CURRÍCULUM VÍTAE NORMALIZADO

Eladio Andrés Velasco Sampedro

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GOBIERNO DE ESPAÑA
MINISTERIO DE ECONOMÍA, INDUSTRIA Y COMPETITIVIDAD

FECYT FUNDACIÓN ESPAÑOLA PARA LA CIENCIA Y LA TECNOLOGÍA

Summary of CV

This section describes briefly a summary of your career in science, academic and research; the main scientific and technological achievements and goals in your line of research in the medium -and long- term. It also includes other important aspects or peculiarities.

Dr. Eladio A. Velasco (EAV) has focused his research interest on the molecular genetics of human inherited disorders, working in a wide range of them such as Childhood Spinal Muscular Atrophies (PhD thesis, 1996), Neurofibromatosis type I, Inherited Ataxias, Darier´s disease and, since 2000, in hereditary breast and ovarian cancer. He has participated in different aspects of the molecular characterization of the responsible genes. His current line of research studies the correlation between aberrant splicing of tumour suppressor genes and genetic susceptibility to cancer. He has developed a new splicing reporter plasmid (pSAD; patent #P201231427 CSIC) that has constituted the backbone of hybrid minigenes of several human disease genes, including BRCA1 and BRCA2 (Hereditary Breast and Ovarian Cancer, MLH1 (Lynch syndrome), COL1A1 (Osteogenesis Imperfecta) and SERPIN1A (Alpha-1-antitrypsin deficiency). By means of this approach, he has constructed the largest minigenes ever reported and tested a large number of DNA variants (~300) of the BRCA1/2 genes. EAV has shown the high frequency of splicing mutations in breast cancer genes, representing a primary disease mechanism. These findings are contributing to clarify the genetic susceptibility spectrum of breast/ovarian cancer and the specific participation of BRCA1/BRCA2 splicing mutations to it. Under the translational viewpoint, he has also provided a functional classification of a huge number of DNA variants reported worldwide in breast/ovarian cancer patients that constitute a valuable catalogue for medical geneticists of Genetic Counselling departments with a view to preventing this group of diseases. By virtue of his expertise in hybrid minigenes, EAV was invited to participate in the H2020 BRIDGES proposal, being participant no. 18 of such grant.

EAV is also involved in the study of the impact of promoter germline mutations of BRCA genes on transcription and their involvement in breast/ovarian carcinogenesis. Promoter regions of cancer-susceptibility genes are under-investigated as they are not routinely screened in genetic testing laboratories. EAV has found that BRCA2 transcription is highly sensitive to promoter variants causing either over or under expression.

Next years EAV plans to extend the minigene approach to other breast cancer susceptibility genes, such as CHEK2, PALB2, ATM, among others, within the H2020 BRIDGES proposal, as well as to study the specific splicing regulation of the non-canonical “GC” donor sites and special huge exons (BRCA1 and BRCA2 exons 11, PALB2, exons 4 and 5) and their roles in health and cancer.

The scientific career of Eladio Velasco is supported by publications in high impact journals of categories Genetics and Heredity, Oncology, Medical Laboratory Technology or Biochemical Research Methods, mainly of the first quartile or decile, such as: Nature Protocols, Clinical Cancer Research, PLoS Genetics or Human Molecular Genetics. He has been the principal investigator of 8 national and 1 European projects and has participated in at least other 12 grants as a collaborator. Finally he has been awarded with several prizes including: “Prize 3M a la innovación en el área de Salud” (2008), “Prize Junta de Castilla y León” de la Real Academia de Medicina y Cirugía de Valladolid (2009) and the Extraordinary Doctorate Award (Universidad Autónoma de Madrid).

General quality indicators of scientific research

This section describes briefly the main quality indicators of scientific production (periods of research activity, experience in supervising doctoral theses, total citations, articles in journals of the first quartile, H index...). It also includes other important aspects or peculiarities.

- 48 publications in scientific journals:
    - 32 articles in Q1 journals (10 of them in the first decile)
    - Average Impact Factor: ~4.5
    - Top 5 cited articles (Scopus): Hum Mol Genet 1996, 5:257-263 (165 citations), Clin Cancer Res 2010, 16: 1957-1967 (47 citations), Hum Mutat 2004, 22: 301-312 (129 citations), Clin Cancer Res 2008, 14: 2861-2869 (60 citations), Clin Chem 2006, 52: 1480-1485 (50 citations).
- h-index (Scopus)= 16
- ~90 communications at national and international Conferences.
- Supervision of 6 PhD Thesis, four of them awarded with the Doctoral Extraordinary Prize of the University of Valladolid.
- One Spanish patent (P201231427): “pSAD plasmid for splicing functional assays”
- Participation in 22 grants, nine of them as principal investigator, including the European H2020 grant “Breast Cancer After Diagnostic Gene Sequencing” (BRIDGES) [see below]
- Principal investigator or scientific coordinator of 7 contracts or agreements with private companies or public organisms.
- Member of the Committee of Familial Breast Cancer of the Spanish Association for Human Genetics (2008-2013)
. Member of the Scientific Advisory Board of the biotechnological company AC GEN Reading Life.
- Inventor of the Spanish patent P201231427 “pSAD plasmid for splicing functional assays”.
- Seven awards, three of which should be highlighted: “Prize 3M a la innovación en el área de Salud” (2008), el “Prize Junta de Castilla y León” de la Real Academia de Medicina y Cirugía de Valladolid (2009) and the Extraordinary Doctorate Award (Universidad Autónoma de Madrid).

Eladio Andrés Velasco Sampedro

Surname(s):	**Velasco Sampedro**
Name:	**Eladio Andrés**
DNI:	**50163233A**
ORCID:	**orcid.org/0000-0002-9682-5589**
ResearcherID:	**D-1432-2016**
Date of birth:	**20/04/1965**
Gender:	**Male**
Nationality:	**Spain**
Country of birth:	**Spain**
Aut. region/reg. of birth:	**Castile and León**
City of birth:	**Riaza**
Contact address:	**c/ Sanz y Forés 3**
Postcode:	**47003**
Contact country:	**Spain**
Contact city:	**Valladolid**
Land line phone:	**983184829**
Fax:	**983184800**
Email:	**eavelsam@ibgm.uva.es**
Mobile phone:	**600844194**

Current professional situation

Employing entity: Consejo Superior de Investigaciones Científicas
Department: Servicio de Genómica, Instituto de Biología y Genética Molecular
Professional category: Coordinador. Investigador Responsable
Start date: 01/01/2011
Primary (UNESCO code): 240900 - Genetics

Employing entity: Consejo Superior de Investigaciones Científicas **Type of entity:** Public Research Body
Department: Splicing y cáncer, Instituto de Biología y Genética Molecular
Professional category: Científico Titular
Phone: (34) 983184829 **Fax:** (34) 983184800 **Email:** eavelsam@ibgm.uva.es
Start date: 21/07/2008
Type of contract: Civil servant **Dedication regime:** Full time
Primary (UNESCO code): 240902 - Genetic engineering; 241007 - Human genetics; 241500 - Molecular biology; 320713 - Oncology
Secondary (UNESCO code): 320101 - Cancerology
Performed tasks: -Estudio de la correlación entre splicing anómalo de genes supresores de tumores y susceptibilidad genética a cáncer de mama/ovario. -Ensayos funcionales de splicing mediante minigenes híbridos construidos en plásmidos reporteros de splicing. -Genética molecular del cáncer de mama/ovario hereditario.
Identify key words: Molecular biology; Medical genetics; Molecular genetics; Genomics

Previous positions and activities

	Employing entity	Professional category	Start date
1	Universidad de Valladolid	Investigador contratado	01/07/1999
2	Hospital General de Segovia	Investigador posdoctoral	01/01/1997
3	Hospital General de Segovia	Becario FIS 96/0337	01/01/1996
4	Hospital Universitario Ramón y Cajal	Becario Proyecto FIS 93/0027-01	01/01/1993
5	Hospital Universitario Ramón y Cajal	Becario predoctoral FPI	01/01/1989

1 **Employing entity:** Universidad de Valladolid **Type of entity:** University
Department: Instituto de Biología y Genética Molecular
Professional category: Investigador contratado
Start date: 01/07/1999 **Duration:** 9 years - 20 days
Type of contract: Temporary employment contract
Dedication regime: Full time
Primary (UNESCO code): 240900 - Genetics; 241007 - Human genetics; 241500 - Molecular biology
Performed tasks: -Plan de prevención de enfermedades congénitas (6 meses) -Plan de prevención de cáncer hereditario de la junta de Castilla y León (8 años, 6 meses, 20 días)

2 **Employing entity:** Hospital General de Segovia **Type of entity:** Healthcare Institutions
Department: Unidad de Investigación
Professional category: Investigador posdoctoral
Start date: 01/01/1997 **Duration:** 2 years - 6 months
Type of contract: Grant-assisted student (pre or post-doctoral, others)
Dedication regime: Full time
Primary (UNESCO code): 240900 - Genetics; 241007 - Human genetics; 241500 - Molecular biology
Performed tasks: -Epidemiología Genética de enfermedades neurodegenerativas hereditarias - Estudio de enfermedades dermatológicas hereditarias: enfermedad de Darier

3 **Employing entity:** Hospital General de Segovia **Type of entity:** Healthcare Institutions
Professional category: Becario FIS 96/0337
Start date: 01/01/1996 **Duration:** 1 year
Type of contract: Grant-assisted student (pre or post-doctoral, others)
Dedication regime: Full time
Primary (UNESCO code): 241007 - Human genetics

4 **Employing entity:** Hospital Universitario Ramón y Cajal **Type of entity:** Healthcare Institutions
Department: Unidad de Genética Molecular
Professional category: Becario Proyecto FIS 93/0027-01
Start date: 01/01/1993 **Duration:** 3 years
Type of contract: Grant-assisted student (pre or post-doctoral, others)
Dedication regime: Full time
Primary (UNESCO code): 240900 - Genetics; 241007 - Human genetics; 241500 - Molecular biology
Performed tasks: Genética Molecular de las Atrofias Musculares Espinales

5 **Employing entity:** Hospital Universitario Ramón y Cajal **Type of entity:** Healthcare Institutions
Department: Unidad de Genética Molecular
City employing entity: Madrid, Community of Madrid, Spain
Professional category: Becario predoctoral FPI

Start date: 01/01/1989 **Duration:** 4 years
Type of contract: Grant-assisted student (pre or post-doctoral, others)
Dedication regime: Full time
Primary (UNESCO code): 240900 - Genetics; 241007 - Human genetics; 241500 - Molecular biology
Performed tasks: -Aislamiento y caracterización de minisatélites de ADN humano emparentados con el minisatélite 113I

Education

University education

1st and 2nd cycle studies and pre-Bologna degrees

University degree: Higher degree
Name of qualification: Licenciado en Ciencias Biológicas Especialidad Biología Fundamental
Degree awarding entity: Universidad Complutense de Madrid **Type of entity:** University
Date of qualification: 1988

Doctorates

Doctorate programme: Programa Oficial de Doctorado en Bioquímica y Biología Molecular
Degree awarding entity: Universidad Autónoma de Madrid **Type of entity:** University
Date of degree: 24/10/1996
Thesis title: GENÉTICA MOLECULAR DE LAS ATROFIAS MUSCULARES ESPINALES
Thesis director: Concepción Hernández Chico
Obtained qualification: Apto cum laude. Premio Extraordinario de Doctorado

Language skills

Language	Listening skills	Reading skills	Spoken interaction	Speaking skills	Writing skills
English		C1	B1	B1	C1

Teaching experience

General teaching experience

1 **Name of the course:** Aplicaciones de la Biología Molecular en Biomedicina
Type of programme: Master's degree
University degree: MÁSTER EN INVESTIGACIÓN BIOMÉDICA
Frequency of the activity: 3
End date: 2011 **Type of hours/ ECTS credits:** Credits
Hours/ECTS credits: 3
Entity: Universidad de Valladolid

2 **Type of teaching:** Official teaching
Name of the course: Enfermedades Genéticas: Fisiopatología Molecular
Type of programme: Doctorate

University degree: Programa de doctorado BIOTECNOLOGÍA: APLICACIONES BIOMÉDICAS"
Frequency of the activity: 5
End date: 2009 **Type of hours/ ECTS credits:** Credits
Hours/ECTS credits: 3
Entity: Universidad Valladolid

3 **Name of the course:** Técnicas avanzadas en Biotecnología: SECUENCIACION AUTOMATICA DE ADN. ANALISIS DE FRAGMENTOS Y METODOS PARA DETECCION DE MUTACIONES"
Type of programme: Doctorate
University degree: Programa de doctorado BIOTECNOLOGÍA: APLICACIONES BIOMÉDICAS"
Frequency of the activity: 5
End date: 2009 **Type of hours/ ECTS credits:** Credits
Hours/ECTS credits: 5
Entity: UNIVERSIDAD DE VALLADOLID

