



CURRÍCULUM VÍTAE NORMALIZADO



Julio Escribano Martínez

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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.

Julio Escribano, Ph.D., is Professor of Genetics at Castilla-La Mancha University Medical School since 2010. In 1992 he completed a PhD at Hospital Ramón y Cajal and Complutense University of Madrid (Spain) on human plasma protein alpha-1 microglobulin characterization. During the predoctoral period, he made several short-term research visits at the universities of Lund (Sweden) and Odense (Denmark). His postdoctoral training took place in the Department of Ophthalmology and Visual Science of Yale University (USA) (1992-1994). In 1994 Dr. Escribano obtained a postdoctoral contract from the Spanish Science and Education Ministry to start a scientific career in Spain, at the Biotechnology Division of the Institute for Regional Development (IDR, UCLM). From 1996-1998 and from 1998-2001 he served as an Assistant Professor at the IDR and at the Medical School of UCLM, respectively. He was appointed as Associate Professor of Genetics at the Medical School (2001) where he was Vice Dean (2001-2004), contributing to implement an innovative medical curriculum, and established the laboratory of Human Molecular Genetics (2001).

During his postdoctoral training in USA he performed the first transcriptomic analysis of the ciliary body using different cDNA libraries and identified more than 700 expressed sequence tags (ESTs), which are part of the ciliary body section of the NEIBank database (<https://neibank.nei.nih.gov/index.shtml>). These studies led to discover several key genes in ciliary body physiology and glaucoma, including PEDF, MYOC and TGFB1. The two later genes were further identified by other groups as the first genes involved in glaucoma and dominant corneal dystrophies, respectively. In addition, the finding of ESTs encoding neuropeptide processing enzymes demonstrated the neuroendocrine properties the ciliary body. In the Biotechnology Section of IDR, he started pioneer studies on biomedical applications of saffron compounds and molecular diagnosis of brucellosis. His initial work as PI of the Human Genetics group contributed to identify the PEDF receptor, to demonstrate the role of PEDF as a niche signal for neural stem cell renewal and to identify genetic risk factors in urological cancers. In recent years his research has focused on studying genetic and molecular bases of glaucoma. Over the past 20 years Dr. Escribano has obtained continuous competitive funding from different sources and since 2008 his group belong to OFTARED, a Spanish research network funded by the Institute of Health Carlos III. Dr Escribano is co-author of four patents, he has published more than 100 indexed journal articles in collaboration with national and international groups, and has directed 12 PhD theses and several master works. He has also obtained six positive evaluations of research activity, including one positive evaluation of translational activity. In addition, his lab has discovered the intracellular proteolytic processing of recombinant myocilin and the presence of this protein in the human blood plasma, providing new insights to explore the biological function of this molecule and to understand the role of MYOC mutations in glaucoma pathogenesis. His work has also identified different gene variants as glaucoma risk factors, including the causative role of rare loss-of-function CYP1B1. Using exome sequencing, CRISPR/Cas9 genome editing and zebrafish as animal model, GPATCH3, GUCA1C and CPAMD8 have been identified to play putative or demonstrated roles in congenital glaucoma



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.

- 1. Six-y ears-positive evaluations of research activity (CNEAI):** 6 (latest 2014-2019).
- 2. JCR articles:** 104 (4-2022). **Q1:** 54% (15% D1). **Q2:** 36%. **First author:** 15%. **Last author:** 40%.
- 3. PhD thesis directed:** 12.
- 4. h-index** (4-2022): 31 (Scopus), 33 (Research Gate), 38 (Google Scholar)
- 5. 10-index** (4-2022): 87 (Google Scholar).
- 6. Total cites** (5-2021) : 3515 (Scopus); 4111 (Research Gate), 5548 (Google Scholar).
- 7. Average cites per year:>115. Average cites per article: >32.**
- 8. Research Gate score (4-2022):** 39,38 (higher than 95% of Research Gate members).
- 9. Top-five impact factor papers:** 15,664 (Nat. Neuroscience), 7,667 (European Urology), 7,725 (Prog. Ret. Eye Res) 6.693 (JBC, two articles) and 6,17 (Ophthalmology).
- 10. Patents:** 4.
- 11. PI of competitive projects:** 16. **Participation in competitive projects:** >30.



Julio Escribano Martínez

Surname(s): **Escribano Martínez**
Name: **Julio**
DNI:
ORCID: **0000-0002-8919-8134**
ScopusID: **35518550700**
ResearcherID: **D-9742-2015**
Date of birth: **25/05/1962**
Gender: **Male**
Nationality: **Spain**
Country of birth: **Spain**
City of birth: **Albacete**
Land line phone: **(34) 967599200 - 2928**
Email: **julio.escribano@uclm.es**
Personal web page: **<https://www.uclm.es/centros-investigacion/idine/laboratorios/genetica-molecular-humana>**

Current professional situation

Employing entity: Universidad de Castilla-La Mancha **Type of entity:** University

Department: Genetics, Facultad de Medicina de Albacete

Professional category: Professor of Genetics

City employing entity: ALBACETE, Castile-La Mancha, Spain

Phone: (34) 967599200 - 2928

Start date: 01/04/2010

Dedication regime: Full time

Primary (UNESCO code): 241007 - Human genetics

Identify key words: Molecular mechanism of disease; Laboratoy animals; Biological sciences

