CV Date	16/06/2025

Part A. PERSONAL INFORMATION

First Name *	Eladio Andrés		
Family Name *	Velasco Sampedro		
Sex *	Not Specified	Date of Birth *	
ID number Social Security, Passport *		Phone Number *	
URL Web			
Email Address			
Researcher's identification number	Open Researcher and Contributor ID (ORCID) *	0000-0002-9682-5589	
	Researcher ID	D-1432-2016	
	Scopus Author ID	57798202600	

* Mandatory

A.1. Current position

Job Title	Científico Titular		
Starting date	2008		
Institution	Consejo Superior de Investigaciones Científicas		
Department / Centre			
Country		Phone Number	(34) 983184829
Keywords	Molecular biology; Medical genetics; Molecular genetics; Genomics		

A.2. Previous positions

Period	Job Title / Name of Employer / Country
1999 -	Investigador contratado / Universidad de Valladolid

A.3. Education

Degree/Master/PhD	University / Country	Year
Programa Oficial de Doctorado en Bioquímica y Biología Molecular	Universidad Autónoma de Madrid	1996
Licenciado en Ciencias Biológicas Especialidad Biología Fundamental	Universidad Complutense de Madrid	1988

A.4. General quality indicators of scientific production

- 72 **articles in scientific journals**, **27 as corresponding author**:
  * 45 articles in top Q1 journals (14 of the first decile) / Average Impact Factor: 4.65
  * Top cited articles: Velasco et al, Hum Mol Genet 1996, 5:257-263 (189 citations); Sanz et al, Clin Cancer Res 2010, 16: 1957-1967 (95 citations); Diez et al, Hum Mutat 2004, 22: 301-312 (159 citations); Milne et al, Clin Cancer Res 2008, 14: 2861-2869 (95 citations); De la Hoya et al, Clin Chem 2006, 52: 1480-1485 (64 citations), Fraile-Bethencourt et al, PLoS Genetics 2017, 13: e1006691 (52 citations).
- h-index = 24 (Scopus), 26 (Google Scholar).
- Patent: “pSAD plasmid for splicing functional assays” (2012).
- Supervision of 9 PhD Thesis, seven of them awarded with the **Extraordinary PhD Award, University of Valladolid**.
- Supervision of 3 Masters Thesis and 1 Bachelor's Thesis.
- **Principal investigator** of 11 National and regional projects, 2 intramural projects, 1 of a private bank foundation and PI of participant #18 of an European H2020 grant (BRIDGES). EAV has been member of the Research Team of another 15 projects.

- Principal investigator or scientific coordinator of 26 contracts of technological support or agreements with private companies or public organisms. It is worth highlighting the contract with the Queensland Institute for Medical Research (Australia) "Splicing analysis of the TP53 gene by hybrid minigenes"; Amount: 60,000 €.
- **Topic editor** of Frontiers in Genetics (IF=4.56): **RNA Splicing and Backsplicing: Disease and Therapy**
- **Academic Editor** of Cancers (IF= 6.64): **"Genetic Variants Associated with Breast and Ovarian Cancer Risk"**

Part B. CV SUMMARY

Dr. Velasco has focused his scientific career on the study of the molecular bases of hereditary diseases.

1) PhD. Doctoral thesis "Molecular Genetics of Spinal Muscular Atrophies (SMA)" (Extraordinary Doctoral Award, UAM). The most relevant contributions were the existence of linkage of Spanish SMA families to chromosome 5q markers and the discovery of a relationship between the number of copies of the SMN2 gene and the severity of the SMA phenotype (Velasco 1996). He also participated in the study of other hereditary diseases (10 publications, 4 as first author)

2) Cancer genetics.

- Characterization of the spectrum of BRCA1/2 mutations in patients with hereditary breast/ovarian cancer: identification of new DNA variants (4 articles, senior author in 2).

Innovation: For screening susceptibility genes, the high-throughput technique of fluorescent heteroduplex analysis in an automatic sequencer was designed (senior author of 3 articles, *Velasco 2007).