4 **Type of teaching:** International teaching
Name of the course: 18th, 19th and 21st Course in Medical Genetics
Type of subject: Doctorate
University degree: Programa de doctorado "BIOTECNOLOGÍA: APLICACIONES BIOMÉDICAS"
Frequency of the activity: 3
End date: 2008
Entity: Escuela Europea de Genética Médica-Universidad de Valladolid
City of entity: Valladolid,

5 **Name of the course:** Genética Molecular e Ingeniería Genética
University degree: Especialista en Ingeniería Biomédica
End date: 2003
Entity: Universidad de Valladolid **Type of entity:** University

6 **Name of the course:** "Orientaciones para la enseñanza de la Biología: Biología Molecular e Ingeniería Genética"
Type of programme: Formación profesorado Enseñanza Secundaria
University degree: Instituto de Formación del Profesorado
End date: 2002
Entity: Universidad de Burgos **Type of entity:** University

7 **Name of the course:** Técnicas Básicas de Biología Molecular"
Type of programme: Doctorate
University degree: Programa de Doctorado de Ciencias de la Visión del Instituto de Oftalmobiología Aplicada (IOBA)
End date: 2000 **Type of hours/ ECTS credits:** Credits
Hours/ECTS credits: 1
Entity: Instituto de Oftalmobiología Aplicada **Type of entity:** University Research Institute

Experience supervising doctoral thesis and/or final year projects

1 **Project title:** SPLICING FUNCTIONAL ANALYSIS OF DNA VARIANTS WITHIN THE BREAST CANCER TYPE 2 SUSCEPTIBILITY GENE (BRCA2), AND ITS EFFECT ON HEREDITARY BREAST AND OVARIAN CANCER (HBOC): A HYBRID MINIGENE APPROACH.
Type of project: Trabajo Fin de Máster
Entity: Universidad de Valladolid
Student: Valeria Velásquez Zapata
Obtained qualification: 10
Date of reading: 15/07/2015

2 **Project title:** Alteraciones de splicing del gen BRCA2 en cáncer de mama y ovario. Diseño del nuevo vector de splicing pSAD para la construcción de minigenes híbridos
Type of project: Doctoral thesis
Entity: Universidad de Valladolid
Student: Alberto Acedo Bécares
Date of reading: 02/12/2013

3 **Project title:** Alteraciones de splicing en genes supresores de tumores como mecanismo etiopatológico en cáncer de mama y ovario hereditario: análisis del exón 17 de BRCA1
Type of project: Trabajo Fin de Máster
Entity: Valladolid
Student: Alvaro Curiel García
Obtained qualification: 9,3
Date of reading: 15/07/2013

4 **Project title:** ESTUDIO DEL SPLICING ABERRANTE EN EL GEN SUPRESOR DE TUMORES BRCA1 MEDIANTE MINIGENES HÍBRIDOS Y SU IMPACTO EN LA PREDISPOSICIÓN GENÉTICA A CÁNCER DE MAMA Y OVARIO
Type of project: Trabajo fin de máster
Entity: Valladolid
Student: Cristina Hernández Moro
Obtained qualification: 9,3
Date of reading: 14/09/2012

5 **Project title:** Estudio genético del síndrome de Lynch. Caracterización de alteraciones recurrentes y de variantes de efecto desconocido
Type of project: Doctoral thesis
Co-director of thesis: Mercedes Durán Domínguez
Entity: Universidad de Valladolid
Student: LUCÍA PÉREZ CABORNERO
Obtained qualification: Sobresaliente cum laude. PREMIO EXTRAORDINARIO DE DOCTORADO
Date of reading: 22/07/2011

6 **Project title:** Splicing anómalo de los genes BRCA1 y BRCA2 y susceptibilidad genética a cáncer de mama y ovario
Type of project: Doctoral thesis
Co-director of thesis: Mercedes Durán Domínguez
Entity: Universidad de Valladolid
City of entity: Valladolid, Castile and León, Spain

Student: David José Sanz San José
Obtained qualification: Sobresaliente cum laude
Date of reading: 18/09/2009

7 **Project title:** CANCER DE MAMA Y OVARIO HEREDITARIOS. ESTUDIO MOLECULAR DE LOS GENES DE PREDISPOSICIÓN BRCA1 Y BRCA2 Y EFECTOS FUNDADORES DE LAS MUTACIONES MÁS FRECUENTES EN CASTILLA Y LEÓN.
Type of project: Doctoral thesis
Co-director of thesis: Mercedes Durán Domínguez
Entity: Universidad de Valladolid
Student: Mª MAR INFANTE SANZ
Obtained qualification: Sobresaliente cum laude. Premio Extraordinario de Doctorado Universidad de Valladolid
Date of reading: 31/03/2008

8 **Project title:** "Análisis funcional de elementos reguladores de splicing en los exones 19 y 20 de BRCA2"
Type of project: Work leading to an ASD
Entity: Universidad de Valladolid **Type of entity:** University
Student: Alberto Acedo Bécares
Obtained qualification: Sobresaliente
Date of reading: 2010
Date of recognition: 03/10/2007
Quality recognition: Yes

9 **Project title:** Estudio Genético y Molecular de los genes de reparación de ADN (MMR) en pacientes con síndrome de Lynch”
Type of project: Work leading to an ASD
Entity: Universidad de Valladolid
Student: Lucía Pérez Cabornero
Obtained qualification: NOTABLE (8,7)
Date of reading: 2007

10 **Project title:** ANÁLISIS MOLECULAR DE LOS GENES BRCA1 Y BRCA2 EN PACIENTES CON CÁNCER DE MAMA Y OVARIO. DESARROLLO DE UN NUEVO MÉTODO DE DETECCIÓN DE MUTACIONES.
Type of project: Doctoral thesis
Co-director of thesis: Cristina Miner Pino
Entity: Universidad de Valladolid
Student: EVA MARÍA ESTEBAN CARDEÑOSA
Obtained qualification: Sobresaliente cum laude. Premio Extraordinario de Doctorado Universidad de Valladolid
Date of reading: 24/06/2005

11 **Project title:** Aplicación de ayuda para la búsqueda de mutaciones en genes BRCA1 y BRCA2 - II”
Type of project: End of course project
Co-director of thesis: Javier Pérez Turiel
Entity: Universidad de Valladolid-E.T.S. DE INGENIERÍA INFORMÁTICA **Type of entity:** University
Student: David San León Granado
Date of reading: 2005

12 **Project title:** Aplicación de ayuda para la búsqueda de mutaciones en genes BRCA1 y BRCA2 - I”
Type of project: End of course project
Co-director of thesis: Javier Pérez Turiel

Entity: Universidad de Valladolid-E.T.S. DE INGENIERÍA INFORMÁTICA **Type of entity:** University
Student: Alfonso Almohalla González
Date of reading: 2005

13 **Project title:** Aplicación para la detección de mutaciones en genes, susceptibles de provocar enfermedades hereditarias"-II
Type of project: End of course project
Co-director of thesis: Javier Pérez Turiel
Entity: Universidad de Valladolid-E.T.S. DE INGENIERÍA INFORMÁTICA **Type of entity:** University
Student: Francisco Javier González Zorita
Date of reading: 2005

14 **Project title:** Aplicación para la detección de mutaciones en genes, susceptibles de provocar enfermedades hereditarias"
Type of project: End of course project
Co-director of thesis: Javier Pérez Turiel
Entity: Universidad de Valladolid-E.T.S. DE INGENIERÍA INFORMÁTICA
Student: Jesús María Recio Rincón
Date of reading: 2003

15 **Project title:** ESTUDIO CLÍNICO Y GENÉTICO DE FAMILIAS ESPAÑOLAS AFECTADAS DE LA ENFERMEDAD DE DARIER-WHITE
Type of project: Doctoral thesis
Co-director of thesis: Susana Gómez Barrero
Entity: Universidad de Valladolid
Student: MARÍA FERNANDA RODRÍGUEZ SANZ
Obtained qualification: SOBRESALIENTE CUM LAUDE
Date of reading: 08/03/2002

16 **Project title:** Investigación en enfermedades poligénicas – Problemática de la Genética del Asma"
Type of project: Work leading to an ASD
Entity: Universidad de Valladolid **Type of entity:** University
Student: Ignacio Díez López
Obtained qualification: Sobresaliente
Date of reading: 2002

Other activities/achievements not included above

1 **Description of the activity:** Presidente del Tribunal de Tesis de María Alejandra Bernardi "Métodos de Modificación génica dirigida en células humanas y su aplicación en el síndrome de Wiskott-Aldrich"
Organising entity: Universidad de Valladolid
End date: 17/04/2015

2 **Description of the activity:** Examiner of a research higher degree thesis of student Brooke Brewster: "The Role of the BRCA1 3'UTR in Breast Cancer"
Organising entity: University of Queensland, Australia
End date: 22/04/2014

3 **Description of the activity:** Evaluador externo de la Tesis "SÍNDROMES HEREDITARIOS DE CÁNCER DE MAMA FAMILIAR: VARIANTES DE SIGNIFICADO CLÍNICO INCIERTO Y CONSEJO GENÉTICO" del doctorando Gorka Ruiz de Garibay Ponce
Organising entity: Universidad Complutense de Madrid
End date: 31/01/2014

4 **Description of the activity:** Tutor de la Beca de Investigación Villalar 2014, Cristina Hernández Moro: "Aplicación de nuevas tecnologías de diagnóstico molecular mediante minigenes híbridos para la prevención de enfermedades hereditarias de gran impacto clínico en Castilla y León"
Organising entity: Fundación Villalar (Castilla y León)
End date: 2014

5 **Description of the activity:** Tutor de la Beca de Investigación Villalar 2013, Álvaro Curiel García: "Alteraciones de splicing en genes supresores de tumores como mecanismo etiopatológico en cáncer de mama y ovario hereditario en pacientes de Castilla y León"
Organising entity: Fundación Villalar (Castilla y León)
End date: 2013

6 **Description of the activity:** Miembro del Tribunal de Tesis de Francisco Martínez Redondo "Caracterización del procesamiento proteolítico de miocilina: papel de las calpaínas, influencia sobre la regulación de genes de matriz extracelular e implicaciones en glaucoma."
Organising entity: Universidad de Castilla-La Mancha
End date: 26/07/2012

7 **Description of the activity:** Miembro del Tribunal de Tesis de Laura Barrio Real "Nuevas aportaciones a la caracterización molecular del gen CHN2"
Organising entity: Universidad de Salamanca **Type of entity:** University
End date: 04/03/2011

8 **Description of the activity:** Secretario del Tribunal de Tesis de Blanca López Posadas
Organising entity: Universidad de Valladolid
End date: 04/06/2007

Healthcare experience

Healthcare innovation projects

1 **Name of the project:** PROGRAMA DE PREVENCIÓN DE CÁNCER HEREDITARIO. INVESTIGADOR CONTRATADO
Type of project: Public health
Entity where project took place: INSTITUTO DE BIOLOGÍA Y GENÉTICA MOLECULAR. UNIVERSIDAD DE VALLADOLID
Funding entity: CONSEJERÍA DE SANIDAD. JUNTA DE CASTILLA Y LEÓN
Start date: 01/01/2000 **Duration:** 8 years - 6 months - 20 days

2 **Name of the project:** PROGRAMA DE PREVENCIÓN DE METABOLOPATÍAS. INVESTIGADOR CONTRATADO
Type of project: Public health
Entity where project took place: INSTITUTO DE BIOLOGÍA Y GENÉTICA MOLECULAR. UNIVERSIDAD DE VALLADOLID
Funding entity: CONSEJERÍA DE SANIDAD. JUNTA DE CASTILLA Y LEÓN

Start date: 01/07/1999 **Duration:** 6 months

Scientific and technological experience

Scientific or technological activities

R&D projects funded through competitive calls of public or private entities

1 **Name of the project:** Splicing aberrante en cáncer de mama hereditario: Análisis funcional de genes de susceptibilidad mediante minigenes híbridos // Aberrant splicing in hereditary breast cancer: Functional analysis of susceptibility genes by hybrid minigenes
Entity where project took place: Instituto de Biología y Genética Molecular **Type of entity:** University Research Institute
Name principal investigator (PI, Co-PI....): Eladio A. Velasco
Nº of researchers: 6
Funding entity or bodies:
Instituto de Salud Carlos III **Type of entity:** Public Research Body
City funding entity: Majadahonda, Community of Madrid, Spain