Previous positions and activities

	Employing entity	Professional category	Start date
1	Universidad de Castilla-La Mancha	Profesor Titular de Universidades	01/02/2001
2	Universidad de Castilla-La Mancha	Profesor Asociado	07/03/1996
3	Universidad de Castilla-La Mancha	Contrato Reincorporacion doctores y tecnologos (MEC)	01/08/1994
4	Yale University Medical School, Department of Ophthalmology and Visual Science, USA	Investigador postdoctoral	01/08/1992
5	Yale University Medical School, Department of Ophthalmology and Visual Science, USA	Becario postdoctoral	01/08/1992
6	Hospital Universitario Ramón y Cajal	Becario predoctoral	01/01/1989



- 1** **Employing entity:** Universidad de Castilla-La Mancha **Type of entity:** University
Professional category: Profesor Titular de Universidades
Start-End date: 01/02/2001 - 31/03/2010
- 2** **Employing entity:** Universidad de Castilla-La Mancha **Type of entity:** University
Professional category: Profesor Asociado
Start-End date: 07/03/1996 - 26/02/2001
- 3** **Employing entity:** Universidad de Castilla-La Mancha **Type of entity:** University
Professional category: Contrato Reincorporacion doctores y tecnologos (MEC)
Start-End date: 01/08/1994 - 31/12/1995
- 4** **Employing entity:** Yale University Medical School, Department of Ophthalmology and Visual Science, USA
Professional category: Investigador postdoctoral
Start-End date: 01/08/1992 - 01/08/1994 **Duration:** 2 years
- 5** **Employing entity:** Yale University Medical School, Department of Ophthalmology and Visual Science, USA
Professional category: Becario postdoctoral
Start-End date: 01/08/1992 - 28/02/1993 **Duration:** 2 years
Type of contract: Grant-assisted student (pre or post-doctoral, others)
- 6** **Employing entity:** Hospital Universitario Ramón y Cajal **Type of entity:** Healthcare Institutions
Professional category: Becario predoctoral
Start-End date: 01/01/1989 - 30/06/1992



Education

University education

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

1 University degree: Higher degree

Name of qualification: Licenciado en Ciencias Biológicas Especialidad Biología Fundamental

Degree awarding entity: Universidad Complutense **Type of entity:** University de Madrid

Date of qualification: 06/1988

2 Name of qualification: Diplomado en Enfermería

Degree awarding entity: Universidad de Murcia

Date of qualification: 30/06/1983

Doctorates

Doctorate programme: Programa de doctorado en Bioquímica y Biología Molecular

Degree awarding entity: Universidad Complutense **Type of entity:** University de Madrid

Date of degree: 25/06/1992

Teaching experience

Experience supervising doctoral thesis and/or final year projects

1 Project title: Identificación de genes candidato en glaucoma congénito mediante secuenciación masiva de exomas: papel del gen GPATCH3 en glaucoma congénito y en desarrollo craneofacial

Entity: Universidad de Castilla-La Mancha **Type of entity:** University

Student: Jesús José Ferre Fernández

Obtained qualification: Sobresaliente cum laude

Date of reading: 20/09/2017

European doctorate: Yes

Date of recognition: 20/09/2017

Quality recognition: Yes

Date of award: 20/09/2017

2 Project title: Comportamiento cinético de los sistemas enzimáticos: aplicación a la activación de cascadas monocíclicas.

Entity: Universidad de Castilla-La Mancha **Type of entity:** University

City of entity: Albacete, Castile-La Mancha, Spain

Student: María Dolores Masiá Mondejar

Obtained qualification: Sobresaliente cum laude

Date of reading: 05/02/2016



3 Project title: Interacción de las proteínas matricelulares miocilina, hevin y SPARC. Expresión tisular, papel en la adhesión celular e implicaciones en glaucoma

Type of project: Doctoral thesis

Entity: Facultad de Medicina, UCLM

Type of entity: University

City of entity: Albacete, Castile-La Mancha, Spain

Student: Ana Fernández Navarro

Obtained qualification: sobresaliente cum laude

Date of reading: 03/12/2015

4 Project title: Identificación y análisis genético y funcional de variantes del gen FOXC1 en glaucoma congénito primario

Entity: Universidad de Castilla-La Mancha

Type of entity: University

Student: Cristina Medina Trillo

Obtained qualification: Sobresaliente cum laude por unanimidad

Date of reading: 14/03/2014

5 Project title: Procesamiento proteolítico de miocilina: caracterización, papel de las calpaínas e influencia sobre la regulación de genes de matriz extracelular. Implicaciones en glaucoma.

Entity: Universidad de Castilla-La Mancha

Type of entity: University

Student: Francisco Martínez Redondo

Obtained qualification: sobresaliente cum laude por unanimidad

Date of reading: 25/06/2012

6 Project title: Variaciones de la secuencia de los genes WDR36 y P53 en glaucoma primario de ángulo abierto: interacción génica y correlación genotipo-fenotipo

Entity: Universidad de Castilla La-Mancha

Student: Cristina Blanco-marchite

Obtained qualification: Sobresaliente cum laude

Date of reading: 01/07/2011

7 Project title: Variaciones de la Secuencia del Gen CYP1B1 en Glaucoma Primario de Ángulo Abierto: Mecanismos Genéticos de Patogenicidad y Relación Genotipo-Fenotipo. Universidad de Castilla La-Mancha. Facultad de Medicina

Entity: CASTILLA-LA MANCHA

Student: María Del Pilar López Garrido

Obtained qualification: Sobresaliente cum laude.