- Founder effect of BRCA1/2 variants: 4 variants showed a founder effect in Castilla y León. The most prevalent mutation in BRCA2 arose from at least 3 different mutational events (hotspot) (senior author of 3 articles).
- National and international collaborations (8 publications) on the genetics of hereditary breast/ovarian cancer: distribution of mutations and rearrangements of BRCA1/2; risk associated with SNPs of TP53; role of PALB2 in pancreatic and male breast cancer.
- Identification and clinical interpretation of variants in hereditary colorectal cancer genes (5 publications).
- Study of the BRCA2 promoter. Using luciferase assays, activator and repressor regions were identified, as well as high variability in gene expression caused by DNA variants in the promoter (senior author, 1 publication)

3) Correlation between aberrant splicing and genetic susceptibility to cancer (main line of his research career).

He has received funding as Principal Investigator from 6 national ISCIII projects, 5 regional ones, 2 CSIC-intramural ones and the European H2020 BRIDGES project, and grants for hiring pre- and postdoctoral researchers.

Innovation. A splicing vector (pSAD–Splicing And Disease) was designed and patented, a fundamental tool for the construction of hybrid minigenes of the genes involved and the findings of this line of research.

Four key contributions stand out (24 publications, 17 as corresponding author):

- Analysis of the main genes for susceptibility to breast cancer

A systematic study of variants of the BRCA1/2 genes showed that splicing alterations are a frequent mechanism of pathogenicity and minigenes are a valuable strategy for studying splicing (2 publications as senior author, *Sanz et al 2010).

The largest splicing studies of variants of the main breast cancer susceptibility genes (functional assays of 706 variants) have been carried out using minigenes for BRCA1, BRCA2, PALB2, CHEK2, ATM, RAD51C and RAD51D, the last 5 within the European BRIDGES project. Aberrant transcripts are generated by different splicing events: exon skipping, use of alternative sites, intron retention, or combinations of these. Variant-induced splicing alterations are a prevalent deleterious mechanism: 433 variants (61%) affected splicing and 245 were classified

as pathogenic (clinically actionable), allowing for the adoption of personalized preventive/ therapeutic measures in carrier women. These pathogenic variants appear in more than 1600 independent records in the ClinVar database, underlining the health impact of these results.

- We also showed that splicing is finely regulated by a wide collection of sequences and factors:Functional mapping of 25 essential regulatory intervals in the BRCA2, RAD51D and CHEK2 genes enriched in splicing enhancers, where 108 spliceogenic variants were detected.
- Characterization of non-canonical 5'/3' splice sites: GC donors (<1% of human 5'SS) of exons 17-BRCA2, 12-PALB2 and 50-ATM, GG-donors-ATM (0.01% of 5'SS) and TG acceptor-CHEK2 (0.02% of 3'SS).
- Coordinator of the Minigenes Facility (http://www.ibgm.med.uva.es/servicios/). Through technological support contracts, other hereditary disease genes such as MLH1, COL1A1, SERPINA1, CHD7, GRN, UGT1A1, TRPM4 and TP53 have been studied, in collaboration with National and International institutions.
- Member of the International Consortium ENIGMA: Evidence-based Network for the Interpretation of Germline Mutant Alleles.

Quality Indicators (summary) **:** - Author of 72 articles (45 Q1, 14 D1, 27 as corresponding author; Average Impact Factor: 4.65)

- h-index = 24 (Scopus), 26 (Google Scholar).- Patent: splicing vector (pSAD–Splicing And Disease).
- Principal Investigator of 15 projects, including one European grant, and 26 technological support contracts.
- Director of the Consolidated Research Unit No. 236 of Castilla y León.
- Supervision of 9 doctoral theses (7 with Extraordinary PhD Award, University of Valladolid), 3 Master's and 2 Bachelor's thesis.

Part C. RELEVANT ACCOMPLISHMENTS

C.1. Publications

AC: corresponding author. (nº x / nº y): position / total authors. If applicable, indicate the number of citations

1 **Scientific paper**. Canson DM; Llinares-Burguet I; Fortuno C,; Sanoguera-Miralles L; Bueno-Martínez E; de la Hoya M; Spurdle AB; Velasco-Sampedro EA. 2025. TP53 minigene analysis of 161 sequence changes provides evidence for role of spatial constraint and regulatory elements on variant-induced splicing impact.Npj Genomic Medicine. 10, pp.37.