Start-End date: 01/01/2018 - 31/12/2020
Total amount: 99.220 €

2 **Name of the project:** Breast Cancer Risk after Diagnostic Gene Sequencing (BRIDGES). Participant no. 18
Entity where project took place: Instituto De Biología Y Genética Molecular-CSIC
Name principal investigator (PI, Co-PI....): (IP) Eladio Andrés Velasco
Nº of researchers: 1
Funding entity or bodies:
European Commission/ Project ID 634935

Start-End date: 01/01/2016 - 31/12/2019
Total amount: 75.000 €

3 **Name of the 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
Entity where project took place: Instituto de Biología y Genética Molecular - CSIC **Type of entity:** Public Research Body
Name principal investigator (PI, Co-PI....): (IP) Eladio Velasco Sampedro
Nº of researchers: 6
Funding entity or bodies:
Consejería de Educación. Junta de Castilla y León. Ref. CSI090U14

Start-End date: 01/01/2015 - 31/12/2016
Total amount: 28.980 €

4 **Name of the project:** Alteraciones de la transcripción y splicing de los genes BRCA1 y BRCA2 y susceptibilidad genética a cáncer de mama y ovario
Entity where project took place: Instituto de Biología y Genética Molecular
Name principal investigator (PI, Co-PI....): (IP) Eladio A. Velasco
Nº of researchers: 6
Funding entity or bodies:

Instituto de Salud Carlos III/ Expdte: PI13/01749 **Type of entity:** State agency

Start-End date: 2014 - 2016 **Duration:** 3 years
Total amount: 74.838,5 €

5 **Name of the project:** "Metástasis a pulmón en el cáncer de mama:función de Beta2-quimerina e implicaciones terapéuticas"
Entity where project took place: Instituto de Biología y Genética Molecular (IBGM)
Name principal investigator (PI, Co-PI....): (IP) María José Caloca Roldán; Eladio A. Velasco Sampedro
Nº of researchers: 2
Funding entity or bodies:
Junta de Castilla y León. Consejería de Sanidad.Expdte:BIO/VA34/15

Start-End date: 2015 - 2015
Total amount: 9.889 €

6 **Name of the project:** Papel de las quimerinas en cáncer de mama: mecanismos moleculares, valor diagnóstico y evaluación como diana farmacológica
Entity where project took place: Instituto de Biología y Genética Molecular (IBGM)
Name principal investigator (PI, Co-PI....): (IP) María José Caloca Roldán; Eladio A. Velasco Sampedro
Nº of researchers: 3
Funding entity or bodies:
Junta de Castilla y León. Consejería de Sanidad.Expdte:BIO/VA22/14

Start-End date: 2014 - 2014
Total amount: 11.166 €

7 **Name of the 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."
Entity where project took place: Instituto de Biología y Genética Molecular **Type of entity:** State agency
Name principal investigator (PI, Co-PI....): (IP) Eladio A. Velasco
Nº of researchers: 7
Funding entity or bodies:
Instituto de Salud Carlos III/ Expdte: 10/02910

Start-End date: 2011 - 2013
Total amount: 121.000 €

8 **Name of the 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
Entity where project took place: Instituto de Biología y Genética Molecular
Name principal investigator (PI, Co-PI....): (IP) Eladio A. Velasco
Nº of researchers: 6
Funding entity or bodies:
Junta de Castilla y León. Consejería de Sanidad.Expdte:BIO/VA08/13

Start date: 2013 **Duration:** 1 year
Total amount: 12.500 €

9 **Name of the project:** IMPLICACIÓN DE LOS DEFECTOS EN EL PROOFREADING DE LAS POLIMERASAS EN EL ALTO RIESGO DE CÁNCER DE COLON EN FAMILIAS SIN MUTACIÓN MMR
Entity where project took place: Instituto de Biología y Genética Molecular (IBGM)

Name principal investigator (PI, Co-PI....): (IP) MERCEDES DURÁN DOMÍNGUEZ
Nº of researchers: 7
Funding entity or bodies:
Junta de Castilla y León. Consejería de Sanidad.Expdte:BIO/VA16/13

Start date: 2013 **Duration:** 1 year
Total amount: 16.960 €

10 **Name of the 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"
Entity where project took place: Instituto de Biología y Genética Molecular **Type of entity:** State agency
Name principal investigator (PI, Co-PI....): (IP) Eladio A. Velasco Sampedro
Funding entity or bodies:
Junta de Castilla y León. Consejería de Educación. Ref. CSI004A10-2

Start date: 2010 **Duration:** 2 years
Total amount: 32.850 €

11 **Name of the project:** CARACTERIZACIÓN Y CLASIFICACIÓN DE VARIANTES UV EN GENES IMPLICADOS EN SÍNDROME DE LYNCH"
Entity where project took place: Instituto de Biología y Genética Molecular **Type of entity:** State agency
Name principal investigator (PI, Co-PI....): Enrique Lastra Aras
Funding entity or bodies:
Caja de Burgos

Start date: 2010
Total amount: 4.000 €

12 **Name of the 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."
Entity where project took place: Instituto de Biología y Genética Molecular **Type of entity:** State agency
Name principal investigator (PI, Co-PI....): (IP) Eladio A. Velasco Sampedro
Funding entity or bodies:
Junta de Castilla y León. Consejería de Sanidad.Expdte: BIO39/VA27/10

Start date: 2010 **Duration:** 2 years
Total amount: 29.608 €

13 **Name of the project:** Splicing anómalo de los genes BRCA1 y BRCA2 y predisposición genética a cáncer de mama y/o ovario"
Entity where project took place: Instituto de Biología y Genética Molecular **Type of entity:** State agency
Name principal investigator (PI, Co-PI....): (IP) Eladio A. Velasco Sampedro
Funding entity or bodies:
CSIC-Proyectos Intramurales. Ref. 200820I135

Start date: 2008 **Duration:** 2 years
Total amount: 30.000 €

14 **Name of the project:** Ataxias en nuestro medio: Un estudio longitudinal en pacientes y evaluaciones del papel de proteínas neuroprotectoras"
Entity where project took place: Hospital Clínico Universitario Valladolid
Name principal investigator (PI, Co-PI....): Benedicta Catalán Bernardos
Funding entity or bodies:
Caja de Burgos

Start date: 2007 **Duration:** 1 year

15 **Name of the 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"
Entity where project took place: Instituto de Biología y Genética Molecular **Type of entity:** State agency
Name principal investigator (PI, Co-PI....): (IP) Eladio A. Velasco Sampedro
Funding entity or bodies:
Instituto de Salud Carlos III/ Expdte: 06/1102

Start date: 2007 **Duration:** 3 years
Total amount: 60.600 €

16 **Name of the project:** Incidencia de reorganizaciones genómicas de los genes MMR en la población de Castilla y León. Comparativa con población española"
Entity where project took place: Instituto de Biología y Genética Molecular **Type of entity:** State agency
Name principal investigator (PI, Co-PI....): Mercedes Durán Domínguez
Funding entity or bodies:
Junta de Castilla y León. Ref. VA018B06

Start date: 2006 **Duration:** 2 years
Total amount: 6.600 €

17 **Name of the project:** ESTUDIO DE BETA-TALASEMIAS EN LA POBLACIÓN DE CASTILLA Y LEON."
Entity where project took place: Instituto de Biología y Genética Molecular **Type of entity:** State agency
Name principal investigator (PI, Co-PI....): Mª Jesús Alonso Ramos
Funding entity or bodies:
Junta de Castilla y León. Ref.: VA088/04.

Start date: 2004 **Duration:** 1 year
Total amount: 4.840 €

18 **Name of the project:** Densidad mineral ósea y polimorfismos del enzima convertidor de la angiotensina (ECA), del cotransportador Na-Cl distal y del receptor estrogénico en la población general"
Entity where project took place: Hospital Río Hortega (Valladolid)
Name principal investigator (PI, Co-PI....): José Luis Pérez Castrillón
Funding entity or bodies:
Instituto de Salud Carlos III. Expdte.: 02/1690

Start date: 2003 **Duration:** 3 years
Total amount: 75,1 €

19 **Name of the project:** Influencia de los polimorfismos genéticos en las hemorragias gastrointestinales asociadas a fármacos: un estudio multicéntrico de casos y controles"
Entity where project took place: Universidad de Valladolid
Name principal investigator (PI, Co-PI....): Alfonso Carvajal García-Pando
Funding entity or bodies:
Instituto de Salud Carlos III. Expdte.02/1572

Start date: 2003 **Duration:** 3 years
Total amount: 49.680 €

20 **Name of the project:** Análisis molecular del gen CFTR en formas atípicas de fibrosis quística en la población de Castilla y León"
Entity where project took place: Instituto de Biología y Genética Molecular **Type of entity:** State agency
Name principal investigator (PI, Co-PI....): Mª Jesús Alonso Ramos
Funding entity or bodies:
Junta de Castilla y León. Ref.: VA083/01.

Start date: 2001 **Duration:** 3 years
Total amount: 9.222 €

21 **Name of the project:** Epidemiología Genética e Historia Natural de las Ataxias Degenerativas Hereditarias y Esporádicas, y condiciones afines" Expdte. 98/0188
Entity where project took place: Hospital General de Segovia **Type of entity:** Healthcare Institutions
Name principal investigator (PI, Co-PI....): Jacinto Duarte García-Luis
Funding entity or bodies:
Instituto de Salud Carlos III **Type of entity:** Public Research Body
City funding entity: Majadahonda, Community of Madrid, Spain

Start date: 1998 **Duration:** 3 years

22 **Name of the project:** Epidemiología Genética y prevalencia de las distonías en la provincia de Segovia y otras zonas geográficas de España, integrado en el estudio epidemiológico de las distonías en Europa (ESDE)" Expdte. 98/0591
Entity where project took place: Hospital General de Segovia **Type of entity:** Healthcare Institutions
Name principal investigator (PI, Co-PI....): Luis Erik Clavería Soria
Funding entity or bodies:
Instituto de Salud Carlos III **Type of entity:** Public Research Body
City funding entity: Majadahonda, Community of Madrid, Spain

Start date: 1998 **Duration:** 3 years

R&D non-competitive contracts, agreements or projects with public or private entities

1 **Name of the project:** Clasificación clínica de variantes de significado clínico desconocido en genes de predisposición a síndromes cancerosos hereditarios mediante ensayos funcionales de splicing en minigenes híbridos construidos en el vector de splicing pSAD
Degree of contribution: Scientific coordinator
Name principal investigator (PI, Co-PI....): (co-IP) Eladio Velasco Sampedro; Alberto Acedo
Nº of researchers: 2

Participating entity/entities: AC-GEN Reading Life, S.L.; Instituto de Biología y Genética Molecular (CSIC-UVa)
Funding entity or bodies:
Convenio colaboración con la empresa AC-GEN Reading Life

Start date: 30/04/2014 **Duration:** 3 years

2 **Name of the project:** Ensayo funcional de splicing de los exones 1B y 1C del gen de la SERPINA1
Degree of contribution: Scientific coordinator
Name principal investigator (PI, Co-PI....): (IP) Eladio A. Velasco Sampedro
Participating entity/entities: Instituto de Biología y Genética Molecular (CSIC-UVa); Instituto de Salud Carlos III
Funding entity or bodies:
Instituto de Salud Carlos III

Start date: 01/12/2013 **Duration:** 5 months
Total amount: 562,65 €

3 **Name of the project:** Ensayo funcional de splicing de la mutación c.117-1G>T del gen MLH1 y construcción de minigen ad hoc.
Degree of contribution: Scientific coordinator
Name principal investigator (PI, Co-PI....): (IP) Eladio Andrés Velasco Sampedro
Nº of researchers: 1
Funding entity or bodies:
LORGEN GP, S.L. **Type of entity:** Technological Centre
City funding entity: Granada

Start date: 01/08/2013
Total amount: 530 €

4 **Name of the project:** HC-GEN TEST: Análisis de 32 genes de cáncer hereditario mediante ultrasecuenciaciación en pacientes con cáncer de mama/ovario de alto riesgo
Degree of contribution: Scientific coordinator
Name principal investigator (PI, Co-PI....): (co-IP) Eladio Andrés Velasco Sampedro; Alberto Acedo Bécares; Beatriz Sobrino
Participating entity/entities: AC-GEN Reading Life SL; FUNDACION GALEGA DE MEDICINA XENOMICA; Instituto de Biología y Genética Molecular
Funding entity or bodies:
AC-GEN READING LIFE S.L. **Type of entity:** Business