Date of reading: 01/07/2010

8 Project title: Análisis cinético de la acción simultánea de dos inhibidores diferentes sobre la activación de zimógenos

Entity: Castilla-La Mancha

Student: J. MasiÁ Pérez

Obtained qualification: Sobresaliente Cum laude por unanimidad

Date of reading: 18/12/2009

9 Project title: Secreción, procesamiento proteolítico e interacciones moleculares de miocilina. Implicaciones en la relación genotipo-fenotipo en glaucoma.

Entity: de Castilla La-Mancha

Student: Jose Daniel Aroca Aguilar

Obtained qualification: Sobresaliente cum laude (PREMIO EXTRAORDINARIO DE DOCTORADO).

Date of reading: 05/06/2009



10 Project title: Papel de los genes MIOCILINA y OPTINEURINA en pacientes del sureste de España con glaucoma primario de ángulo abierto
Entity: de Castilla-La Mancha
Student: Francisco Lopez Martinez
Obtained qualification: sobresaliente cum laude por unanimidad
Date of reading: 23/12/2008

11 Project title: ISOLATION, STRUCTURAL CHARACTERISATION AND PHARMACOLOGICAL PROPERTIES OF BIOACTIVE SAPONINS FROM CORMS OF CROCUS SATIVUS L. (DOCTORADO EUROPEO).
Entity: CASTILLA-LA MANCHA
Student: Angela Rubio Moraga
Obtained qualification: SOBRESALIENTE CUM LAUDE
Date of reading: 12/08/2004

12 Project title: POLIMORFISMOS GENÉTICOS DEL SISTEMA RENINA-ANGIOTENSINA Y DEL GEN DE LA ÓXIDO NÍTRICO SINTASA EN LA REGULACIÓN DE LA PRESIÓN ARTERIAL EN UNA POBLACIÓN ESPAÑOLA
Entity: AUTONOMA DE MADRID
Student: Angel Puras
Obtained qualification: SOBRESALIENTE CUM LAUDE
Date of reading: 22/04/2002

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:** Analysis of the genetic and molecular bases of congenital and juvenile glaucoma: diagnostic and therapeutic implications (Ref.: PI19/00208)
Entity where project took place: Facultad de medicina de Albacete **Type of entity:** University
City of entity: Albacete, Castile-La Mancha, Spain
Name principal investigator (PI, Co-PI....): Julio Escribano Martínez
Start-End date: 01/01/2020 - 31/12/2022
Total amount: 101.640 €
- 2 Name of the project:** OFTARED, red temática de investigación cooperativa en salud. prevención, detección precoz, tratamiento y rehabilitación de las patologías oculares
Entity where project took place: Facultad de Medicina de Albacete **Type of entity:** University
City of entity: Albacete, Castile-La Mancha, Spain
Name principal investigator (PI, Co-PI....): Julio Escribano Martínez
Nº of researchers: 4
Funding entity or bodies:
Instituto de Salud Carlos III **Type of entity:** Public Research Body
City funding entity: Majadahonda, Community of Madrid, Spain



Code according to the funding entity: RD16/0008/0019

Start-End date: 01/01/2017 - 31/12/2021

Total amount: 83.996 €

3 Name of the project: Establecimiento y caracterización de una línea de pez cebra knock-out para el gen cyp1b1: obtención de un modelo animal para el estudio del glaucoma congénito y para el desarrollo de nuevos tratamientos farmacológicos

Entity where project took place: Facultad de Medicina de Albacete **Type of entity:** University

City of entity: Albacete, Castile-La Mancha, Spain

Name principal investigator (PI, Co-PI....): Julio Escribano Martínez; Jose Daniel Aroca Aguilar

Nº of researchers: 5

Funding entity or bodies:

Consejería de Educación, JCCM

Code according to the funding entity: SBPLY/17/180501/000404

Start-End date: 01/03/2018 - 31/12/2020

Total amount: 155.000 €

4 Name of the project: Análisis funcional in vitro y en un modelo de pez cebra de genes candidato en glaucoma congénito primario. Identificación de nuevos genes mediante secuenciación masiva de exomas.

Entity where project took place: Facultad de Medicina, UCLM **Type of entity:** University

City of entity: Albacete, Castile-La Mancha, Spain

Name principal investigator (PI, Co-PI....): Julio Escribano Martínez

Nº of researchers: 4

Funding entity or bodies:

Instituto de Salud Carlos III

Type of entity: Public Research Body

City funding entity: Majadahonda, Community of Madrid, Spain

Code according to the funding entity: PI15/01193

Start-End date: 01/01/2016 - 31/12/2019

Total amount: 126.445 €

Dedication regime: Full time

5 Name of the project: Expresión del gen MYOC en tejidos no oculares y efecto de miocilina sobre la expresión de genes de matriz extracelular: implicaciones en la relación genotipo-fenotipo en glaucoma.