2 **Scientific paper**. Velasco-Sampedro EA; Sánchez-Vicente C; Caloca MJ. 2025. β2-Chimaerin Deficiency Favors Polyp Growth in the Colon of ApcMin/+ Mice. Molecules. 30, pp.824.

3 **Scientific paper**. Fortuno C; Llinares-Burguet I; Canson DM; et al; Spurdle AB. 2025. Exploring the role of splicing in TP53 variant pathogenicity through predictions and minigene assays. Human Genomics. 19, pp.2.

4 **Scientific paper**. Carmona-Carmona CA; Zini P; (3/6) Velasco-Sampedro EA; Cózar-Castellano I; Perdomo G; Caloca MJ. 2024. β2-Chimaerin, a GTPase-Activating Protein for Rac1, Is a Novel Regulator of Hepatic Insulin Signaling and Glucose Metabolism. Molecules. 29, pp.5301. https://doi.org/10.3390/molecules29225301

5 **Scientific paper**. Llinares-Burguet I; Sanoguera-Miralles L; Valenzuela-Palomo A; García-Álvarez A; Bueno-Martínez E; (6/6) Velasco-Sampedro EA (AC). 2024. Splicing Dysregulation of Non-Canonical GC-5′ Splice Sites of Breast Cancer Susceptibility Genes ATM and PALB2. Cancers. 16, pp.3562. https://doi.org/10.3390/cancers16213562

6 **Scientific paper**. Sanoguera-Miralles L; Llinares-Burguet I; Bueno-Martínez E; et al; (11/11) Velasco-Sampedro EA (AC). 2024. Comprehensive splicing analysis of the alternatively spliced CHEK2 exons 8 and 10 reveals three enhancer/silencer-rich regions and 38 spliceogenic variants. The Journal of Pathology. 262, pp.395-409. https://doi.org/10.1002/path.6243

7 **Scientific paper**. Sanoguera-Miralles L; Valenzuela-Palomo A; Bueno-Martínez E; et al; (14/14) Velasco-Sampedro EA (AC). 2024. Systematic Minigene-Based Splicing Analysis and Tentative Clinical Classification of 52 CHEK2 Splice-Site Variants.Clinical Chemistry. 70, pp.319-338. https://doi.org/10.1093/clinchem/hvad125

8 **Scientific paper**. Valenzuela-Palomo A.; Sanoguera-Miralles L; Bueno-Martínez E; et al; (10/10) Velasco-Sampedro EA (AC). 2022. Splicing Analysis of 16 PALB2 ClinVar Variants by Minigene Assays: Identification of Six Likely Pathogenic Variants. Cancers. 14, pp.4541. https://doi.org/10.3390/cancers14184541

9 **Scientific paper**. Bueno-Martínez E; Sanoguera-Miralles L; Valenzuela-Palomo A; et al; (15/15) Velasco-Sampedro EA (AC). 2022. Minigene-based splicing analysis and ACMG/AMP-based tentative classification of 56 ATM variants.The Journal of Pathology. 258, pp.83-101. https://doi.org/10.1002/path.5979

10 **Scientific paper**. Sanoguera-Miralles L; Bueno-Martínez E; Valenzuela-Palomo A; et al; (9/9) Velasco-Sampedro EA (AC). 2022. Minigene Splicing Assays Identify 20 Spliceogenic Variants of the Breast/Ovarian Cancer Susceptibility Gene RAD51C. Cancers. 14, pp.2960. https://doi.org/10.3390/cancers14122960

11 **Scientific paper**. Valenzuela-Palomo A.; Bueno-Martínez E.; Sanoguera-Miralles L.; et al; (17/17) Velasco EA (AC). 2022. Splicing predictions, minigene analyses, and ACMG-AMP clinical classification of 42 germline PALB2 splice-site variants.The Journal of Pathology. 256, pp.321-334. https://doi.org/10.1002/path.5839

12 **Scientific paper**. Zhu, L.; Miao, B.; Dymerska, D.; et al; Bandapalli, O.R.; Hemminki, K.; Försti, A; (7/10) Velasco EA. 2022. Germline Variants of CYBA and TRPM4 Predispose to Familial Colorectal Cancer.CANCERS. 14, pp.670. https://doi.org/10.3390/cancers14030670