Start date: 21/06/2013 **Duration:** 1 year

5 **Name of the project:** Estudio in vitro del splicing de BRCA2. Extracción de ARN de sangre. Ensayo de RT-PCR del gen BRCA2
Degree of contribution: Scientific coordinator
Name principal investigator (PI, Co-PI....): (IP) Eladio Andrés Velasco Sampedro
Nº of researchers: 1
Funding entity or bodies:
SERVICIO EXTREMEÑO DE SALUD **Type of entity:** Administrative Body of the National Health System
City funding entity: Cáceres

Start date: 01/05/2013

6 **Name of the project:** Reacciones de secuencia para Departamento de Microbiología
Degree of contribution: Scientific coordinator
Name principal investigator (PI, Co-PI....): (IP) Eladio A. Velasco Sampedro
Nº of researchers: 1
Funding entity or bodies:
FUND GRAL DE LA UNIV. DE VALLADOLID
Type of entity: University Centres and Structures and Associated Bodies
Start date: 01/01/2013

7 **Name of the project:** PROGRAMA DE PREVENCIÓN DE CÁNCER HEREDITARIO DE LA JUNTA DE CASTILLA Y LEÓN
Type of project: Research and development, including transfer
Geographical area: Regional
Degree of contribution: Researcher
Name principal investigator (PI, Co-PI....): Cristina Miner Pino
Participating entity/entities: Junta de Castilla y León; Universidad de Valladolid
Funding entity or bodies:
Consejería de Sanidad
Type of entity: Administrative Body of the National Health System
City funding entity: Valladolid, Castile and León, Spain
Start date: 01/01/2000
Duration: 8 years - 6 months - 20 days

8 **Name of the project:** PLAN DE PREVENCIÓN DE ENFERMEDADES CONGÉNITAS
Geographical area: Regional
Degree of contribution: Researcher
Entity where project took place: Universidad de Valladolid
Type of entity: University
Name principal investigator (PI, Co-PI....): Alfredo Blanco Quirós
Participating entity/entities: Junta de Castilla y León; Universidad de Valladolid
Funding entity or bodies:
Consejería de Sanidad
Type of entity: Administrative Body of the National Health System
City funding entity: Valladolid, Castile and León, Spain
Start date: 01/07/1999
Duration: 6 months

Results

Industrial and intellectual property

Title registered industrial property: Plásmido pSAD para ensayos funcionales de splicing
Inventors/authors/obtainers: Eladio Andrés Velasco Sampedro; Alberto Acedo Bécares; Beatriz Díez Gómez
Entity holder of rights: Consejo Superior de Investigaciones Científicas
Nº of application: P201231427
Country of inscription: Spain
Date of register: 14/09/2012
Conferral date: 04/08/2015

Scientific and technological activities

Scientific production

Publications, scientific and technical documents

1 Montalban G; Fraile-Bethencourt E; López-Perolio I; Pérez-Segura P; Infante M; Durán M; Alonso-Cerezo MC; López-Fernández A; Diez O; de la Hoya M; Velasco EA; Gutiérrez-Enríquez S. Characterization of spliceogenic variants located in regions linked to high levels of alternative splicing: BRCA2 c.7976+5G > T as a case study.Human mutation. 27/06/2018. ISSN 1059-7794

DOI: 10.1002/humu.23583

PMID: 29969168

Type of production: Scientific paper

Corresponding author: Yes

Impact source: ISI

Category: Science Edition - GENETICS & HEREDITY

Impact index in year of publication: 5.359

Journal in the top 25%: Yes

Position of publication: 24

No. of journals in the cat.: 167

2 Fraile-Bethencourt E; Valenzuela-Palomo A; Díez-Gómez B; Infante M; Durán M; Marcos G; Lastra E; Gómez-Barrero S; Velasco EA. Genetic dissection of the BRCA2 promoter and transcriptional impact of DNA variants. Breast Cancer Res Treat. 15/05/2018.

DOI: 10.1007/s10549-018-4826-7

Type of production: Scientific paper

Corresponding author: Yes

Impact source: ISI

Category: Science Edition - ONCOLOGY

Impact index in year of publication: 3.626

Position of publication: 82

No. of journals in the cat.: 217

3 Fraile-Bethencourt E; Valenzuela-Palomo A; Díez-Gómez B; Acedo A; Velasco EA. Identification of Eight Spliceogenic Variants in BRCA2 Exon 16 by Minigene Assays. Frontiers in Genetics. 9, pp. 188. 05/2018.

DOI: 10.3389/fgene.2018.00188

Type of production: Scientific paper

Corresponding author: Yes

Impact source: ISI

Category: Science Edition - GENETICS & HEREDITY

Impact index in year of publication: 4.151

Journal in the top 25%: Yes

Position of publication: 47

No. of journals in the cat.: 167

4 Villate O.; Ibarluzea N.; Fraile-Bethencourt E.; Valenzuela A; Velasco E.A.; Grozeva D.; Raymond F.L.; Botella M.P.; Tejada M.I.Functional analyses of a novel splice variant in the CHD7 gene, found by Next Generation Sequencing, confirm its pathogenicity in a Spanish patient and diagnose him with CHARGE syndrome. Frontiers in Genetics. 9, pp. 7. 08/01/2018.

DOI: 10.3389/fgene.2018.00007

Type of production: Scientific paper

Impact source: ISI

Category: Science Edition - GENETICS & HEREDITY

Impact index in year of publication: 4.151 (2017)

Position of publication: 47

No. of journals in the cat.: 162

5 Pérez-Alonso M; Briongos LS; Ruiz-Mambrilla M; Velasco EA; Linares L; Cuellar L; Olmos JM; De Luis D; Dueñas-Laita A; Pérez-Castrillón JL. The Effect of Genistein Supplementation on Vitamin D Levels and Bone Turnover Markers during the Summer in Healthy Postmenopausal Women: Role of Genotypes of Isoflavone Metabolism.Journal of Nutrigenetics and Nutrigenomics. 11/2017.

DOI: 10.1159/000484480

Type of production: Scientific paper

Format: Journal

Impact source: ISI

Category: Nutrition and Dietetics

Impact index in year of publication: 1.5

6 Fraile-Bethencourt E; Díez-Gómez B; Velásquez-Zapata V; Acedo A; Sanz DJ; Velasco EA. Functional classification of DNA variants by hybrid minigenes: Identification of 30 spliceogenic variants of BRCA2 exons 17 and 18. PLoS Genetics. 13, pp. e1006691 [doi. 10.1371/journal.pgen.1006691]. 14/03/2017.

Type of production: Scientific paper

Corresponding author: Yes

Impact source: ISI

Category: Science Edition - GENETICS & HEREDITY

Impact index in year of publication: 6.1 (IF 2016)

Position of publication: 16

No. of journals in the cat.: 167

7 Acedo A; Díez-Gómez B; Hernández-Moro C; Curiel-García A; Velasco EA. Functional classification of BRCA2 DNA variants by splicing assays in a large minigene with 9 exons. Human Mutation. 36, pp. 210 - 221. 2015.

DOI: 10.1002/humu.22725

Type of production: Scientific paper

Format: Journal

Corresponding author: Yes

Impact source: ISI

Category: Science Edition - GENETICS & HEREDITY

Impact index in year of publication: 5.089

Journal in the top 25%: Yes

Position of publication: 23

No. of journals in the cat.: 166

Source of citations: SCOPUS

Citations: 4

8 Lara B; Martínez MT; Blanco I; Hernández-Moro C; Velasco EA; Ferrarotti I; Rodríguez-Frías F; Pérez L; Vázquez I; Alonso J; Posada M; Martínez-Delgado B. Severe Alpha-1 Antitrypsin Deficiency in composite heterozygotes inheriting a new splicing mutation QOMadrid. Respiratory Research. 15, pp. 125. 01/10/2014.

DOI: 10.1186/s12931-014-0125-y

Type of production: Scientific paper

Impact source: ISI

Category: Science Edition - RESPIRATORY SYSTEM

Impact index in year of publication: 3.093

Position of publication: 18

No. of journals in the cat.: 58

9 Ruiz de Garibay G; Acedo A; García-Casado Z; Gutiérrez-Enríquez S, Tosar A, Romero A, Garre P, Llort G; Thomassen M, Díez O, Pérez-Segura P, Eduardo Díaz-Rubio E,; Velasco EA; Caldés T, de la Hoya M. Capillary Electrophoresis Analysis of Conventional Splicing Assays: IARC Analytical and Clinical Classification of 31 BRCA2 Genetic Variants. Human Mutation. 35, pp. 53 - 57. 01/01/2014.

DOI: 10.1002/humu.22456

Type of production: Scientific paper

Impact source: ISI

Category: Science Edition - GENETICS & HEREDITY

Impact index in year of publication: 5.34

Journal in the top 25%: Yes

Position of publication: 25

No. of journals in the cat.: 167

Source of citations: SCOPUS

Citations: 11

10 Infante M; Durán M; Acedo A; Sánchez-Tapia EM; Díez-Gómez B; Barroso A; García-González M; FeliubadalóL; Lasa A; de la Hoya M; Esteban-Cardeñosa E; Díez O; Martínez-Bouzas C; Godino J; Teulé A; Osorio A; Lastra E; González-Sarmiento R; Miner C; Velasco EA. The highly prevalent BRCA2 mutation c.2808_2811del (3036delACAA) is located in a mutational hotspot and has multiple origins. Carcinogenesis. 34, pp. 2505 - 2511. 08/08/2013.

DOI: 10.1093/carcin/bgt272
Type of production: Scientific paper
Corresponding author: Yes
Impact source: ISI
Category: Science Edition - ONCOLOGY
Impact index in year of publication: 5.266
Journal in the top 25%: Yes
Position of publication: 32
No. of journals in the cat.: 203

11 Pérez-Cabornero L; Infante M; Velasco EA; Lastra E; Miner C; Durán M. Genotype-phenotype correlation in MMR mutation-positive families with Lynch syndrome. International Journal of Colorectal Disease. 28, pp. 1195 - 1201. 16/04/2013.

DOI: 10.1007/s00384-013-1685-x
Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: GASTROENTEROLOGY & HEPATOLOGY
Impact index in year of publication: 2.415
Position of publication: 37
No. of journals in the cat.: 75
Source of citations: SCOPUS
Citations: 11

12 Pérez-Cabornero L; Infante M; Velasco E; Lastra E; Miner C; Durán M. Evaluating the Effect of Unclassified Variants Identified in MMR Genes Using Phenotypic Features, Bioinformatics Prediction, and RNA Assays.Journal of Molecular Diagnostics. 15, pp. 380 - 390. 20/03/2013.

DOI: 10.1016/j.jmoldx.2013.02.003
Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Pathology
Impact index in year of publication: 3.955
Journal in the top 25%: Yes
Position of publication: 13
No. of journals in the cat.: 76
Source of citations: SCOPUS
Citations: 3

13 Ana Blanco; Miguel de la Hoya; Ana Osorio; Orland Diez; María Dolores Miramar; Mar Infante; Cristina Martinez Bouzas; Asunción Torres; Adriana Lasa; Gemma Llort; Joan Brunet; Begoña Graña; Pedro Perez Segura; María José Garcia; Sara Gutiérrez Enríquez; Ángel Carracedo; María-Isabel Tejada; Eladio A Velasco; María-Teresa Calvo; Judith Balmaña; Javier Benitez; Trinidad Caldés; Ana Vega. Analysis of PALB2 gene in BRCA1/BRCA2 negative Spanish hereditary breast/ovarian cancer families with pancreatic cancer cases.PloS One. 8 - 7, pp. e67538. 2013.