Entity where project took place: Universidad de Castilla-La Mancha **Type of entity:** University

City of entity: Albacete, Castile-La Mancha, Spain

Name principal investigator (PI, Co-PI....): Julio Escribano Martínez

Nº of researchers: 4

Code according to the funding entity: PEII-2014-002-P

Start-End date: 01/01/2014 - 01/11/2017

Total amount: 89.600 €

6 Name of the project: Mutaciones de los genes FOXC1, FOXC2 Y PITX2 en glaucoma congénito primario. Identificación de nuevos genes mediante secuenciación del exoma y estudio de relación genotipo-fenotipo

Geographical area: National

Name principal investigator (PI, Co-PI....): Julio Escribano Martinez

Funding entity or bodies:

Instituto de Salud Carlos III



Code according to the funding entity: PI11/00662

Start date: 01/01/2012

Duration: 3 years

Total amount: 115.830 €

- 7 Name of the project:** PAPEL DEL PROCESAMIENTO PROTEOLÍTICO DE MIOCILINA EN LAS INTERACCIONES PROTEÍNA-PROTEÍNA Y EN LAS UNIONES CELULARES: IMPLICACIONES EN LA RELACIÓN GENOTIPO-FENOTIPO EN GLAUCOMA.

Geographical area: National

Name principal investigator (PI, Co-PI....): Julio Escribano Martinez

Funding entity or bodies:

SECRETARÍA DE ESTADO DE UNIVERSIDADES E INVESTIGACIÓN

Start date: 01/01/2009

Duration: 3 years

Total amount: 96.800 €

- 8 Name of the project:** PATOLOGÍA OCULAR DEL ENVEJECIMIENTO, CALIDAD VISUAL Y CALIDAD DE VIDA

Geographical area: National

Name principal investigator (PI, Co-PI....): Julio Escribano Martinez

Funding entity or bodies:

INSTITUTO DE LA SALUD CARLOS III

Start date: 15/02/2008

Duration: 3 years

Total amount: 74.677 €

- 9 Name of the project:** ANÁLISIS DE MUTACIONES DEL GEN CYP1B1 EN PACIENTES ESPAÑOLES AFECTADOS POR GALUCOMA PRIMARIO DE ÁNGULO ABIERTO

Geographical area: National

Name principal investigator (PI, Co-PI....): Julio Escribano Martinez

Funding entity or bodies:

INST.SALUD CARLOS III

Start date: 04/07/2005

Duration: 3 years

Total amount: 20.000 €

- 10 Name of the project:** ANALISIS DEL PAPEL DE LA PROTEINA HUMANA MIOCILINA EN LA ADHESIÓN CELULAR Y CARACTERIZACIÓN DE SU PROCESAMIENTO ENDOPROTEOLITICO

Geographical area: Regional

Name principal investigator (PI, Co-PI....): Julio Escribano Martinez

Funding entity or bodies:

JUNTA DE COMUNIDADES DE CASTILLA-LA MANCHA

Start date: 22/06/2005

Duration: 3 years

Total amount: 72.800 €



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

1 Name of the project: PREMIO ALCON A LA EXCELENCIA INVESTIGADORA (01 MAR 2013).

Funding entity or bodies:

Alcon Cusí, S.A.

Type of entity: R&D Centre

City funding entity: El Masnou, Catalonia, Spain

Start date: 01/03/2013

Total amount: 4.000 €

2 Name of the project: REALIZACIÓN DE DIAGNÓSTICO GENÉTICO DE DIABETES TIPO MODY

Degree of contribution: Coordinator of total project, network or consortium

Participating entity/entities: FUNDACIÓN PARA LA DIABETES DE CASTILLA-LA MANCHA (FUCAMDI)

Funding entity or bodies:

FUNDACIÓN PARA LA DIABETES DE CASTILLA-LA MANCHA (FUCAMDI)

Start date: 01/01/2009

Duration: 3 years

Total amount: 150.725 €

3 Name of the project: DIAGNOSTICO GENETICO DE DIABETES TIPO MODY

Degree of contribution: Coordinator of total project, network or consortium

Participating entity/entities: FUNDACION DE CASTILLA-LA MANCHA PARA LA DIABETES

Funding entity or bodies:

FUNDACION DE CASTILLA-LA MANCHA PARA LA DIABETES

Start date: 01/09/2006

Duration: 2 years - 9 months

Total amount: 102.718,26 €

Results

Industrial and intellectual property

1 Title registered industrial property: USE OF THE PEDF FACTOR TO INDUCE CELL REGENERATION

Type of industrial property: Patent of invention Author's rights: Yes

Related rights: Yes

Inventors/authors/obtainers: Isabel Fariñas; Carmen Ramirez Castillejo; Celia Andreu-agulló; Sacramento Ferrón; P Sánchez; H Mira; Francisco Sanchez Sanchez; Jose Daniel Aroca Aguilar; Julio Escribano Martinez

Entity holder of rights: Universidad de Valencia/UCLM

Nº of application: 07735311.8

Country of inscription: Spain

Date of register: 16/02/2007

Conferral date: 16/11/2011

Nº of patent: EP 2 044 952 B1

Spanish patent: Yes

EU patent: Yes

2 Title registered industrial property: Procedimiento de producción y purificación del factor derivado del epitelio pigmentado de la retina en un sistema de levadura.

