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Dra. Esther Pomares

Coordinator of the Genetics Department, Institute of Ocular Microsurgery.

Master's Degree in Experimental Biology, Diploma in Advanced Studies on the Genetics Programme and Doctor of Genetics, with honours, from the University of Barcelona.

Patron of the IMO Foundation since 2014.

License Number: 21.659

Curriculum

Professional Profile

Coordinator in the Genetics Department at the Ocular Microsurgery Institute. Master's Degree in Experimental Biology, Diploma in Advanced Studies on the Genetics Programme and Doctor of Genetics, with honours, from the University of Barcelona. Patron of the IMO Foundation since 2014.

Her specialty is the genetic diagnosis and study of the molecular bases of eye diseases.

She received a Training of Research Staff (FPI) grant and a CIBERER pre-doctoral contract at the UB. Later, she was working on the CIBERER post-doctoral contract, also at the University of Barcelona..

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Training

Master's Degree in Experimental Biologyfrom the University of Barcelona, 2003.

Diploma of Advanced Studies in the Genetics Programme from the University of Barcelona, 2005.

Doctor in Genetics. Doctorate with honours, from the University of Barcelona, 2009.

Collaboration scholarship at the Germans Trias i Pujol Hospital, 2003.

FPI fellowship at the University of Barcelona, 2004-2007.

Pre-doctoral CIBERER contract at the University of Barcelona, 2007-2009.

Post-doctoral CIBERER contract at the University of Barcelona, 2009-2012.

Memberships

- European Society of Human Genetics

Publications

Riera, M; Patel, A; Corcóstegui, B; Sparrow, J; **Pomares, E**; Corneo, B. *Establishment and characterization of an iPSC line (FRIMOi001-A) derived from a retinitis pigmentosa patient carrying PDE6A mutations.*
Stem Cell Research https://doi.org/10.1016/j.scr.2019.101385

Riera, M; Patel, A; Corcóstegui, B; Sparrow, J; **Pomares, E**; Corneo, B. *Generation of an induced pluripotent stem cell line (FRIMOi002-A) from a retinitis pigmentosa patient carrying compound heterozygous mutations in USH2A gene .*
Stem Cell Research https://doi.org/10.1016/j.scr.2019.101386

Riera, M; Patel, A; Burés-Jelstrup, A; Corcóstegui, B; Sparrow, J; **Pomares, E**; Corneo, B. *Generation of iPS cell lines derived from two Stargardt patients carrying ABCA4 compound heterozygous mutations* Stem Cell Research

Riera, M; Wert, A; Nieto, I; **Pomares, E**. *Panel-based whole exome sequencing identifies novel mutations in microphthalmia and anophthalmia patients showing complex Mendelian inheritance patterns. Mol Genet Genomic Med Nov;5(6):709-719, 2017.*

Riera, M; Navarro, R; Ruiz-Nogales, S; Méndez, P; Burés-Jelstrup, A; Corcóstegui, B; **Pomares, E**. *Whole exome sequencing using Ion Proton system enables reliable genetic diagnosis of inherited retinal dystrophies.* Sci Rep. Feb 9; 7:42078, 2017.

De Castro-Miró, M.; **Pomares, E.**; Lorés, L.; Tonda, R.; Dopazo, J.; Marfany, G. & González-Duarte, R. *Combined genetic and high-throughput strategies for molecular diagnosis of inherited retinal dystrophies.* PLoS One 9(2):e88410, 2014.

Perrault I, Estrada-Cuzcano A, Lopez I, Kohl S, Li S, Testa F, Zekveld-Vroon R, Wang X, **Pomares E**, Andorf J, Aboussair N, Banfi S, Delphin N, Den Hollander Ai, Edelson C, Florijn R, Jean-Pierre M, Leowski C, Megarbane A, Villanueva C, Flores B, Munnich A, Ren H, Zobor D, Bergen A, Chen R, Cremers Fp, Gonzalez-Duarte R, Koenekoop RK, Simonelli F, Stone E, Wissinger B, Zhang Q, Kaplan J, Rozet JM. *Union makes strength: a worldwide collaborative genetic and clinical study to provide a comprehensive survey of RD3 mutations and delineate the associated phenotype*. PLoS One 8(1):e51622, 2013.

Pomares, E.; Burés-Jelstrup, A.; Ruiz-Nogales, S.; Corcóstegui, B.; González-Duarte, R. & Navarro, R. *Nonsense-mediated decay as the molecular cause for autosomal recessive bestrophinopathy in two unrelated families.* Investigative Ophthalmology & Visual Science 53(1):532-7, 2012.