DOI: 10.1371/journal.pone.0067538
Type of production: Scientific paper
Impact source: ISI
Category: Science Edition - MULTIDISCIPLINARY SCIENCES
Impact index in year of publication: 3.534
Journal in the top 25%: Yes
Position of publication: 8
No. of journals in the cat.: 55
Source of citations: SCOPUS
Citations: 16

14 Acedo A; Sanz DJ; Durán M,; Infante M; Pérez-Cabornero L; Miner C,; Velasco EA (Corresponding author). Comprehensive splicing functional analysis of DNA variants of the BRCA2 gene by hybrid minigenes. Breast Cancer Res. 14, pp. R87. 25/05/2012.
DOI: 10.1186/bcr3202
Type of production: Scientific paper
Format: Journal
Corresponding author: Yes
Impact source: ISI
Category: Science Edition - ONCOLOGY
Impact index in year of publication: 5.872
Journal in the top 25%: Yes
Position of publication: 26
No. of journals in the cat.: 197
Source of citations: SCOPUS
Citations: 16

15 Ana Blanco; Miguel de la Hoya; Judith Balmaña; Teresa Ramón y Cajal; Alex Teulé; María-Dolores Miramar; Eva Esteban; Mar Infante; Javier Benítez; Asunción Torres; María-Isabel Tejada; Joan Brunet; Begoña Graña; Milagros Balbín; Pedro Pérez Segura; Ana Osorio; Eladio A Velasco; Isabel Chirivella; María-Teresa Calvo; Lidia Feliubadaló; Adriana Lasa; Orland Díez; Angel Carracedo; Trinidad Caldés; Ana Vega. Detection of a large rearrangement in PALB2 in Spanish breast cancer families with male breast cancer.Breast cancer research and treatment. 132 - 1, pp. 307 - 315. 02/2012.
DOI: 10.1007/s10549-011-1842-2
Type of production: Scientific paper
Impact source: ISI
Category: Science Edition - ONCOLOGY
Impact index in year of publication: 4.469
Journal in the top 25%: Yes
Position of publication: 43
No. of journals in the cat.: 197
Source of citations: SCOPUS
Citations: 33

16 Ana Peixoto; Catarina Santos; Manuela Pinheiro; Pedro Pinto; Maria José Soares; Patrícia Rocha; Leonor Gusmão; António Amorim; Annemarie van der Hout; Anne-Marie Gerdes; Mads Thomassen; Torben A Kruse; Dorthe Cruger; Lone Sunde; Yves-Jean Bignon; Nancy Uhrhammer; Lucie Cornil; Etienne Rouleau; Rosette Lidereau; Drakoulis Yannoukakos; Maroulio Pertesi; Steven Narod; Robert Royer; Maurício M Costa; Conxi Lazaro; Lidia Feliubadaló; Begoña Graña; Ignacio Blanco; Miguel de la Hoya; Trinidad Caldés; Philippe Maillet; Gaelle Benais Pont; Bruno Pardo; Yael Laitman; Eitan Friedman; Eladio A Velasco; Mercedes Durán; Maria-Dolores Miramar; Ana Rodriguez Valle; María-Teresa Calvo; Ana Vega; Ana Blanco; Orland Diez; Sara Gutiérrez Enríquez; Judith Balmaña; Teresa Ramon y Cajal; Carmen Alonso; Montserrat Baiget; William Foulkes; Marc Tischkowitz; Rachel Kyle; Nelly Sabbaghian; Patricia Ashton Prolla; Ingrid P Ewald; Thangarajan Rajkumar; Luisa Mota Vieira; Giuseppe Giannini; Alberto Gulino; Maria I Achatz; Dirce M Carraro; Brigitte Bressac de Paillerets; Audrey Remenieras; Cindy Benson; Silvia Casadei; Mary-Claire King; Erik Teugels; Manuel R Teixeira. International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation.Breast cancer research and treatment. 127 - 3, pp. 671 - 679. 06/2011.
DOI: 10.1007/s10549-010-1036-3
Type of production: Scientific paper
Impact source: ISI
Category: Science Edition - ONCOLOGY
Impact index in year of publication: 4.431
Journal in the top 25%: Yes
Position of publication: 37
No. of journals in the cat.: 196
Source of citations: SCOPUS
Citations: 14
Reviews in journals: 14

17 Pérez-Cabornero L,; Borrás E, Infante M,; Velasco EA, Acedo A, Lastra E, Cuevas J, Pineda M,; Ramón y Cajal T, Capellá G, Miner C y Durán M. Characterization of new founder Alu-mediated rearrangements in MSH2 gene associated with a Lynch Syndrome phenotype. Cancer Prevention Research. 4, pp. 1546 - 1555. 2011.
DOI: 10.1158/1940-6207.CAPR-11-0227
Type of production: Scientific paper
Format: Journal

Impact source: ISI
Impact index in year of publication: 4.908
Position of publication: 34
Category: Science Edition - ONCOLOGY
Journal in the top 25%: Yes
No. of journals in the cat.: 196
Source of citations: SCOPUS
Citations: 11

18 Pérez-Cabornero L,; Infante M,; Velasco EA,; Lastra E, Acedo A, Miner C,; Durán M. Frequency of Rearrangements in Lynch Syndrome cases associated to MSH2. Characterization of a new deletion involving both EPCAM and the 5´part of MSH2. Cancer Prevention Research. 4, pp. 1556 - 1562. 2011.
DOI: 10.1158/1940-6207.CAPR-11-0080
Type of production: Scientific paper
Format: Journal
Impact source: ISI
Impact index in year of publication: 4.908
Position of publication: 34
Category: Science Edition - ONCOLOGY
Journal in the top 25%: Yes
No. of journals in the cat.: 196
Source of citations: SCOPUS
Citations: 14

19 Sanz DJ; Acedo A; Infante M; Durán M,; Pérez-Cabornero L; Esteban-Cardeñosa E, Lastra E,; Pagani F, Miner C,; Velasco EA (Corresponding author). A high proportion of DNA variants of BRCA1 and BRCA2 is associated with aberrant splicing in breast/ovarian cancer patients. Clinical Cancer Research. 16, pp. 1957 - 1967. 2010.
DOI: 10.1158/1078-0432.CCR-09-2564
Type of production: Scientific paper
Format: Journal
Corresponding author: Yes
Impact source: ISI
Impact index in year of publication: 7.338
Position of publication: 16
Category: Science Edition - ONCOLOGY
Journal in the top 25%: Yes
No. of journals in the cat.: 185
Source of citations: SCOPUS
Citations: 47

20 Infante M,; Durán M,; Acedo A,; Pérez-Cabornero L,; Sanz DJ, García-González M, Beristain E,; Esteban-Cardeñosa E, Teulé A, Vega A, Tejada I, Lastra E, Miner C; Velasco EA (Corresponding author). BRCA1 5272-1G>A and BRCA2 5374delTATG are founder mutations of high relevance for genetic counselling in breast/ovarian cancer families of Spanish origin.Clinical Genetics. 77, pp. 60 - 69. 2010.
DOI: 10.1111/j.1399-0004.2009.01272.x
Type of production: Scientific paper
Format: Journal
Corresponding author: Yes
Impact source: ISI
Impact index in year of publication: 2.942
Position of publication: 68
Category: Science Edition - GENETICS & HEREDITY
No. of journals in the cat.: 156
Source of citations: SCOPUS
Citations: 7

21 Milne RL; Osorio A; Ramón Y Cajal T; Baiget M; Lasa A; Diaz-Rubio E; de la Hoya M; Caldés T; Teulé A; Lázaro C; Blanco I; Balmaña J; Sánchez-Ollé G; Vega A; Blanco A; Chirivella I; Esteban Cardeñosa E; Durán M; Velasco E; Martínez de Dueñas E; Tejada MI; Miramar MD; Calvo MT; Guillén-Ponce C; Salazar R; San Román C; Urioste M; Benítez J.Parity and the risk of breast and ovarian cancer in BRCA1 and BRCA2 mutation carriers.Breast Cancer Research and Treatment. 119, pp. 221 - 232. 2010.
DOI: 10.1007/s10549-009-0394-1
Type of production: Scientific paper
Format: Journal
Impact source: ISI
Impact index in year of publication: 4.859
Position of publication: 34
Category: Science Edition - ONCOLOGY
Journal in the top 25%: Yes
No. of journals in the cat.: 185

Source of citations: SCOPUS **Citations:** 29

22 Infante M,; Durán M,; Lasa A, Acedo A, de la Hoya M, Esteban-Cardeñosa E,; Sanz DJ, Pérez-Cabornero L, Lastra E, Miner C,; Velasco EA (Corresponding author). Two founder BRCA2 mutations predispose to breast cancer in young women Ref. Revista /Libro:. Breast Cancer Research and Treatment. 122, pp. 567 - 571. 2010.
DOI: 10.1007/s10549-009-0661-1
Type of production: Scientific paper **Format:** Journal
Corresponding author: Yes
Impact source: ISI **Category:** Science Edition - ONCOLOGY
Impact index in year of publication: 4.859 **Journal in the top 25%:** Yes
Position of publication: 34 **No. of journals in the cat.:** 185
Source of citations: SCOPUS **Citations:** 4

23 Pérez-Cabornero L; Velasco E; Infante M; Sanz D; Lastra E; Hernández L; Miner C; Duran M. A New Strategy to Screen MMR genes in Lynch Syndrome: HA-CAE, MLPA and RT-PCR. European Journal of Cancer. 45, pp. 1485 - 1493. 2009.
DOI: 10.1016/j.ejca.2009.01.030
Type of production: Scientific paper **Format:** Journal
Impact source: ISI **Category:** Science Edition - ONCOLOGY
Impact index in year of publication: 4.121
Position of publication: 44 **No. of journals in the cat.:** 166
Source of citations: SCOPUS **Citations:** 13

24 Sánchez-Diz P; Estany-Gestal A; Aguirre C; Blanco A; Carracedo A; Ibáñez L; Passiu M; Provezza L; Ramos-Ruiz R; Ruiz B; Salado-Valdivieso I; Velasco EA; Figueiras A.Prevalence of CYP2C9 polymorphisms in the south of Europe.The Pharmacogenomics Journal. 9, pp. 306 - 310. 2009.
DOI: 10.1038/tpj.2009.16
Type of production: Scientific paper **Format:** Journal
Impact source: ISI **Category:** Science Edition - PHARMACOLOGY & PHARMACY
Impact index in year of publication: 4.398 **Journal in the top 25%:** Yes
Position of publication: 30 **No. of journals in the cat.:** 237
Source of citations: SCOPUS **Citations:** 12

25 Milne RL; Osorio A; Ramón y Cajal T; Vega A; Llort G; de la Hoya M; Díez O; Alonso MC; Lazaro C; Blanco I; Sánchez-de-Abajo A; Caldés T; Blanco A; Graña B; Durán M; Velasco E; Chirivella I; Esteban Cardeñosa E; Tejada MI; Beristain E; Miramar MD; Calvo MT; Martínez E; Guillén C; Salazar R; San Román C; Antoniou AC; Urioste M; Benítez J.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. 2008.
DOI: 10.1158/1078-0432.CCR-07-4436
Type of production: Scientific paper **Format:** Journal
Impact source: ISI **Category:** Science Edition - ONCOLOGY
Impact index in year of publication: 6.488 **Journal in the top 25%:** Yes
Position of publication: 16 **No. of journals in the cat.:** 143
Source of citations: SCOPUS **Citations:** 60

26 Esteban Cardeñosa E; Bolufer Gilabert P; Palanca Suela S; Oltra Soler S; Barragán González E; Velasco Sampedro E; Chirivella González I; Segura Huerta A; Guillén Ponce C; Martínez de Dueñas E. Twenty-three novel BRCA1 and BRCA2 sequence alterations in breast and/or ovarian cancer families of Eastern Spain. Breast Cancer Research Treatment. 112, pp. 69 - 73. 2008.

DOI: 10.1007/s10549-007-9818-y

Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Science Edition - ONCOLOGY
Impact index in year of publication: 5.684
Journal in the top 25%: No
Position of publication: 17
No. of journals in the cat.: 143
Source of citations: SCOPUS
Citations: 6

27 Velasco E (Corresponding author); Infante M; Durán M; Pérez-Cabornero L; Sanz DJ; Esteban-Cardeñosa E; Miner C.Heteroduplex analysis by capillary array electrophoresis for rapid mutation detection in large multiexon genes.Nature Protocols. 2, pp. 237 - 246. 2007.

DOI: 10.1038/nprot.2006.482

Type of production: Scientific paper
Format: Journal
Corresponding author: Yes
Impact source: ISI
Category: Science Edition - BIOCHEMICAL RESEARCH METHODS
Impact index in year of publication: 10.032 (actual)
Journal in the top 25%: Yes
Position of publication: 2
No. of journals in the cat.: 78
Source of citations: SCOPUS
Citations: 29

28 Osorio A; Martínez-Delgado B; Pollán M; Cuadros M; Urioste M; Torrenteras C; Melchor L; Díez O; De La Hoya M; Velasco E; González-Sarmiento R; Caldés T; Alonso C; Benítez J.A haplotype containing the p53 polymorphisms ins16bp and arg72pro modify cancer risk in brca2 mutation carriers. Human Mutation. 27, pp. 242 - 248. 2006.

DOI: 10.1002/humu.20283

Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 6.473
Journal in the top 25%: Yes
Position of publication: 18
No. of journals in the cat.: 131
Source of citations: SCOPUS
Citations: 26

29 Perez-Castrillon JL; Sanz A; Silva J.; Justo I.; Velasco E; Dueñas A. Calcium-sensing receptor gene A968S polymorphism and bone mass in hypertensive women.Archives of Medical Research. 37, pp. 607 - 611. 2006.

Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Science Edition - MEDICINE, RESEARCH & EXPERIMENTAL
Impact index in year of publication: 2.645
Journal in the top 25%: No
Position of publication: 57
No. of journals in the cat.: 123

30 de la Hoya M; Gutiérrez-Enríquez S; Velasco E; Osorio A; Sánchez de Abajo A; Vega A; Salazar R; Esteban E; Llort G; González-Sarmiento R; Carracedo A; Benítez J; Miner C; Díez O; Díaz-Rubio E; Caldés T.Genomic rearrangements at the BRCA1 locus in Spanish Families with Breast/Ovarian Cancer.Clinical Chemistry. 52, pp. 1480 - 1485. 2006.

DOI: 10.1373/clinchem.2006.070110

Type of production: Scientific paper
Format: Journal
Impact source: ISI

Category: Science Edition - MEDICAL LABORATORY TECHNOLOGY
Impact index in year of publication: 5.454
Journal in the top 25%: Yes
Position of publication: 2
No. of journals in the cat.: 25
Source of citations: SCOPUS
Citations: 50

31 Infante M; Durán M; Esteban-Cardeñosa E; Miner C; Velasco E* (Corresponding Author). 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. 2006.
DOI: 10.1007/s10038-006-0404-7
Type of production: Scientific paper
Format: Journal
Corresponding author: Yes
Impact source: ISI
Category: Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 2.205
Position of publication: 74
No. of journals in the cat.: 131
Source of citations: WOS
Citations: 26

32 Bolufer P; Munárriz B; Santaballa A; Velasco E; Lerma E; Barragán E (2005).Mutaciones en BRCA1 y BRCA2 en pacientes con historia familiar de cáncer de mama del Hospital La Fe. Medicina Clínica (Barc) 2005; 124: 10-12. 2005. ISSN 0025-7753
Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: MEDICINE, GENERAL & INTERNAL
Impact index in year of publication: 1.417

33 Velasco E; Infante M; Durán M; Esteban-Cardeñosa E; Lastra E; García-Girón C; Miner C. Rapid mutation detection in complex genes by heteroduplex analysis with capillary array electrophoresis (HA-CAE).Electrophoresis. 26, pp. 2539 - 2552. 2005.
DOI: 10.1002/elps.200410425
Type of production: Scientific paper
Format: Journal
Corresponding author: Yes
Impact source: ISI
Category: Science Edition - CHEMISTRY, ANALYTICAL
Impact index in year of publication: 3.85
Journal in the top 25%: Yes
Position of publication: 3
No. of journals in the cat.: 70
Source of citations: SCOPUS
Citations: 19

34 Esteban-Cardeñosa E; Durán M; Infante M; Velasco E* (corresponding author); Miner C.A high-throughput mutation detection method to scan BRCA1 and BRCA2 based on heteroduplex analysis by capillary array electrophoresis.Clinical Chemistry. 50, pp. 313 - 320. 2004.
DOI: 10.1373/clinchem.2003.023614
Type of production: Scientific paper
Format: Journal
Corresponding author: Yes
Impact source: ISI
Category: Science Edition - MEDICAL LABORATORY TECHNOLOGY
Impact index in year of publication: 6.501
Journal in the top 25%: Yes
Position of publication: 1
No. of journals in the cat.: 24
Source of citations: SCOPUS
Citations: 23

35 Diez O; Osorio A; Duran M; Martinez-Ferrandis JI; de la Hoya M; Salazar R; Vega A; Campos B; Rodriguez-Lopez R; Velasco E; Chaves J; Diaz-Rubio E; Jesus Cruz J; Torres M; Esteban E; Cervantes A; Alonso C; San Roman JM; Gonzalez-Sarmiento R; Miner C; Carracedo A; Eugenia Armengod M; Caldes T; Benitez J; Baiget M.Analysis of BRCA1 and BRCA2 genes in Spanish breast/ovarian cancer patients: A high proportion of mutations unique to Spain and evidence of founder effects.Human Mutation. 22, pp. 301 - 312. 2003.

DOI: 10.1002/humu.10260

Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 6.328
Journal in the top 25%: Yes
Position of publication: 14
No. of journals in the cat.: 120
Source of citations: SCOPUS
Citations: 129

36 Durán M; Esteban-Cardeñosa; Velasco E* (corresponding author); Infante M; Miner C. Mutational analysis of the BRCA2 gene in Spanish breast cancer patients from Castilla-Leon: identification of four novel truncating mutations.Human Mutation. 21 - MIB# 5, pp. 448. 2003.

PMID: 12655567

Type of production: Scientific paper
Format: Journal
Corresponding author: Yes
Impact source: ISI
Category: Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 6.328
Journal in the top 25%: Yes
Position of publication: 14
No. of journals in the cat.: 120
Source of citations: SCOPUS
Citations: 14

37 Velasco E; Esteban E; Infante M; Durán M; Lastra E.; García-Girón; C.; Miner C .Estudio molecular de los genes BRCA1 y BRCA2 en 153 familias con cáncer de mama de Castilla y León (España): Identificación de nueve variantes de efecto desconocido no descritas.Medicina Clínica (Barc.). 119, pp. 441 - 445. 2002. ISSN 0025-7753

Type of production: Scientific paper
Format: Journal
Impact source: ISI
Impact index in year of publication: 1.399

38 Alonso MJ; Blanco A; Fernández-Burriel M; Calleja J; Fernández I; Sanz A; Velasco E; Telleria JJ. Polyvariant mutant genes: different haplotypes determining different alterations causing azoospermia. Gene Function & Disease (Desde 2002, Comparative and Functional Genomics). 1, pp. 189 - 193. 2000.

Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Science Edition - BIOTECHNOLOGY & APPLIED MICROBIOLOGY
Impact index in year of publication: 2.032
Journal in the top 25%: No
Position of publication: 85
No. of journals in the cat.: 163

39 De la Puente A; Velasco E; Pérez-Jurado LA; Hernández-Chico C; van de Rijke FM; Scherer SW; Raap AK; Cruces J. Analysis of the monomeric alphoid sequences in the pericentromeric region of human chromosome 7.Cytogenetics Cell Genetics (ahora Cytogenet Genome Res) (). 83, pp. 176 - 181. 1998. ISSN 0301-0171

Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 1.561
Journal in the top 25%: No

40 Redondo Mateo J; Velasco Sampedro E; Gómez Barrero S; Rodríguez Sanz F; Clavería Soria LE. Estudio clínico y genético de familias españolas afectadas de la enfermedad de Darier. Actas Dermosifiliográficas. 89, pp. 519 - 529. 1998.
Type of production: Scientific paper **Format:** Journal

41 Valero C; Castroviejo IP; Velasco E; Moreno F, Hernández-Chico C. Identification of de novo deletions at the NF1 gene: no preferential paternal origin and phenotypic analysis of patients.Human Genetics. 99, pp. 720 - 726. 1997.
DOI: 10.1007/s004390050438
Type of production: Scientific paper **Format:** Journal
Impact source: ISI **Category:** Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 2.662
Position of publication: 30 **No. of journals in the cat.:** 90
Source of citations: SCOPUS **Citations:** 32

42 Duarte J; Velasco E.Distonía sensible a DOPA. ALDE. 4, pp. 3 - 4. 1996.
Type of production: Scientific paper **Format:** Journal

43 Valero C; Velasco E; Valero A; Moreno F; Hernández-Chico C. Linkage disequilibrium between four intragenic polymorphic microsatellites of the NF1 gene and its implications for genetic counselling. Journal of Medical Genetics. 33, pp. 590 - 593. 1996.
PMID: 8818946
Type of production: Scientific paper **Format:** Journal
Impact source: ISI **Category:** Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 6.335 **Journal in the top 25%:** Yes
Position of publication: 18 **No. of journals in the cat.:** 167
Source of citations: SCOPUS **Citations:** 11

44 Velasco E; Valero C; Valero A; Moreno F; Hernández-Chico C.Molecular analysis of the SMN and NAIP genes in Spanish Spinal Muscular Atrophy (SMA) families and correlation between number of copies of cBCD541 and SMA phenotype. Human Molecular Genetics. 5, pp. 257 - 264. 1996.
PMID: 8824882
Type of production: Scientific paper **Format:** Journal
Impact source: ISI **Category:** Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 8.505 **Journal in the top 25%:** Yes
Position of publication: 18 **No. of journals in the cat.:** 253
Source of citations: SCOPUS **Citations:** 165

45 Hernández-Chico C; Velasco E; Valero C; García E; Moreno F. Diagnóstico Prenatal de atrofia muscular espinal. Anales Españoles de Pediatría. 42, pp. 429 - 435. 1995.
Type of production: Scientific paper **Format:** Journal

46 Viribay M; Tellería D; Velasco E; Moreno F; San Millán JL.Dinucleotide repeat polymorphism at the D4S2458 locus close to the PKD2 locus on human chromosome 4q. Human Genetics. 95, pp. 601 - 602. 1995.
DOI: 10.1007/BF00223883
Type of production: Scientific paper **Format:** Journal
Impact source: ISI **Category:** Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 2.758

47 Velasco E; Valero C; García E; de la Puente A; Cruces J; San Millán JL; del Castillo I; Coloma A; Moreno F; Hernandez-Chico C. Isolation of microsatellites from the spinal muscular atrophy (SMA) candidate region on chromosome 5q and linkage analysis in Spanish SMA families.European Journal of Human Genetics. 3, pp. 96 - 101. 1995. ISSN 1018-4813

Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 4.349
Journal in the top 25%: Yes
Position of publication: 36
No. of journals in the cat.: 167

48 Valero MC; Velasco E; Moreno F; Hernández-Chico C.Characterization of four mutations in the neurofibromatosis type I gene by denaturing gradient gel electrophoresis (DGGE).Human Molecular Genetics. 3, pp. 639 - 641. 1994.

DOI: 10.1093/hmg/3.4.639
Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 4.528
Journal in the top 25%: Yes
Source of citations: SCOPUS
Citations: 32

49 Velasco E; de la Puente A; Cruces J; Valero MC; Garcia-Patiño E; del Castillo I; Coloma A; Moreno F; Hernandez-Chico C.Dinucleotide repeat polymorphism at the D5S1356, D5S1357 and D7S1480 loci. Human Molecular Genetics. 3, pp. 1441. 1994.

DOI: 10.1093/hmg/3.8.1441
Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 4.528
Journal in the top 25%: Yes

50 Velasco E; Sánchez-Corral P; Moreno F; Rodríguez de Córdoba S. Dinucleotide repeat polymorphism between the human C4BPA y C4BPB gene loci. Human Molecular Genetics. 1, pp. 552. 1992.

DOI: 10.1093/hmg/1.7.552-a
Type of production: Scientific paper
Format: Journal
Impact source: ISI
Category: Science Edition - GENETICS & HEREDITY
Impact index in year of publication: 3.783
Journal in the top 25%: Yes
Source of citations: SCOPUS
Citations: 3

51 Velasco E; Bussaglia E; Valero C; Hernandez C; Grinberg D; Balcells S; Moreno F; Baiget M.Linkage analysis in spinal muscular atrophy disease using chromosome 5 markers in 32 Spanish families. American Journal of Human Genetics. 51 - 4 Suppl.:, pp. A204. 1992.

Type of production: 42nd Annual Meeting of the American Society of Human Genetics, San Francisco, California, USA, November 9-13, 1992.
Format: Journal

Works submitted to national or international conferences

1 **Title of the work:** BRCA2 mis-splicing: exons 17 and 18 regulation
Name of the conference: European Human Genetics Conference 2017
City of event: Copenhagen, Denmark
Date of event: 27/05/2017
End date: 30/05/2017
Organising entity: European Society of Human Genetics
Fraile Bethencourt E; Díaz Gomez B; Valenzuela-Palomo A; Acedo A; Sanz DJ; Goina E; Buratti E; Velasco EA.

2 **Title of the work:** BRCA2 mis-splicing: exons 17 and 18 regulation
Name of the conference: European Human Genetics Conference 2017
City of event: Copenhagen, Denmark
Date of event: 27/05/2017
End date: 30/05/2017
Organising entity: European Society of Human Genetics
Fraile Bethencourt E; Díaz Gomez B; Valenzuela-Palomo A; Acedo A; Sanz DJ; Goina E; Buratti E; Velasco EA.

3 **Title of the work:** ANÁLISIS FUNCIONALES CONFIRMAN LA PATOGENICIDAD DE UNA VARIANTE DE SPLICING EN EL GEN CHD7 HALLADA MEDIANTE SECUENCIACIÓN MASIVA
Name of the conference: I Congreso Interdisciplinar en Genética Humana
City of event: Madrid,
Date of event: 25/04/2017
End date: 28/04/2017
Organising entity: Asociación española de Genética Humana
Villate O; Fraile-Bethencourt E; Valenzuela A; Velasco EA; Grozeva D; Raymond L; Botella MP; Tejada MI.