Type of industrial property: Patent of invention Author's rights: Yes



Related rights: Yes

Inventors/authors/obtainers: Francisco Sanchez Sanchez; Jose Daniel Aroca Aguilar; Carmen Ramirez Castillejo; Isabel Fariñas; Julio Escribano Martinez

Entity holder of rights: Universidad de Castilla la Mancha (90%). Universidad de Valencia (10%).

Nº of application: 200600385

Country of inscription: Spain

Date of register: 17/02/2006

Conferral date: 21/09/2006

Nº of patent: ES 2 330 173 B1

Spanish patent: Yes

3 **Title registered industrial property:** PEDF-R RECEPTOR AND USES

Inventors/authors/obtainers: Patricia Becerra; Luigi Notari; Jorge Laborda Fernandez; Julio Escribano Martinez

Entity holder of rights: National Institutes of Health, USA

Reference/registry code: WO2005014645-A2.

Nº of application: US Patent Application 20090162363

Country of inscription: United States of America

Date of register: 2004

4 **Title registered industrial property:** Método de aislamiento de sustancias colorantes y saborizantes del azafrán especia: crocinas y picrocrocina

Type of industrial property: Patent of invention

Author's rights: Yes

Related rights: Yes

Inventors/authors/obtainers: Julio Escribano Martinez; Gonzalo Luis Alonso Diaz-marta; Maria Rosario Salinas Fernandez; Jose Antonio Fernandez Perez

Entity holder of rights: UNIVERSIDAD DE CASTILLA-LA MANCHA

Reference/registry code: P9700664

Nº of application: 009700664

Country of inscription: Spain

Date of register: 15/04/1997

Conferral date: 30/11/1999

Nº of patent: ES 2 130 066 B1

Spanish patent: Yes

EU patent: No

International non-EU patent: No

Operating aut.region/region: Spain



Scientific and technological activities

Scientific production

Publications, scientific and technical documents

- 1** García-García GP; Martínez-Rubio M; Moya-Moya MA; Pérez-Santonja JJ; Escribano J. Identification of novel CYP4V2 genotypes associated with Bietti crystalline dystrophy and atypical anterior segment phenotypes in Spanish patients. *Acta ophthalmologica*. 2018. ISSN 1755-375X

DOI: 10.1111/aos.13768

PMID: 29691984

Type of production: Scientific paper

Position of signature: 5

Total no. authors: 5

Corresponding author: Yes

Impact source: SCOPUS

Category: Ophthalmology

Impact index in year of publication: 3.157

Journal in the top 25%: Yes

Position of publication: 12

No. of journals in the cat.: 59

Relevant publication: Yes

- 2** Jesus-Jose Ferre-Fernandez; Jose-Daniel Aroca-Aguilar; Cristina Medina-Trillo; Juan-Manuel Bonet-Fernandez; Carmen-Dora Mendez-Hernandez; Laura Morales-Fernandez; Marta Corton; Maria-Jose Cabanero-Valera; Marta Gut; Raul Tonda; Carmen Ayuso; Miguel Coca-Prados; Julian Garcia-Feijoo; Julio Escribano. Whole-Exome Sequencing of Congenital Glaucoma Patients Reveals Hypermorphic Variants in GPATCH3, a New Gene Involved in Ocular and Craniofacial Development. *Scientific Reports*. 7, 2017.

DOI: 10.1038/srep46175;28397860

Type of production: Scientific paper

Format: Journal

Position of signature: 14

Corresponding author: Yes

Total no. authors: 14

Category: Multidisciplinary

Impact source: SCOPUS

Journal in the top 25%: Yes

Impact index in year of publication: 4.36

No. of journals in the cat.: 87

Position of publication: 5

Relevant publication: Yes

- 3** Cristina Medina-Trillo; Jesus-Jose Ferre-Fernandez; Jose-Daniel Aroca-Aguilar; Juan-Manuel Bonet-Fernandez; Julio Escribano. Functional characterization of eight rare missense CYP1B1 variants involved in congenital glaucoma and their association with null genotypes. *Acta Ophthalmologica*. 94 - 7, pp. E555 - E560. 2016.

Type of production: Scientific paper

Format: Journal

Position of signature: 5

Corresponding author: Yes

Total no. authors: 5

Category: Ophthalmology

Impact source: SCOPUS

Journal in the top 25%: Yes

Impact index in year of publication: 3.157

No. of journals in the cat.: 59

Position of publication: 12

Relevant publication: Yes



- 4** Luis Alberto Marín Rubio; José-Daniel Aroca Aguilar; Mar Luis Hidalgo; Julio Escribano; Jesús Ontañón. RNA and protein expression analysis of HLA-DQB1*03:01:01:21Q allele: A null allele renamed as HLA-DQB1*03:01:01:21N. HLA. 99 - 3, pp. 160 - 166. 01/03/2022. Available on-line at: <[10.1111/tan.14537](https://doi.org/10.1111/tan.14537)>.

Type of production: Scientific paper

Format: Journal

- 5** Null cyp1b1 Activity in Zebrafish Leads to Variable Craniofacial Defects Associated with Altered Expression of Extracellular Matrix and Lipid Metabolism Genes. 16/06/2021.