Pomares, E.; Marfany, G. & Gonzàlez-Duarte, R. *High-throughput approaches for the genetic diagnosis of retinal dystrophies.* Advances in Experimental Medicine and Biology 723:329-35, 2012.

Garanto, A.; Riera, M.; **Pomares, E.**; Permanyer, J.; De Castro-Miró, M.; Sava, F.; Abril J.F.; Marfany, G. & Gonzàlez-Duarte, R. *High transcriptional complexity of the retinitis pigmentosa CERKL gene in human and mouse*. Investigative Ophthalmology & Visual Science 52(8):5202-14, 2011.

Permanyer, J.; Navarro, R.; Friedman, J.; **Pomares, E**.; Castro-Navarro, J.; Marfany, G.; Swaroop, A. & Gonzàlez-Duarte, R. *Autosomal recessive Retinitis Pigmentosa with early macular affectation caused by premature truncation in PROM1.* Investigative Ophthalmology & Visual Science 51(5):2656-63, 2010.

Pomares, E.; Riera, M.; Permanyer, J.; Mendez, P.; Castro-Navarro, J.; Andrés-Gutiérrez, A.; Marfany, G. & Gonzàlez-Duarte, R. *Comprehensive SNP-chip for Retinitis Pigmentosa-Leber Congenital Amaurosis diagnosis: new mutations and detection of mutational founder effects*. European Journal of Human Genetics 18 (1): 118-24, 2010.

Pomares, E.; Riera, M.; Castro-Navarro, J.; Andrés-Gutiérrez, A.; Marfany, G. & Gonzàlez-Duarte, R. *An intronic single point mutation in RP2 causes semidominant X-linked Retinitis Pigmentosa.* Investigative Ophthalmology & Visual Science 50 (11): 5107-14, 2009.

Brea-Fernández, A. J.; **Pomares, E**.; Brión, M. J.; Marfany, G.; Blanco, M. J.; Sánchez-Salorio, M.; Gonzàlez-Duarte, R. & Carracedo, A. *Novel splice donor site mutation in MERTK gene associated with retinitis with retinitis pigmentosa.* British Journal of Ophthalmology 92: 1419-1423, 2008.

Pomares, E. *Estrategias automatizadas para el diagnóstico genético de la Retinosis Pigmentaria.*Visión 32: 28-29, 2008.

Pomares, E.; Marfany, G.; Brión, Ma J.; Carracedo, A. & Gonzàlez-Duarte, R. *Novel high-throughput SNP genotyping cosegregation analysis for genetic diagnosis of autosomal recessive Retinitis Pigmentosa and Leber Congenital Amaurosis.* Human Mutation 28 (5): 511-16, 2007.

Cervantes, S.; Saura, C.A.; **Pomares, E**.; Gonzàlez-Duarte, R. & Marfany, G. *Functional implications of the presenilin dimerization: reconstitution of gamma-secretase activity by assembly of a catalytic site at the dimer interface of two catalytically inactive presenilins.* Journal of Biological Chemistry 279 (35): 36519-29, 2004.

Clinical guide to Low Vision:

Gómez, A; Castillón, L; **Pomares, E**; Castañé, E; Sánchez-Ramo, C; Aguirre, M; Yagüe, F; Alcocer, A; Marco, E. "Tengo Baja Visión ¿Qué puedo hacer?" Spanish Society of Low Vision Specialists, SEEBV. 2014

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Conferences and courses

Pomares E. (2016) Oral communication. Genetic heterogeneity in retinal dystrophies: challenges in molecular diagnosis. AARPC 2016. Barcelona

Pomares E. (2016) Oral communication. Genetic study of diseases in children. When to request it and what it provides. 1st Adult Strabismus and Children with Low Vision Course, IMO Foundation. Barcelona

Pomares E. (2016) Poster. Clinical and molecular characterization of the ABCA4-associated dystrophies: novel genotype-phenotype correlations. M. Riera, A. Burés-Jelstrup, P. Méndez, J. C. D'Antin, B. Corcóstegui, R. Navarro, E. Pomares. European Society of Human Genetics 2016. Barcelona

Pomares E. (2015) Oral communication. Genetics of retinal dystrophies. Rare diseases, IMO Foundation. Barcelona

Pomares E. (2015) Oral communication. Gene therapy vs cell therapy. 3rd Low Vision and Visual Rehabilitation Sessions, Spanish Society of Low Vision Specialists. Madrid

Pomares E. (2015) Speaker. "Value of genetic analysis." 1st Retina Specialists Meeting. Allergan. Oxford

Pomares E. (2014) Speaker. "Genetics in retinal dystrophies." Retina project multidisciplinary sessions. Open your Eyes Foundation and the Spanish Society of Low Vision Specialists (SEEBV). Valencia

Pomares E. (2014) Speaker. "Genética de las patologías oculares".Course for the assessment of paediatric and/or strabic patients. Ocular Microsurgery Institute, IMO. Barcelona.