4 **Title of the work:** BRCA 2 MIS-SPLICING: REGULACIÓN DE LOS EXONES 17 Y 18
Name of the conference: I Congreso Interdisciplinar en Genética Humana
City of event: Madrid,
Date of event: 25/04/2017
End date: 28/04/2017
Organising entity: Asociación Española de Genética Humana
Fraile Bethencourt E; Díaz Gomez B; Valenzuela A; Acedo A; Sanz DJ; Goina E; Buratti E; Velasco EA.

5 **Title of the work:** CARACTERIZACIÓN FUNCIONAL DE VARIANTES CANDIDATAS DE SPLICING EN GENES DE SUSCEPTIBILIDAD MEDIANTE MINIGENES HÍBRIDOS: PALB2
Name of the conference: I Congreso Interdisciplinar en Genética Humana
City of event: Madrid,
Date of event: 25/04/2017
End date: 28/04/2017
Organising entity: Asociación Española de Genética Humana
Valenzuela Palomo A; Fraile Bethencourt E; Díez Gómez B; Entrala C; Martínez- Delgado B; Villate O; Tejada MI; Velasco EA.

6 **Title of the work:** Functional characterization of DNA variants from exons 17 and 18 of the BRCA2 gene
Name of the conference: European Human Genetics Conference 2016
Type of event: Conference
Type of participation: 'Participatory - poster
City of event: Barcelona, Spain
Date of event: 21/05/2016
End date: 24/05/2016
Organising entity: European Society of Human Genetics
Fraile-Bethencourt E; Díez-Gómez B; Velásquez-Zapata V; Acedo A; Sanz DJ; Hernandez-Moro C; Marcos G; Infante M; Durán M; Velasco EA.

7 **Title of the work:** Caracterización funcional de mutaciones reguladoras en el promotor de BRCA2 en cáncer de mama y ovario hereditario
Name of the conference: XL CONGRESO DE LA SOCIEDAD ESPAÑOLA DE GENÉTICA
Corresponding author: Yes
City of event: Córdoba,
Date of event: 16/09/2015
End date: 18/09/2015
Organising entity: SOCIEDAD ESPAÑOLA DE GENÉTICA
Díez B; Fraile E; Infante M; Durán M; Velasco EA.

8 **Title of the work:** Clasificación funcional y clínica de variantes de ADN del gen BRCA2 mediante minigenes híbridos
Name of the conference: XL CONGRESO DE LA SOCIEDAD ESPAÑOLA DE GENÉTICA
Type of participation: Participatory - oral communication
Corresponding author: Yes
City of event: Córdoba,
Date of event: 16/09/2015
End date: 18/09/2015
Organising entity: SOCIEDAD ESPAÑOLA DE GENÉTICA
Fraile E; Velásquez V; Hernández C; Díez B; Sanz DJ; Acedo A; Infante M; Velasco EA.

9 **Title of the work:** CONSTRUCCIÓN DE UN MINIGEN HÍBRIDO PARA EL ESTUDIO DE MUTACIONES DE SPLICING EN LOS EXONES 17 Y 18 DE BRCA2
Name of the conference: XXVIII Congreso Nacional de Genética Humana
Corresponding author: Yes
City of event: Palma de Mallorca,
Date of event: 13/05/2015
End date: 15/05/2015
Organising entity: Asociación Española de Genética Humana
FRAILE E; Velásquez V; HERNÁNDEZ MORO C; Díez B; Sanz DJ; Acedo A; Infante M; Velasco EA.

10 **Title of the work:** IDENTIFICACIÓN DE ALTERACIONES DE SPLICING EN EL GEN BRCA1 MEDIANTE MINIGENES HÍBRIDOS
Name of the conference: XXVIII Congreso Nacional de Genética Humana
Corresponding author: Yes
City of event: Palma de Mallorca,
Date of event: 13/05/2015
End date: 15/05/2015
Organising entity: Asociación Española de Genética Humana
HERNÁNDEZ MORO C; Curiel A; Acedo A; FRAILE E; Díez B; Infante M; Durán M; Velasco EA.

11 **Title of the work:** New splicing vector pSAD: Splicing functional analysis of a hybrid “maxi-minigene” with exons 19 to 27 of BRCA2
Name of the conference: European Human Genetics Conference 2013
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: París, France
Date of event: 08/06/2013
End date: 11/06/2013
Organising entity: European Society of Human Genetics
Acedo A; Díez-Gómez B; Curiel A; Hernández-Moro C; Infante M; Miner C; Durán M; Velasco EA.

12 **Title of the work:** Splicing functional assays of a BRCA1 minigene with exons 15-19
Name of the conference: European Human Genetics Conference 2013
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: París, France
Date of event: 08/06/2013
End date: 11/06/2013
Organising entity: European Society of Human Genetics
Hernández-Moro C; Curiel A; Díez-Gómez B; Acedo A; Infante M; Miner C; Durán M; Velasco EA.

13 **Title of the work:** Estudios funcionales de splicing de los exones 16 y 17 de brca1 mediante minigenes híbridos
Name of the conference: XXVII Congreso Nacional de Genética Humana
Type of event: Conference
Type of participation: 'Participatory - poster
City of event: Madrid,
Date of event: 10/04/2013
End date: 12/04/2013
Organising entity: Asociación Española de Genética Humana
Hernández-Moro C; Curiel-García A; Díez Gómez B; Acedo; Infante M; Durán M; Velasco EA.

14 **Title of the work:** Nuevo vector de splicing pSAD. Validación mediante ensayos funcionales de splicing en el "maxi-minigen"con los exones 19 a 27 de BRCA2
Name of the conference: XXVII Congreso Nacional de Genética Humana
Type of event: Conference
Type of participation: 'Participatory - poster
City of event: Madrid,
Date of event: 10/04/2013
End date: 12/04/2013
Organising entity: Asociación Española de Genética Humana
Acedo; Díez Gómez B; Curiel-García A; Hernández-Moro C; Infante M; Durán M; Velasco EA.

15 **Title of the work:** New splicing vector pSAD: Splicing functional analysis of a hybrid “maxi-minigene” with exons 19 to 27 of BRCA2
Name of the conference: IV Annual IMPPC Conference/1st ICO-IDIBELL Hereditary Cancer Program Meeting.
Type of event: Conference
Type of participation: 'Participatory - poster
City of event: Barcelona, Spain

Date of event: 14/03/2013
End date: 15/03/2013
Organising entity: Instituto Catalán de Oncología. IMPCC
City organizing entity: Barcelona, Spain
Acedo A; Díez-Gómez B; Curiel A; Hernández-Moro C; Infante M; Durán M; Velasco EA.

16 **Title of the work:** "Splicing y cáncer de mama y ovario hereditario"
Name of the conference: JORNADA NACIONAL DE GRUPOS DE TRABAJO EN CÁNCER HEREDITARIO
Type of event: Workshop
Type of participation: Participatory - invited/keynote talk **Reasons for participation:** Upon invitation
City of event: Barcelona,
Date of event: 13/03/2013
Organising entity: ICO-IMPCC **Type of entity:** Public Research Body
Eladio A. Velasco Sampedro.

17 **Title of the work:** Distribution of mutations in breast/ovarian cancer susceptibility genes in the north-east of Spain.
Name of the conference: European Human Genetics Conference 2012
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Nüremberg, Germany
Date of event: 23/06/2012
Organising entity: European Society of Human Genetics
1; Infante M; Esteban Cardeñosa EM; Velasco EA; Lastra Aras E; Marcos García G; Hernández Sanz L; Martínez Martín N; Durán Domínguez M; Miner C.

18 **Title of the work:** Frequency of germ line MUTYH mutations in patients diagnosed with colorectal cancer in Castilla y León (Spain)
Name of the conference: European Human Genetics Conference 2012
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Nüremberg, Germany
Date of event: 23/06/2012
Organising entity: European Society of Human Genetics
Tascón M; Pérez-Cabornero L; Infante M; Velasco EA; Lastra E; Marcos G; Esteban-Cardeñosa E; Hernández Sanz L; Miner C; Durán M.

19 **Title of the work:** Splicing functional assays of a single minigene with eight exons of the BRCA2 gene
Name of the conference: European Human Genetics Conference 2012
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Nüremberg, Germany
Date of event: 23/06/2012
Organising entity: European Society of Human Genetics
Acedo A; Díez-Gómez B; Curiel A; Hernández-Moro C; Infante M; Miner C; Durán M; Velasco EA.

20 **Title of the work:** Splicing aberrante de BRCA1 y BRCA2 y susceptibilidad genética a cáncer de mama/ovario"
Name of the conference: SEMINARIOS DEL SERVICIO DE GENÉTICA
Type of event: Seminar

Type of participation: Participatory - invited/keynote talk **Reasons for participation:** Upon invitation
City of event: Barcelona,
Date of event: 29/11/2011
Organising entity: HOSPITAL DE LA SANTA CREU I SANT PAU
Eladio Velasco Sampedro.

21 **Title of the work:** "Análisis funcional de splicing de variantes de ADN de los genes BRCA1 y BRCA2 mediante minigenes híbridos"
Name of the conference: XXXVIII Congreso de la Sociedad Española de Genética
Type of event: Conference **Geographical area:** National
Type of participation: Participatory - oral communication
City of event: Murcia,
Date of event: 21/09/2011
Organising entity: Sociedad Española de Genética
Acedo A, Sanz DJ, Durán M, Infante M, Lucía Pérez-Cabornero, Beatriz Díez, Cristina Miner; Velasco EA.

22 **Title of the work:** Analysis of PALB2 gene in BRCA1/2 negative Spanish Hereditary Breast Ovarian Cancer Families with pancreatic cancer cases.
Name of the conference: European Human Genetics Conference 2011
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Amsterdam, Holland
Date of event: 28/05/2011
Organising entity: European Society of Human Genetics
Blanco A; Diez O; de la Hoya M; García M J; Miramar MD; Infante M; Martínez Bouzas C; Torres A; Lasa A; Llort G; Brunet J; Bosch N; Pérez Segura P; Osorio A; Calvo MT; Velasco E; Tejada MI; Caldés T; J. Benitez; Carracedo A; Balmaña J; Vega A. "Analysis of PALB2 gene in BRCA1/2 negative Spanish Hereditary Breast Ovarian Cancer Families with pancreatic cancer cases".

23 **Title of the work:** Influence of MLH1 -93G>A promoter polymorphism in hereditary vs. sporadic colon cancer
Name of the conference: European Human Genetics Conference 2011
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Amsterdam, Holland
Date of event: 28/05/2011
Organising entity: European Society of Human Genetics
Durán M, Martínez N, Pérez-Cabornero L, Infante M,; E. Velasco,; C. Miner. "Influence of MLH1 -93G>A promoter polymorphism in hereditary vs. sporadic colon cancer.".

24 **Title of the work:** Mutation screening of RAD51C in Breast and Ovarian Cancer families from Castilla-León (Spain)
Name of the conference: European Human Genetics Conference 2011
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Amsterdam, Holland
Date of event: 28/05/2011
Organising entity: European Society of Human Genetics
Infante M; Durán M; Díez-Gómez B; Curiel A; A. Acedo, Pérez-Cabornero L; Lastra E; Marcos G; Miner C; Velasco EA. "Mutation screening of RAD51C in Breast and Ovarian Cancer families from Castilla-León (Spain).".

25 **Title of the work:** EFECTO DEL POLIMORFISMO MLH1-93G>A EN CÁNCER COLORRECTAL
Name of the conference: XXVI Congreso Nacional de Genética Humana
Type of event: Conference **Geographical area:** National
Type of participation: 'Participatory - poster
City of event: Murcia,
Date of event: 30/03/2011
Organising entity: Asociación Española de Genética Humana
Martínez N; Pérez-Cabornero L; Infante M; Velasco E; Acedo A; Lastra E; Martínez N; Hernández L; Miner C; Durán M. "ESTUDIO MUTACIONAL DEL GEN RAD51C EN PACIENTES CON CÁNCER DE MAMA Y OVARIO DE LA ZONA ESTE DE CASTILLA Y LEÓN".

26 **Title of the work:** ESTUDIO MUTACIONAL DEL GEN RAD51C EN PACIENTES CON CÁNCER DE MAMA Y OVARIO DE LA ZONA ESTE DE CASTILLA Y LEÓN
Name of the conference: XXVI Congreso Nacional de Genética Humana
Type of event: Conference **Geographical area:** National
Type of participation: 'Participatory - poster
City of event: Murcia,
Date of event: 30/03/2011
Organising entity: Asociación Española de Genética Humana
Infante M; Durán M; Díez-Gómez B, Curiel A, Acedo A, Pérez-Cabornero L, Lastra E, Marcos G, Miner C; Velasco EA. "ESTUDIO MUTACIONAL DEL GEN RAD51C EN PACIENTES CON CÁNCER DE MAMA Y OVARIO DE LA ZONA ESTE DE CASTILLA Y LEÓN".

