valverde D**, Piñeiro-Gallego T, Baiget M, Borrego S, Ayuso C, Searby, CC, Nishimura DY. *Mol Vis* (2010); 16:137-143. IF: 1,85.

37. Discovery and functional analysis of a novel retinitis pigmentosa gene, C2ORF71. DY Nishimura, LM Baye, R Perveen, CC Searby, A Avila-Fernandez, I Pereiro, C Ayuso, **D Valverde**, PN Bishop, FD Manson, J Urquhart, EM Stone, DC Slusarski, GC Black, VC Sheffield. *Am. J. Hum. Genet* (2010).86: 686-695. IF: 12,3.

38. Genetic variations of NAT2 and CYP2E1 and isoniazid hepatotoxicity in a diverse population. Leiro-Fernandez V, **Valverde D**, Vázquez-Gallardo R, Constenla L, Fernández-Villar A. *Pharmacogenomics* (2010), 11: 1205-1206. IF: 3,89.

39. Arrayed Primer Extension (APEX) technology simplifies mutation detection in Bardet Biedl and Alström Syndrome. I Pereiro, BE. Hoskins, JD. Marshall, GB. Collin, JK. Naggert, T

Piñeiro-Gallego, E Oitmaa, N Katsanis, **D Valverde**, PL. Beales. Eur. J. Hum. Genet. (2011); 19: 485-488. IF: 3,5.

40. NAT2 polymorphisms and risk of antituberculosis drug-induced hepatotoxicity in Caucasians. Leiro-Fernandez V, **Valverde D**, Vázquez-Gallardo R, Botana-Rial M, Constenla L, Agúndez JA, Fernández-Villar A. The International Journal of Tuberculosis and Lung Disease (2011) 15 (10):1403-1408. IF?

41. Clinical evaluation of two consanguineous families with homozygous mutations in BEST1. Piñeiro-Gallego T, Álvarez M, Pereiro I, Campos S, Dror Sharon, Patrik Schatz, **Valverde D**. Mol Vis (2011), 17:1607-1617. F?

42. Polymorphisms in the serotonin transporter protein (SERT) gene in patients with pulmonary arterial hypertension. Balóira A, Núñez M, Cifrián J, Vilariño C, Ojeda M, **Valverde D**. Arch Bronconeumol. (2012) Mar;48(3):77-80. IF: 1,6.

43. Association of CTLA4 gene polymorphism with ophthalmopathy of Graves disease in Spanish population. Álvarez-Vázquez P., Constenla L., García-Mayor R. **Valverde D**. International Journal Of Endocrinology and Metabolism (2011); 9: 397- 402. F?

44. Molecular approach in the study of Alström Syndrome: analysis of ten Spanish families.

Teresa Piñeiro-Gallego, Marta Cortón, Carmen Ayuso, Montserrat Baiget, Diana **Valverde**. Mol Vis (2012) 18: 1794-1802. F?

45. Prognostic value of aberrant hypermethylation in pleural effusion of lung adenocarcinoma. Botana-Rial M, de Chiara L, **Valverde D**, Leiro-Fernández V, Represas-Represas C, Del Campo-Pérez V, Fernández-Villar A. Cancer Biol Ther. (2012) 6;13(14). IF: 1,04.

46. Bardet-Biedl syndrome: A rare genetic disease. Diana Valverde, Sheila Castro-Sánchez and María Álvarez-Satta. Pediatric Genetics (2013)2:77-83. F?

47. Overview of Bardet-Biedl syndrome in Spain: identification of novel mutations in BBS1, BBS10 and BBS12 genes. Alvarez-Satta M, Castro-Sánchez S, Pereiro I, Piñeiro-Gallego T, Baiget M, Ayuso C, **Valverde D**. Clin Genet (2014) 86:601-602. IF: 3.931.

48. Novel mutations in BMPR2, ACVRL1 and KCNA5 genes and hemodynamic parameters in patients with pulmonary arterial hypertension. Pousada G, Balóira A, Vilariño C, Cifrián JM, **Valverde D**. PLoS One (2014) 9(6):e100261. doi: 10.1371/journal.pone.0100261. IF: 3.234.

49. Viabilidad de las muestras ganglionares obtenidas por ecobroncoscopia para el estudio de alteraciones epigenéticas en pacientes con cáncer de pulmón. Virginia Leiro-Fernández, Loretta De Chiara, Maribel Botana-Rial, Ana González-Piñeiro, Antoni Tardio-Baiges, Manuel Núñez-Delgado, Diana **Valverde**, Alberto Fernández-Villar. Arch Bronconeumol (2014) 50:213-220. IF: 1.823.

50. Estudio de la repetición del pentanucleótido CCTTT en el gen de la sintasa inducible del óxido nítrico en pacientes con hipertensión arterial pulmonar. Adolfo Balóira Villar, Guillermo Pousada Fernández, Carlos Vilariño Pombo, Marta Núñez Fernández, Jose Cifrián Martínez, Diana **Valverde**. Arch Bronconeumol (2014) 50:141-145. IF: 1.823.