Type of production: Scientific paper

Format: Journal

- 6** Knockout of myoc Provides Evidence for the Role of Myocilin in Zebrafish Sex Determination Associated with Wnt Signalling Downregulation. 30/01/2021.

Type of production: Scientific paper

Format: Journal

- 7** Juan-Manuel Bonet-Fernández; José-Daniel Aroca-Aguilar; Marta Cortón; Ana-Isabel Ramírez; Susana Alexandre-Moreno; María-Teresa García-Antón; Juan-José Salazar; Jesús-José Ferre-Fernández; Raquel Atienzar-Aroca; Cristina Villaverde; Ionut Iancu; Alejandra Tamayo; Carmen-Dora Méndez-Hernández; Laura Morales-Fernández; Blanca Rojas; Carmen Ayuso; Miguel Coca-Prados; José-María Martínez-de-la-Casa; Julián García-Feijoo; Julio Escribano. CPAMD8 loss-of-function underlies non-dominant congenital glaucoma with variable anterior segment dysgenesis and abnormal extracellular matrix. Human Genetics. 139 - 10, pp. 1209 - 1231. 2020.

DOI: [10.1007/s00439-020-02164-0](https://doi.org/10.1007/s00439-020-02164-0)

Type of production: Scientific paper

Format: Journal

Position of signature: 20

Total no. authors: 20

Corresponding author: Yes

- 8** Laura Morales-Fernández; José María Martínez-de-la-Casa; Blanca Benito-Pascual; Federico Saénz-Francés; Enrique Santos-Bueso; Pedro Arriola-Villalobos; Julio Escribano-Martínez; Julián García-Feijoo. Cataract extraction in patients with primary congenital glaucoma. Eur J Ophthalmol. 30 - 3, pp. 525 - 532. 2020.

DOI: [10.1177/1120672119841190](https://doi.org/10.1177/1120672119841190)

Type of production: Scientific paper

Format: Journal

Position of signature: 7

Total no. authors: 8

Corresponding author: No

- 9** Susana Alexandre-Moreno; Samuel Morales-Cámaras; Juan-Manuel Bonet-Fernández; Raquel Atienzar-Aroca; José-Daniel Aroca-Aguilar; Jesús-José Ferre-Fernández; Carmen-Dora Méndez; Laura Morales; Laura Fernández-Sánchez; Nicolas Cuenca; Miguel Coca-Prados; José-María Martínez-de-la-Casa; Julián García-Feijoo; Julio Escribano. Role of GUCA1C in Primary Congenital Glaucoma and in the Retina: Functional Evaluation in Zebrafish. Genes (Basel). 11 - 5, pp. 550. MDPI, 2020.

DOI: doi: [10.3390/genes11050550](https://doi.org/10.3390/genes11050550).

Type of production: Scientific paper

Format: Journal

Position of signature: 14

Total no. authors: 14

Corresponding author: Yes

- 10** Cristina Medina Trillo; José Daniel Aroca Aguilar; Jesús Ferre Fernández; Susana Alexandre Moreno; Laura Morales Fernández; Carmen Mendez Hernández; Julián García Feijoo; Julio Escribano. Role of FOXC2 and PITX2 rare variants associated with mild functional alterations as modifier factors in congenital glaucoma. Plos One. 14 - 1, pp. e0211029. 2019.

Type of production: Scientific paper

Position of signature: 8

Total no. authors: 8

Corresponding author: Yes



11 Ezequiel Campos-Mollo; Yago Varela-Conde; Pedro Arriola-Villalobos; Rubén Cabrera-Beyrouti; José-Manuel Benitez-del-Castillo; Miguel Maldonado; Julio Escribano. TGFBI p.(L558P) variant is associated with autosomal dominant lattice corneal dystrophy type IV in a large cohort of Spanish patients. Clinical and Experimental Ophthalmology. 2019.

Type of production: Scientific paper

Position of signature: 7

Total no. authors: 7

Corresponding author: Yes

12 Aroca-Aguilar JD; Fernández-Navarro A; Ontañón J; Coca-Prados M; Escribano J. Identification of myocilin as a blood plasma protein and analysis of its role in leukocyte adhesion to endothelial cell monolayers. Plos One. 13, pp. e0209364. 2018.

DOI: 10.1371/journal.pone.0209364

PMID: 30557320

Type of production: Scientific paper

Position of signature: 5

Total no. authors: 5

Corresponding author: Yes

Impact source: ISI

Impact index in year of publication:

Category: Multidisciplinary

Journal in the top 25%: Yes

13 Maria T. Garcia-Anton; Juan J. Salazar; Rosa de Hoz; Blanca Rojas; Ana I. Ramirez; Alberto Trivino; Jose-Daniel Aroca-Aguilar; Julian Garcia-Feijoo; Julio Escribano; Jose M. Ramirez. Goniodygenesis variability and activity of CYP1B1 genotypes in primary congenital glaucoma. Plos One. 12 - 4, pp. e0176386. 2017.