13 **Scientific paper**. Bueno-Martínez E.; Sanoguera-Miralles L.; Valenzuela-Palomo A.; et al; (15/15) Velasco E.A. (AC). 2021. RAD51D Aberrant Splicing in Breast Cancer: Identification of Splicing Regulatory Elements and Minigene-Based Evaluation of 53 DNA Variants. Cancers. 13, pp.2845. https://doi.org/10.3390/cancers13112845

14 **Scientific paper**. Sanoguera-Miralles L.; Valenzuela-Palomo A.; Bueno-Martínez E.; et al; (16/16) Velasco E.A. (AC). 2020. Comprehensive Functional Characterization and Clinical Interpretation of 20 Splice-Site Variants of the RAD51C Gene. Cancers. 12, pp.3771. https://doi.org/10.3390/cancers12123771

15 **Scientific paper**. Gailite L; Valenzuela-Palomo A; Sanoguera-Miralles L; Rots D; Kreile M; (6/6) Velasco EA (AC). 2020. UGT1A1 variants c.864+5G>T and c.996+2_996+5del of a Crigler-Najjar patient induce aberrant splicing in minigene assays. Front Genet. 11, pp.169. https://doi.org/10.3389/fgene.2020.00169

16 **Scientific paper**. Fraile-Bethencourt E; Valenzuela-Palomo A; Díez-Gómez B; Caloca MJ; Gómez-Barrero S; (6/6) Velasco EA (AC). 2019. Minigene splicing assays identify 12 spliceogenic variants of BRCA2 exons 14 and 15. Front Genet. 10, pp.503. https://doi.org/10.3389/fgene.2019.00503

17 **Scientific paper**. Fraile-Bethencourt E; Valenzuela-Palomo A; Díez-Gómez B; Goina E; Acedo A; Buratti E; (7/7) Velasco EA (AC). 2019. Mis-splicing in breast cancer: identification of pathogenic BRCA2 variants by systematic minigene assays. The Journal of Pathology. 248, pp.409-420. https://doi.org/10.1002/path.5268

18 **Scientific paper**. 2019. Alternative splicing and ACMG-AMP-2015-based classification of PALB2 genetic variants: an ENIGMA report.J Med Genet. 56, pp.453-460. https://doi.org/10.1136/jmedgenet-2018-105834

19 **Scientific paper**. Montalban G; Fraile-Bethencourt E; López-Perolio I; et al; Gutiérrez-Enríquez S; (11/12) Velasco EA (AC). 2018. Characterization of spliceogenic variants located in regions linked to high levels of alternative splicing: BRCA2 c.7976+5G>T as a case study.Hum Mutat. 39, pp.1155-1160. ISSN 1059-7794. https://doi.org/10.1002/humu.23583

20 **Scientific paper**. Fraile-Bethencourt E; Valenzuela-Palomo A; Díez-Gómez B; et al; (9/9) Velasco EA (AC). 2018. Genetic dissection of the BRCA2 promoter and transcriptional impact of DNA variants. Breast Cancer Res Tr. https://doi.org/10.1007/s10549-018-4826-7

21 **Scientific paper**. Fraile-Bethencourt E; Valenzuela-Palomo A; Díez-Gómez B; Acedo A; (5/5) Velasco EA (AC). 2018. Identification of Eight Spliceogenic Variants in BRCA2 Exon 16 by Minigene Assays. Front Genet. 9, pp.188. https://doi.org/10.3389/fgene.2018.00188

22 **Scientific paper**. Fraile-Bethencourt E; Díez-Gómez B; Velásquez-Zapata V; Acedo A; Sanz DJ; (6/6) Velasco EA (AC). 2017. Functional classification of DNA variants by hybrid minigenes: Identification of 30 spliceogenic variants of BRCA2 exons 17 and 18. PLoS Genet. 13, pp.e1006691 [doi. 10.1371/journal.pgen.1006691]. https://doi.org/10.1371/journal.pgen.1006691

23 **Scientific paper**. Acedo A; Hernández-Moro C; Díez-Gómez B; Curiel-García A; (5/5) Velasco EA (AC). 2015. Functional classification of BRCA2 DNA variants by splicing assays in a large minigene with 9 exons. Hum Mutat. 36, pp.210-221. https://doi.org/10.1002/humu.22725