Pomares E. (2013) Speaker. "L'estudi genètic del glaucoma familiar". Semana Mundial del Glaucoma 2013. Fundación IMO. Barcelona.

Pomares E. (2013) Speaker. "Genética en Distrofias Retinianas". 2as Jornadas de Baja Visión y Rehabilitación Visual. Spanish Society of Low Vision Specialists, SEEBV. Barcelona.

Pomares E. (2012) Speaker. "The RD-chip as a diagnosis tool for familial retina dystrophies". CIBERER Neurosensorial Pathology Meeting. Madrid.

Pomares E. (2012) Poster. An optimised high-throughput SNP chip for the genetic diagnosis of Retinal Dystrophies. ISER 2012. International Society for Eye Research. Berlin, Germany

Pomares E. (2011) Speaker. "Chip optimised for the high-throughput diagnosis of the most common hereditary retina dystrophies". CIBERER Neurosensorial Pathology Meeting. Madrid.

Pomares E. (2011) Speaker. "Chip optimised for the high-throughput diagnosis of the most common hereditary retina dystrophies". 5th Annual Meeting of the CIBERER. Madrid

Pomares E. (2011) Poster. "High transcriptional complexity of the retinitis pigmentosa CERKL gene in human and mouse". ARVO 2011. Annual Meeting. Florida. USA

Pomares E. (2010) Attendee. "High transcriptional complexity of the retinitis pigmentosa CERKL gene in human retina". 4th Annual Meeting of the CIBERER. Madrid.

Pomares E. (2009) Attendee. "Análisis genómico a gran escala en familias afectas de retinosis pigmentaria (RP)". 3rd Annual Meeting of the CIBERER. El Escorial.

Pomares E. (2008) Speaker. "Retos en el diagnóstico de la Retinosis Pigmentaria". 2nd Annual Meeting of the CIBERER, Valencia.

Pomares E. (2008) Poster. "Genetic diagnosis of autosomal dominant and recessive Retinitis Pigmentosa using SNP high-throughput genotyping". ESHG 2008. European Society of Human Genetics. Barcelona.

Pomares E. (2008) Poster. "Rapid and efficient SNP genotyping for the genetic diagnosis of autosomal dominant and recessive Retinitis Pigmentosa". ARVO 2008. Annual Meeting. Fort Lauderdale. USA.

Pomares E. (2007) Poster. "Construcción de un modelo murino knockout condicional para el gen ceramida kinasa-like (CERKL)". 1st Annual Meeting of the CIBERER. Barcelona.

Pomares E. (2007) Speaker. "Nova estratègia de diagnòstic indirecte per la Retinitis Pigmentària". XV JORNADES DE BIOLOGIA MOLECULAR de la Societat Catalana de Biologia. Barcelona.

Pomares E. (2007) Poster. "Nueva estrategia de diagnóstico para la retinosis pigmentaria". XXXVI Congress of the Spanish Genetics Society. León.

Pomares E. (2003) Poster. ”Homodimerization of presenilins: Implications for the presinilinase and gamma-secretase activities”. 6th International Conference on Alzheimer’s and Parkinson’s Diseases (AD/PD). Sevilla

Research

Principal researcher. iPS cells as a tool of study and model for implementation of the CRISPR/Cas system in retinal dystrophies

Principal researcher. Search and identification of mutations in non-coding regions of the ABCA4 gene. Population study on a cohort of Spanish patients

Principal researcher. Identification of new retinal dystrophy genes and characterisation of genotype/phenotype correlations.

Collaborating researcher. Mass sequencing of exomes for the identification of new genes and variants responsible for retinal dystrophies.

Collaborating researcher. BIOIMAGE. Phase IV clinical trial to assess the genetic variants of the VEGF pathway as efficacy biomarkers of the treatment with aflibercept in patients with neovascular age-related macular degeneration (AMD). (IMO- AFLI-2013-01)

Teaching

She took part in the subject “Human genetic diseases” on the Master’s course in Developmental biology and Genetics at the University of Barcelona in 2009-2010 and 2010-2011.

Laboratory practice teacher in Human Genetics for the Biology Degree course at the University of Barcelona, from 2004 to 2008.