51. Algorithm for the molecular analysis of Bardet-Biedl syndrome in Spain. Castro-Sánchez S, Alvarez-Satta M, Pereiro I, Piñeiro-Gallego MT, **Valverde D**. Med Clin (Barc) (2015) 145:147-152. IF: 1.417.

52. K198N polymorphism in the gene EDN1 in patients with pulmonary arterial hypertension. Pousada G, Balóira A, Vilaríño C, **Valverde D**. *Med Clin (Barc)* (2015); 144:348-352. IF: 1.417.
53. Pulmonary arterial hypertension and portal hypertension in a patient with hereditary hemorrhagic telangiectasia. Guillermo Pousada, Adolfo Balóira, Diana **Valverde**. *Med Clin (Barc)* (2015) 144:261-264. IF: 1.417.
54. Molecular and clinical analysis of TRPC6 and AGTR1 genes in patients with pulmonary arterial hypertension. Pousada G, Balóira A, **Valverde D**. *Orphanet J Rare Dis* (2015) 10:1. doi: 10.1186/s13023-014-0216-3. IF: 3.358.
55. Estudio clínico y molecular de 4 casos de hipertensión arterial pulmonar asociada a Sarcoidosis. Balóira-Villar A, Pousada-Fernández G, Núñez-Fernández M, **Valverde-Pérez D**. *Arch Bronconeumol* (2015) 51:19-21. IF: 1.823.
56. Exploring genotype-phenotype relationships in Bardet-Biedl syndrome families. Castro-Sánchez S, Álvarez-Satta M, Cortón M, Guillén E, Ayuso C, **Valverde D**. *J Med Genet* (2015) 52:503-513. IF: 6.335.
57. Prevalence study of the genetic markers associated with slow progression of human immunodeficiency virus type 1 in the Galician population (Northwest of Spain). Rodríguez-Da Silva A, Miralles C, Ocampo A, **Valverde D**. *Enferm Infecc Microbiol Clin.* (2015) pii: S0213-005X(15)00153-6. doi: 10.1016/j.eimc.2015.04.006 2015. IF: 2.172.
58. Complex inheritance in Pulmonary Arterial Hypertension patients with several mutations. Pousada G, Balóira A, **Valverde D**. *Sci Rep.* (2016) Sep 15;6:33570. doi: 10.1038/srep33570. IF: 5.228.
59. HLA-DQ B1\*0201 and A1\*0102 Alleles Are Not Responsible for Antituberculosis Drug-Induced Hepatotoxicity Risk in Spanish Population. Leiro-Fernández V, **Valverde D**, Vázquez-Gallardo R, Constenla-Caramés L, Del Campo-Pérez V, Fernández-Villar A. *Front Med (Lausanne)*. (2016) Aug 22;3:34. doi: 10.3389/fmed.2016.00034. IF: 1.863
60. Pulmonary arterial hypertension associated with hereditary spherocytosis and splenectomy in a patient with a mutation in the BMPR2 gene. Balóira A, Bastos M, Pousada G, **Valverde D**. *Clin Case Rep.* (2016) Jun 30;4(8):752-5. doi: 10.1002/ccr3.610. **F?**
61. Mutational and clinical analysis of the ENG gene in patients with pulmonary arterial hypertension. Pousada G, Balóira A, Fontán D, Núñez M, **Valverde D**. *BMC Genet* (2016) Jun 4;17(1):72. doi: 10.1186/s12863-016-0384-3. IF: 2.152.
62. Pulmonary arterial hypertension associated with human immunodeficiency virus infection: study of 4 cases. Pousada G, Balóira A, Castro-Añón O, **Valverde D**. *Med Clin (Barc)*. (2016) Apr 15;146(8):350-3. doi: 10.1016/j.medcli.2015.12.014. IF: 1.267.
63. Methylation Analysis of the BMPR2 Gene Promoter Region in Patients With Pulmonary Arterial Hypertension. Pousada G, Balóira A, **Valverde D**. *Arch Bronconeumol* (2016) Jun;52(6): 293-8. doi: 10.1016/j.arbres.2015.10.006. 5. IF: 1.77.
64. Mutational screening in genes related to porto-pulmonary hypertension: An analysis of 6 cases. Pousada G, Balóira A, **Valverde D**. *Med Clin (Barc)* (2017) Feb 25. pii: S0025-7753(17)30091-X. doi: 10.1016/j.medcli.2017.01.020. [Epub ahead of print]. IF: 1,267

65. Predictive value of spindle retardance in embryo implantation rate. García-Oro S, Rey MI, Rodríguez M, Durán Á, Devesa R, **Valverde D**. *J Assist Reprod Genet* (2017) Mar 9. doi: 10.1007/s10815-017-0897-3. [Epub ahead of print]. IF: 1,858
66. Functional analysis by minigene assay of putative splicing variants found in Bardet–Biedl syndrome patients. María Alvarez-Satta, Sheila Castro-Sánchez, Guillermo Pousada, Diana **Valverde**. *J. Cell. Mol. Med.* Vol XX, No X, 2017 pp. 1-8. IF: 4,938