24 **Scientific paper**. Acedo A; Sanz DJ; Durán M; Infante M; Pérez-Cabornero L; Miner C,; (7/7) Velasco EA (AC). 2012. Comprehensive splicing functional analysis of DNA variants of the BRCA2 gene by hybrid minigenes. Breast Cancer Res. 14, pp.R87. https://doi.org/10.1186/bcr3202

25 **Scientific paper**. Sanz DJ; Acedo A; Infante M; et al; (10/10) Velasco EA (AC). 2010. A high proportion of DNA variants of BRCA1 and BRCA2 is associated with aberrant splicing in breast/ovarian cancer patients. Clin Cancer Res. 16, pp.1957-1967. https://doi.org/10.1158/1078-0432.CCR-09-2564

26 **Scientific paper**. Infante M,; Durán M,; Acedo A,; et al; (15/15) Velasco EA (AC). 2010. BRCA1 5272-1G>A and BRCA2 5374delTATG are founder mutations of high relevance for genetic counselling in breast/ovarian cancer families of Spanish origin.Clin Genet. 77, pp.60-69. https://doi.org/10.1111/j.1399-0004.2009.01272.x

27 **Scientific paper**. Milne RL; Osorio A; Ramón y Cajal T; et al; Benítez J.; (16/29) Velasco E. 2008. The average cumulative risks of breast and ovarian cancer for carriers of mutations in BRCA1 and BRCA2 attending genetic counselling units in Spain.Clinical Cancer Research. 14, pp.2861-2869. https://doi.org/10.1158/1078-0432.CCR-07-4436

28 **Scientific paper**. (1/7) Velasco E (AC); Infante M; Durán M; Pérez-Cabornero L; Sanz DJ; Esteban-Cardeñosa E; Miner C.2007. Heteroduplex analysis by capillary array electrophoresis for rapid mutation detection in large multiexon genes.Nature Protocols. 2, pp.237-246. https://doi.org/10.1038/nprot.2006.482

29 **Scientific paper**. Infante M; Durán M; Esteban-Cardeñosa E; Miner C; (5/5) Velasco E (AC). 2006. High proportion of novel mutations of BRCA1 and BRCA2 in breast/ovarian cancer patients from Castilla-León (Central Spain).Journal of Human Genetics. 51, pp.611-617. https://doi.org/10.1007/s10038-006-0404-7

C.2. Conferences and meetings

1 Llinares-Burguet I; Sanoguera-Miralles L; García-Álvarez A; Bueno-Martínez E; Velasco-Sampedro EA. ANÁLISIS DE VARIANTES +2T EN GENES DE SUSCEPTIBILIDAD A CÁNCER DE MAMA: GENERACIÓN DE DONADORES ATÍPICOS (NO CANÓNICOS) FUNCIONALES. [Comunicación Oral]. XVI Jornada de Actualización en Genética Humana. Nuevos Horizontes en Genómica y Cáncer Hereditario. Asociación Española de Genética Humana. 2024.

2 Sanoguera-Miralles L; Llinares-Burguet I; Bueno-Martínez E; et al; Velasco EA. Comprehensive splicing analysis of the alternatively spliced CHEK2 exons 8 and 10 by hybrid minigenes [Poster P01.059.C]. European Human Genetics Conference 2024. European Society of Human Genetics. 2024. Germany.

3 Llinares-Burguet I; Sanoguera-Miralles L; Bueno-Martínez E; García-Álvarez A; Velasco EA. Mapping of Splicing Regulatory Elements-rich intervals and identification of spliceogenic variants in ATM exon 7 [Poster P01.002.B]. European Human Genetics Conference 2024. European Society of Human Genetics. 2024. Germany.

4 Llinares-Burguet I; Sanoguera-Miralles L; Bueno-Martínez E; García-Álvarez A; Velasco-Sampedro EA. Identificación de Elementos Reguladores de splicing y variantes espliceogénicas en el exón 7 de ATM [Comunicación Oral]. X Jornadas de Investigadoras de Castilla y León, la Aventura de la Ciencia y la Tecnología. Universidad de Valladolid. 2024.