DOI: 10.1371/journal.pone.0176386

PMID: 28448622

Type of production: Scientific paper

Format: Journal

Impact source: ISI

Category: Multidisciplinary

Impact index in year of publication:

Journal in the top 25%: Yes

Source of citations: WOS

Citations: 4

14 Montserrat Garcia; Lydia Alvarez; Angela Fernandez; Hector Gonzalez-Iglesias; Julio Escribano; Beatriz Fernandez-Vega; Eva Villota; Luis Fernandez-Vega Cueto; Alvaro Fernandez-Vega; Miguel Coca-Prados. Metallothionein polymorphisms in a Northern Spanish population with neovascular and dry forms of age-related macular degeneration. Ophthalmic genetics. 38 - 5, pp. 451 - 458. 2017.

Type of production: Scientific paper

Format: Journal

15 Prieto-Martin, Ana I.; Daniel Aroca-Aguilar, J.; Sanchez-Sanchez, Francisco; Manoz, Luis J.; Lopez, Dolores E.; Escribano, Julio; de Cabo, Carlos. Molecular and neurochemical substrates of the audiogenic seizure strains: The GASH:Sal model. EPILEPSY & BEHAVIOR. 71, pp. 218 - 225. 2017. ISSN 1525-5050

DOI: 10.1016/j.yebeh.2015.05.025

PMID: 26071997

Type of production: Scientific paper

Source of citations: WOS

Citations: 3

16 Maria D. Masia; Manuela Garcia-Moreno; Francisco Garcia-Sevilla; Julio Escribano; Milagros Molina-Alarcon; Maria L. Amo-Saus; Carmen V. Ortiz-Ruiz; Ramon Varon. A novel transient phase kinetic analysis of the fractional modification of monocyclic enzyme cascades. Journal of Mathematical Chemistry. 54 - 10, pp. 1952 - 1972. 2016.

Type of production: Scientific paper

Format: Journal



- 17** Cristina Medina-Trillo; Jose-Daniel Aroca-Aguilar; Carmen-Dora Mendez-Hernandez; Laura Morales; Maite Garcia-Anton; Julian Garcia-Feijoo; Julio Escribano. Rare FOXC1 variants in congenital glaucoma: identification of translation regulatory sequences. European Journal of Human Genetics. 24 - 5, pp. 672 - 680. 2016.
- Type of production:** Scientific paper **Format:** Journal
- Position of signature:** 7 **Corresponding author:** Yes
- Total no. authors:** 7 **Category:** Genetics
- Impact source:** SCOPUS **Journal in the top 25%:** Yes
- Impact index in year of publication:** 3.05 **No. of journals in the cat.:** 91
- Position of publication:** 30
- 18** Montserrat Garcia; Lydia Alvarez; Alicja M. Nogacka; Hector Gonzalez-Iglesias; Julio Escribano; Beatriz Fernandez-Vega; Alvaro Fernandez-Vega; Luis Fernandez-Vega; Miguel Coca-Prados. CFH polymorphisms in a Northern Spanish population with neovascular and dry forms of age-related macular degeneration. Acta Ophthalmologica. 93 - 8, pp. E658 - E666. 2015.
- Type of production:** Scientific paper **Format:** Journal
- Impact source:** ISI **Category:** Ophthalmology
- Impact index in year of publication:** **Journal in the top 25%:** Yes
- 19** Laura Morales-Fernandez; Jose M. Martinez-de-la-Casa; Javier Garcia-Bella; Carmen Mendez; Federico Saenz-Frances; Maite Garcia; Julio Escribano; Julian Garcia-Feijoo. Clinical Variability of Primary Congenital Glaucoma in a Spanish Family With Cyp1b1 Gene Mutations. Journal of Glaucoma. 24 - 8, pp. 630 - 634. 2015.
- Type of production:** Scientific paper **Format:** Journal
- 20** Cristina Medina-Trillo; Francisco Sanchez-Sanchez; Jose-Daniel Aroca-Aguilar; Jesus-Jose Ferre-Fernandez; Laura Morales; Carmen-Dora Mendez-Hernandez; Fiona Blanco-Kelly; Carmen Ayuso; Julian Garcia-Feijoo; Julio Escribano. Hypo-and Hypermorphic FOXC1 Mutations in Dominant Glaucoma: Transactivation and Phenotypic Variability. Plos One. 10 - 3, 2015.
- Type of production:** Scientific paper **Format:** Journal
- Position of signature:** 10 **Corresponding author:** Yes
- Total no. authors:** 10 **Category:** Biochemistry, Genetics and Molecular Biology (miscellaneous)
- Impact source:** SCOPUS **Journal in the top 25%:** Yes
- Impact index in year of publication:** 3.01 **No. of journals in the cat.:** 186
- Position of publication:** 42
- 21** Lydia Alvarez; Montserrat Garcia; Hector Gonzalez-Iglesias; Julio Escribano; Pedro P. Rodriguez-Calvo; Luis Fernandez-Vega; Miguel Coca-Prados. LOXL1 gene variants and their association with pseudoexfoliation glaucoma (XFG) in Spanish patients. BMC Medical Genetics. 16, 2015.
- Type of production:** Scientific paper **Format:** Journal
- Source of citations:** WOS **Citations:** 6
- 22** Cristina Medina-Trillo; Jose-Daniel Aroca-Aguilar; Jesus-Jose Ferre-Fernandez; Carmen-Dora Mendez-Hernandez; Laura Morales; Julian Garcia-Feijoo; Julio Escribano. The Role of hsa-miR-548I Dysregulation as a Putative Modifier Factor for Glaucoma-Associated FOXC1 Mutations. MicroRNA (Shariqah, United Arab Emirates). 4 - 1, pp. 50 - 6. 2015.
- Type of production:** Scientific paper **Format:** Journal
- Position of signature:** 7 **Corresponding author:** Yes
- Total no. authors:** 7



23 Gerardo-Pedro García-García; M A Martínez-Rubio; MA Moya-Moya; J J Pérez-Santonja. Current perspectives in Bietti crystalline dystrophy. *Clin Ophthalmol.* 13, pp. 1379 - 1399. 2019.