27 **Title of the work:** Analysis of the splicing effect of 28 DNA variants from BRCA2 exons 19 and 20 by hybrid minigenes
Name of the conference: French workshop on RNA splicing and genetic diseases
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Paris, France
Date of event: 14/10/2010
Organising entity: EURASNET (European Alternative Splicing Network) **Type of entity:** Associations and Groups
A. Acedo; M. Durán; D. J. Sanz; M. Infante; L. Pérez-Cabornero; E. Lastra; C. Miner; E.A. Velasco.

28 **Title of the work:** Evaluation of pathogenicity of single-base germline changes involving the mismatch repair genes MLH1, MSH2 and MSH6 in diagnostics of Lynch syndrome
Name of the conference: European Human Genetics Conference 2010
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Gothenburg, Sweden
Date of event: 12/06/2010
Organising entity: European Society of Human Genetics
Pérez-Cabornero L; Infante Sanz M; Velasco Sampedro E; Lastra Aras E; Cuevas González J; Acedo Bécares A; Alonso Ramos M; Fernández Carvajal I; Hernández Sanz L; Martínez Martín N; Miner Pino C; Durán Domínguez M.

29 **Title of the work:** Risk measure for expansion upon transmission in FMR1 grey alleles.
Name of the conference: European Human Genetics Conference 2010
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Gothenburg, Sweden

Date of event: 12/06/2010
Organising entity: European Society of Human Genetics
B. Lopez; J. A. Garrote; E. Velasco; M. J. Alonso; M. Duran; A. Blanco; I.Fernández-Carvajal.

30 **Title of the work:** Splicing Functional Assays of DNA variants of the BRCA1 and BRCA2 genes.
Name of the conference: European Human Genetics Conference 2010
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Gothenburg, Sweden
Date of event: 12/06/2010
Organising entity: European Society of Human Genetics
Acedo A; Sanz DJ; Pérez-Cabornero L; Infante M; Durán M; Esteban-Cardeñosa E; Miner C; Velasco EA.

31 **Title of the work:** Two deleterious BRCA1 and BRCA2 mutations in a Spanish family.
Name of the conference: European Human Genetics Conference 2010
Type of event: Conference **Geographical area:** European Union
Type of participation: 'Participatory - poster
City of event: Gothenburg, Sweden
Date of event: 12/06/2010
Organising entity: European Society of Human Genetics
Infante M; Durán M; Sanz D; Pérez-Cabornero L; Acedo A; Lastra E; Hernández L; Martínez N; Miner C; Velasco E.

32 **Title of the work:** Effect of DNA Variants from BRCA2 Exons 19 and 20 on the Splicing Process by Hybrid Minigenes
Name of the conference: 4th FAMILIAL CANCER CONFERENCE
Type of event: Conference **Geographical area:** Internacional
Type of participation: 'Participatory - poster
City of event: Madrid,
Date of event: 07/06/2010
Organising entity: Centro Nacional de Investigaciones Oncológicas **Type of entity:** Healthcare Institutions
Acedo A; Sanz DJ; Infante M; Pérez-Cabornero L; Lastra E; Miner C; Durán M; Velasco EA.

R&D management and participation in scientific committees

Scientific, technical and/or assessment committees

1 **Committee title:** Comisión de Cáncer Familiar-Asociación Española de Genética Humana
Primary (UNESCO code): 240900 - Genetics; 241007 - Human genetics
Secondary (UNESCO code): 320713 - Oncology
Affiliation entity: Asociación Española de Genética Humana
Start-End date: 2008 - 01/04/2013

2 **Committee title:** Comité Científico Asesor
Primary (UNESCO code): 241007 - Human genetics; 241500 - Molecular biology
Affiliation entity: AC-GEN Reading Life SL
City affiliation entity: Valladolid, Spain
Start date: 16/06/2013

Organization of R&D activities

1 **Title of the activity:** DNA challenge
Type of activity: Miembro del Jurado del Concurso Día ADN **Geographical area:** National
Convening entity: AC-GEN Reading Life
City convening entity: Valladolid, Spain
Start-End date: 25/04/2014 - 25/04/2014

2 **Title of the activity:** II Simposio de Cáncer Hereditario
Type of activity: Miembro del Comité Científico **Geographical area:** National
Convening entity: Hospital General Yagüe **Type of entity:** Healthcare Institutions
City convening entity: Burgos, Castile and León, Spain
Start date: 17/04/2008 **Duration:** 2 days

3 **Title of the activity:** Congreso de la Asociación Española de Genética Humana
Type of activity: Miembro del Comité Organizador y Científico **Geographical area:** National
Convening entity: Asociación Española de Genética Humana
City convening entity: Valladolid, Castile and León, Spain
Start date: 15/06/2006 **Duration:** 2 days

Evaluation and revision of R&D projects and articles

1 **Name of the activity:** Evaluación de 10 proyectos
Performed tasks: EVALUADOR DE PROYECTOS NACIONALES
Entity where activity was carried out: Agencia Nacional de Evaluación y Prospectiva **Type of entity:** -
Start-End date: 2010 - 2018

2 **Performed tasks:** Revisor de artículos-revista Clinical Genetics
Entity where activity was carried out: Wiley-Blackwell
Type of activity: Review of articles in scientific or technological journals **Frequency of the activity:** 2
Geographical area: Non EU International
Start-End date: 2008 - 2011

3 **Performed tasks:** Evaluador de 8 proyectos de investigación - Convocatoria de ayudas para la financiación de la investigación biomédica básica traslacional en oncología y oncohematología para el año 2017.
Entity where activity was carried out: Consejería de Salud Junta de Andalucía
Start date: 2018

4 **Name of the activity:** Referee
Performed tasks: Revisor- Frontiers in Genetics
Entity where activity was carried out: Frontiers
Type of activity: Review of articles in scientific or technological journals
Geographical area: Non EU International
Start date: 2018

5 **Performed tasks:** Evaluador de 7 proyectos de investigación - Consejería de Salud
Entity where activity was carried out: Fundación Pública Andaluza Progreso y Salud - Junta de Andalucía
Start date: 2016

6 **Performed tasks:** Evaluador del Biobanco del Sistema Sanitario Público de Andalucía (SSPA). Miembro del Comité Científico provisional
Entity where activity was carried out: Fundación Pública Andaluza Progreso y Salud - Junta de Andalucía
Start date: 01/07/2013

7 **Name of the activity:** Referee
Performed tasks: Referee-Carcinogenesis
Entity where activity was carried out: Oxford University Press
Type of activity: Review of articles in scientific or technological journals **Frequency of the activity:** 2
Geographical area: Non EU International
Start date: 2013

8 **Performed tasks:** Revisor- Human Mutation
Entity where activity was carried out: Wiley-Blackwell
Type of activity: Review of articles in scientific or technological journals **Frequency of the activity:** 2
Geographical area: Non EU International
Start date: 2013

9 **Performed tasks:** EVALUADOR DE CONTRATOS AECC INVESTIGADORES MODALIDAD A
Entity where activity was carried out: Agencia Nacional de Evaluación y Prospectiva **Type of entity:** MINECO
Frequency of the activity: 2
Geographical area: National
Start date: 2012

10 **Performed tasks:** Revisor-Clinical Genetics
Entity where activity was carried out: Wiley-Blackwell
Type of activity: Review of articles in scientific or technological journals **Frequency of the activity:** 2
Geographical area: Non EU International
Start date: 2011

11 **Performed tasks:** Revisor-Journal of Medical Genetics
Entity where activity was carried out: BMJ group
Type of activity: Review of articles in scientific or technological journals
Geographical area: Non EU International
Start date: 2011

12 **Performed tasks:** Revisor revista Familial Cancer
Entity where activity was carried out: Springer
Type of activity: Review of articles in scientific or technological journals **Frequency of the activity:** 1
Geographical area: Non EU International
Start date: 2010

13 **Performed tasks:** Revisor de artículos-revista Clinical Chemistry
Entity where activity was carried out: The American Association for Clinical Chemistry
Type of activity: Review of articles in scientific or technological journals **Frequency of the activity:** 2
Geographical area: Non EU International
Start date: 2009

14 **Performed tasks:** Revisor-Revista Cancer Detection and Prevention
Entity where activity was carried out: International Society for Preventive Oncology
Type of activity: Review of articles in scientific or technological journals **Frequency of the activity:** 1
Geographical area: Non EU International
Start date: 2006

15 **Performed tasks:** Revisor de artículos-Electrophoresis
Entity where activity was carried out: Editorial Wiley-VCH
Type of activity: Review of articles in scientific or technological journals
Geographical area: Non EU International
Start date: 2005

16 **Performed tasks:** Revisor de artículos-revista Biotechniques
Type of activity: Review of articles in scientific or technological journals
Geographical area: Non EU International
Start date: 2005

17 **Performed tasks:** Revisor de artículos-revista Clinical Chemistry
Entity where activity was carried out: The American Association for Clinical Chemistry
Type of activity: Review of articles in scientific or technological journals **Frequency of the activity:** 2
Geographical area: Non EU International
Start date: 2005

Other achievements

Stays in public or private R&D centres

Entity: International Centre of Genetic Engineering and Biotechnology **Type of entity:** Public Research Body
City of entity: Trieste, Italy
Start date: 02/11/2006 **Duration:** 2 months - 17 days
Goals of the stay: Científico Visitante
Provable tasks: Aprendizaje de ensayos funcionales de splicing mediante minigenes híbridos

Scientific societies and professional associations

1 **Name of the society:** Socio Sociedad Española de Genética
Affiliation entity: Sociedad Española de Genética
Start date: 2011

2 **Name of the society:** Socio SEOM
Affiliation entity: Sociedad Española de Oncología Médica (SEOM)
Start date: 2005

3 **Name of the society:** Socio de la Asociación Española de Genética Humana
Affiliation entity: Asociación Española de Genética Humana
Professional category: Vocal de la Comisión de Cáncer Familiar (2008-actualidad)
Start date: 2001

Prizes, mentions and distinctions

1 **Description:** Premio a la mejor comunicación del XXVII Congreso Nacional de la AEGH
Awarding entity: Asociación Española de Genética Humana
City awarding entity: Madrid, Spain
Conferral date: 12/04/2013

2 **Description:** Premio Junta de Castilla y León" de la Real Academia de Medicina y Cirugía de Valladolid (2009). Trabajo: Análisis molecular de los genes BRCA1 Y BRCA2 en pacientes de Castilla y León con cáncer de mama y ovario hereditarios como estrategia de prevención eficaz de esta enfermedad"
Awarding entity: REAL ACADEMIA DE MEDICINA Y CIRUGIA DE VALLADOLID
Conferral date: 2009

3 **Description:** X Premio 3M a la Innovación (2008). Área de Salud. Trabajo PREVENCIÓN EFICAZ DE CÁNCER DE MAMA Y OVARIO HEREDITARIOS: DESARROLLO DE UN MÉTODO RÁPIDO Y SENSIBLE PARA LA DETECCIÓN DE ALTERACIONES EN GENES DE PREDISPOSICIÓN HEREDITARIA"
Awarding entity: Fundación 3M **Type of entity:** Foundation
Conferral date: 2008

4 **Description:** Acreditación en Genética Humana: Reconocimiento de méritos en Asistencia Sanitaria, Investigación y Docencia en Genética Humana
Awarding entity: Asociación Española de Genética Humana
Conferral date: 2007

5 **Description:** Premio de la Real Academia de Medicina y Cirugía de Valladolid al trabajo Efectos del polimorfismo A986S del receptor sensor del calcio sobre la densidad mineral y marcadores del remodelado de mujeres hipertensas"
Awarding entity: REAL ACADEMIA DE MEDICINA Y CIRUGIA DE VALLADOLID
Conferral date: 2005

6 **Description:** Premio a la mejor Comunicación-Póster con el trabajo Importancia de un rastreo exhaustivo del gen CFTR en los pacientes que presentan fenotipos atípicos F.Q."
Awarding entity: Sociedad Española de Fibrosis Quística
City awarding entity: Murcia

Conferral date: 1999

7 **Description:** Premio Profesor Joaquín Piñol Aguadé. Trabajo: Estudio clínico y genético de familias afectadas de la enfermedad de Darier"
Awarding entity: Academia Española de Dermatología y Venereología
Type of entity: Sociedad Científica
Conferral date: 1998