5 Sanoguera-Miralles L; Llinares-Burguet I; Bueno-Martínez E; Esteban Sánchez A; Valenzuela Palomo A; García-Álvarez A; de la Hoya M; Velasco Sampedro EA. Comunicación Oral: CARACTERIZACIÓN FUNCIONAL MEDIANTE MINIGENES REPORTEROS DE SPLICING E INTERPRETACIÓN CLÍNICA DE 52 VARIANTES DEL GEN CHEK2 (Premio a la mejor comunicación oral). IV Congreso Interdisciplinar en Genética Humana. Asociación Española de Genética Humana. 2023.

6 Sanoguera-Miralles L; Llinares-Burguet I; Bueno-Martínez E; Esteban Sánchez A; Valenzuela Palomo A; García-Álvarez A; de la Hoya M; Velasco Sampedro EA. Comunicación Oral: Análisis funcional mediante minigenes híbridos de las variantes del gen CHEK2 detectadas en el proyecto europeo BRIDGES. I Congreso Iberoamericano de Genética Médica y Medicina Genómica. ACMGen / AEGH / AMGH / RELAGH. 2023. Colombia.

7 Llinares-Burguet I; Sanoguera-Miralles L; Bueno-Martínez E; Esteban Sánchez A; García-Álvarez A; de la Hoya M; Velasco Sampedro EA. Comunicación Oral: Caracterización funcional de variantes candidatas de splicing en el gen de susceptibilidad a cáncer de mama ATM. I Congreso Iberoamericano de Genética Médica y Medicina Genómica. ACMGen / AEGH / AMGH / RELAGH. 2023. Colombia.

8 Valenzuela Palomo A; Llinares-Burget I; Sanoguera-Miralles L; Bueno-Martínez E; García-Álvarez A; Esteban A; de la Hoya M; Velasco EA. Comunicación Oral: DESREGULACIÓN DEL SPLICING DEL GEN PALB2 EN CÁNCER DE MAMA HEREDITARIO. XV Jornada de Actualización en Genética Humana. Asociación Española de Genética Humana. 2022.

C.3. Research projects and contracts

1 **Project**. Desregulación del splicing de macroexones y microexones en genes de susceptibilidad a cáncer de mama (Ref. PI23/00047). Instituto de Salud Carlos III. (IP) Eladio A. Velasco Sampedro. (Instituto de Biomedicina y Genética Molecular de Valladolid). 01/01/2024-31/12/2026. 91.250 €. Principal investigator.

2 **Project**. SPLICING ABERRANTE DE GENES DE SUSCEPTIBILIDAD A CÁNCER DE MAMA (Ref. 202320E182). CSIC. Proyectos Intramurales Especiales. (IP) Eladio A. Velasco Sampedro. (Instituto de Biomedicina y Genética Molecular de Valladolid). 01/11/2023-31/10/2024. 16.000 €. Principal investigator.

3 **Project**. Splicing regulation of alternative and atypical exons of breast cancer genes in physiological and DNA damage conditions: implications in disease susceptibility (Ref. PI20/00225). Instituto de Salud Carlos III. (IP) Eladio A. Velasco Sampedro. (Instituto de Biología y Genética Molecular (CSIC)). 01/01/2021-31/12/2023. 93.170 €. Principal investigator.

4 **Project**. Desregulación del splicing en cáncer de mama hereditario: análisis funcional de genes de susceptibilidad mediante minigenes híbridos.. Consejería de Educación. Junta de Castilla y León. Ref.: CSI242P18. (IP) Eladio A. Velasco Sampedro. (Instituto de Biología y Genética Molecular (CSIC)). 01/01/2019-31/12/2021. 120.000 €. Principal investigator.

5 **Project**. Aberrant splicing in hereditary breast cancer: Functional analysis of susceptibility genes by hybrid minigenes. Instituto de Salud Carlos III. Expdte. PI17/00227. (IP) Eladio A. Velasco Sampedro. (Instituto de Biología y Genética Molecular). 01/01/2018-31/12/2020. 99.220 €. Principal investigator.