DOI: 10.2147/OPTH.S185744

Type of production: Review

Position of signature: 5

Total no. authors: 5

Corresponding author: Yes

24 Montserrat Garcia; Lydia Alvarez; Alicia Maria Nogacka; Hector Gonzalez-Iglesias; Julio Escribano; Beatriz Fernandez-Vega; Alvaro Fernandez-Vega; Luis Fernandez-Vega; Miguel Coca-Prados. Complement factor H polymorphisms associated with exudative or dry AMD in the Spanish population. *Investigative Ophthalmology & Visual Science.* 56 - 7, 2015.

Type of production: Congress abstract

Format: Journal

R&D management and participation in scientific committees

Scientific, technical and/or assessment committees

1 Committee title: Presidente de la Comisión de Control de Calidad y Evaluación de Oftared

Affiliation entity: Oftared, Red Temática de Investigación en Oftalmología (ISCIII) **Type of entity:** Public Research Body

City affiliation entity: Spain

Start-End date: 2012 - 2018

2 Committee title: Vocal del Comité de Ética en Investigación con Organismos Modificados, Agentes Biológicos y Bioseguridad (CIOMABB)

Affiliation entity: Vicerrectorado de Investigación, UCLM **Type of entity:** University

City affiliation entity: Spain

Start-End date: 01/01/2005 - 2012

3 Committee title: VOCAL DE LA COMISION DE INVESTIGACIÓN DE LA GERENCIA DE ATENCIÓN INTEGRADA DE ALBACETE

Affiliation entity: COMPLEJO HOSPITALARIO UNIVERSITARIO DE ALBACETE **Type of entity:** Administrative Body of the National Health System

City affiliation entity: Albacete, Spain

Start-End date: 01/01/2003 - 2010

4 Committee title: Vocal de la Junta Directiva de la SEG

Primary (UNESCO code): 240900 - Genetics

Affiliation entity: Sociedad Española de Genética **Type of entity:** Associations and Groups

City affiliation entity: Spain

Start-End date: 2002 - 2006

5 Committee title: Equipo decanal de la Facultad de Medicina de Albacete, Vicedecano.

Affiliation entity: Facultad de Medicina de Albacete, UCLM

City affiliation entity: Albacete,

Start-End date: 2001 - 2004



- 6 Committee title:** Comisión de Acreditacion A5 (Biología Celular y Molecular), ANECA
Primary (UNESCO code): 240700 - Cell biology; 240900 - Genetics; 241500 - Molecular biology
Start date: 01/10/2020

Other achievements

Stays in public or private R&D centres

- 1 Entity:** Yale University, Dept. Ophthalmology and Vis Sci **Type of entity:** University

City of entity: New Haven, United States of America

Start-End date: 01/08/1992 - 31/07/1994

Duration: 2 years

Goals of the stay: Post-doctoral

- 2 Entity:** YALE UNIVERSITY MEDICAL SCHOOL

City of entity: NEW HAVEN, Unknown

Start date: 29/06/2001

Duration: 1 month

Goals of the stay: Guest

- 3 Entity:** Odense University

Type of entity: University Department

Faculty, institute or centre: Department of Biochemistry and Molecular Biology

City of entity: Odense, Denmark

Start date: 1991

Duration: 15 days

Goals of the stay: Doctorate

Provable tasks: Análisis por espectrometría de masas de la proteína humana HC

- 4 Entity:** Lund University

Type of entity: University

Faculty, institute or centre: Medical School

City of entity: Malmo, Sweden

Start date: 01/10/1989

Duration: 1 month

Goals of the stay: Doctorate

Provable tasks: Analisis mediante tecnicas inmunoquimicas de la proteína humana HC

Periods of research activity

- 1 Nº of recognized periods:** 1

Certifying entity: CNEAI

Date of recognition: 01/01/2020

- 2 Nº of recognized periods:** 1

Certifying entity: CNAEI

Date of recognition: 01/01/2019

- 3 Nº of recognized periods:** 1

Certifying entity: CNEAI

City certifying entity: Spain

Date of recognition: 09/06/2014



4 Nº of recognized periods: 1

Geographical area: National

Certifying entity: CNEAI

City certifying entity: Spain

Date of recognition: 01/01/2008

5 Nº of recognized periods: 1

Geographical area: National

Certifying entity: CNEAI

City certifying entity: Spain

Date of recognition: 01/01/2002

6 Nº of recognized periods: 1

Geographical area: National

Certifying entity: CNEAI

City certifying entity: Spain

Date of recognition: 01/01/2002