6 **Project**. Breast Cancer Risk after Diagnostic Gene Sequencing (BRIDGES). Participant no. 18. European Commission/ Project ID 634935. (IP) Eladio Andrés Velasco Sampedro. (Instituto De Biología Y Genética Molecular-CSIC). 01/09/2015-31/08/2020. 105.559,96 €. Principal investigator.

7 **Project**. Impacto de las mutaciones reguladoras de transcripción y splicing de genes supresores de tumores en el diagnóstico molecular y prevención de cáncer de mama. Consejería de Educación. Junta de Castilla y León. Ref. CSI090U14. (IP) Eladio Velasco Sampedro. (Instituto de Biología y Genética Molecular - CSIC). 01/01/2015-31/12/2016. 28.980 €. Principal investigator.

8 **Project**. Alteraciones de la transcripción y splicing de los genes BRCA1 y BRCA2 y susceptibilidad genética a cáncer de mama y ovario. Instituto de Salud Carlos III/ Expdte: PI13/01749. (IP) Eladio A. Velasco Sampedro. (Instituto de Biología y Genética Molecular). 2014-2016. 74.838,5 €. Principal investigator.

9 **Project**. Análisis de secuencias reguladoras de splicing (enhancer y silenciadores) de BRCA1 y BRCA2 mediante minigenes híbridos: splicing aberrante y cáncer de mama/ovario. Análisis global de los patrones de splicing en pacientes BRCA negativos.". Instituto de Salud Carlos III/ Expdte: 10/02910. (IP) Eladio A. Velasco Sampedro. 2011-2013. 121.000 €. Principal investigator.

10 **Project**. Alteraciones de la expresión de genes supresores de tumores en cáncer de mama y ovario hereditario. Implicaciones en el consejo genético y la prevención de esta enfermedad. Junta de Castilla y León. Consejería de Sanidad.Expdte:BIO/VA08/13. (IP) Eladio A. Velasco Sampedro. (Instituto de Biología y Genética Molecular). From 2013. 12.500 €. Principal investigator.

11 **Project**. Alteraciones del procesamiento del ARNm de los genes supresores de tumores BRCA1 Y BRCA2 en pacientes con cáncer de mama/ovario de CASTILLA Y LEÓN. Correlación con la susceptibilidad genética a esta enfermedad". Junta de Castilla y León. Consejería de Educación. Ref. CSI004A10-2. (IP) Eladio A. Velasco Sampedro. (Instituto de Biología y Genética Molecular). From 2010. 32.850 €. Principal investigator.

12 **Project**. Correlación entre anomalías del procesamiento de ARNm (splicing) de los genes BRCA1 y BRCA2 y la predisposición hereditaria a cáncer de mama y ovario. Mapeo de secuencias reguladoras de splicing mediante minigenes híbridos.". Junta de Castilla y León. Consejería de Sanidad.Expdte: BIO39/VA27/10. (IP) Eladio A. Velasco Sampedro. (Instituto de Biología y Genética Molecular). From 2010. 29.608 €. Principal investigator.

13 **Project**. Splicing anómalo de los genes BRCA1 y BRCA2 y predisposición genética a cáncer de mama y/o ovario". CSIC-Proyectos Intramurales. Ref. 200820I135. (IP) Eladio A. Velasco Sampedro. (Instituto de Biología y Genética Molecular). From 2008. 30.000 €. Principal investigator.

14 **Project**. Influencia de las variantes de efecto fisiológico desconocido de los genes BRCA1 y BRCA2 en las alteraciones del procesamiento del ARNm. Correlación entre eliminación de elementos clave reguladores del splicing y susceptibilidad genética a cáncer de mama". Instituto de Salud Carlos III/ Expdte: 06/1102. (IP) Eladio A. Velasco Sampedro. (Instituto de Biología y Genética Molecular). From 2007. 60.600 €. Principal investigator.

15 **Healthcare project**. PROGRAMA DE PREVENCIÓN DE METABOLOPATÍAS (1999). INVESTIGADOR CONTRATADO. CONSEJERÍA DE SANIDAD. JUNTA DE CASTILLA Y LEÓN. (INSTITUTO DE BIOLOGÍA Y GENÉTICA MOLECULAR. UNIVERSIDAD DE VALLADOLID). 1999-1999. €.

16 **Healthcare project**. PROGRAMA DE PREVENCIÓN DE CÁNCER HEREDITARIO (2000-2008). INVESTIGADOR CONTRATADO. CONSEJERÍA DE SANIDAD. JUNTA DE CASTILLA Y LEÓN. (INSTITUTO DE BIOLOGÍA Y GENÉTICA MOLECULAR. UNIVERSIDAD DE VALLADOLID). €.

17 **Contract**. Basic functional assays (wild type and mutant minigenes) of the BRCA1 gene (9 variants) INSTITUTO DE INVESTIGACIÓN BIOMÉDICA DE BELLVITGE. (IP) Eladio A. Velasco Sampedro. 21/03/2024-21/03/2025. 2.722,5 €.

18 **Contract**. Basic functional assays (wild type and mutant minigenes) of the BRCA1 gene (9 variants) FUNDACIÓ HOSPITAL UNIVERSITARI VALL D'HEBRON INSTITUT DE RECERCA. (IP) Eladio A. Velasco Sampedro. 13/03/2024-13/03/2025. 2.722,5 €.

19 **Contract**. Splicing analysis of the TP53 gene by hybrid minigenes Queensland Institute of Medical Research (QIMR Berghofer). (IP) Eladio A. Velasco Sampedro. 01/01/2022-01/07/2023. 60.000 €.

20 **Contract**. SPLICING FUNCTIONAL STUDY OF DNA VARIANTS IN PSAD-DERIVED MINIGENES11 NATIONAL HUMAN GENOME RESEARCH INSTITUTE (NHGRI). (IP) Eladio A. Velasco Sampedro. 11/11/2021-11/02/2022. 750 €.

21 **Contract**. Construction of an ad hoc minigene of the TRPM4 gene in the pSAD vector. Splicing functional assay of TRPM4 variant c.25-1G>T for Dr. Hemminki and Dr. Bandapalli German Cancer Research Center (DKFZ). (IP) Eladio A. Velasco Sampedro. 09/04/2019-09/07/2019. 600 €.

22 **Contract**. SPLICING FUNCTIONAL STUDY OF DNA VARIANTS IN PSAD-DERIVED MINIGENES. BASIC FUNCTIONAL ASSAYS (WILD TYPE AND MUTANT MINIGENES). INSERTION OF EXÓN I Empresa SIA BioAVots y Riga Stradins University (Riga, Letonia). (IP) Eladio A. Velasco Sampedro. 18/03/2019-18/06/2019. 400 €.

23 **Contract**. Splicing Functional Assay of variant c.996+2_996+5del of the UGT1A1 gene (Crigler-Najjar Syndrome) and construction of a custom minigene for Dr. Linda Gaïlite Empresa SIA BioAVots y Riga Stradins University (Riga, Letonia).. (IP) Eladio A. Velasco Sampedro. 30/10/2018-30/01/2019. 600 €.

24 **Contract**. Splicing Functional Assay of variant IVS7-1G>A of the GRN gene (Frontotemporal Dementia) and custom minigene construction for Dr. Ana Belén de la Hoz Rastrollo ASOCIACIÓN INSTITUTO DE INVESTIGACIÓN SANITARIA BIOCRUCES. HOSPITAL UNIVERSITARIO CRUCES, Barakaldo, Bizkaia.. (IP) Eladio A. Velasco Sampedro. From 12/03/2018. 665,5 €.

25 **Contract**. Splicing Functional Assay of variant c.5665+1G>T of the CHD7 gene (Intellectual disability) and custom minigene construction for Dr. María Isabel Tejada Mínguez ASOCIACIÓN INSTITUTO DE INVESTIGACIÓN SANITARIA BIOCRUCES. HOSPITAL UNIVERSITARIO CRUCES, Barakaldo, Bizkaia. (IP) Eladio A. Velasco Sampedro. From 29/06/2016. 605 €.

C.4. Activities of technology / knowledge transfer and results exploitation

Eladio Andrés Velasco Sampedro; Alberto Acedo Bécares; Beatriz Díez Gómez. P201231427. Plásmido pSAD para ensayos funcionales de splicing Spain. 04/08/2015. Consejo Superior de Investigaciones Científicas.