CURRICULUM VITAE

Part A. PERSONAL INFORMATION

CV date	2025

First and Family name	**Gemma Marfany Nadal**		
Social Security, Passport, ID number		Age	
Researcher numbers	Researcher ID		
	Scopus Author ID		
	ORCID code		

A.1. Current position

Name of University/Institution	Universitat de Barcelona		
Department	Dept. de Genética, Microbiología y Estadística		
Address and Country			
Phone number			
Current position	Full Professor of Genetics	From	2020
Espec. cód. UNESCO	2409 / 2410.07 / 2409.02 / 2415		
Palabras clave	Genetics/Human molecular genetics/Genetic engineering/ Molecular Biology		

A.2. Previous positions (research activity interuptions, indicate total months)

Posoition	University	Year
Pre-doctoral fellow	Universitat de Barcelona	1987-90
Assistant Professor LRU	Universitat de Barcelona	1991-92
Post-doctoral fellow, Dept. Biochemistry	University of Oxford	1992-94
Associate Professor of Genetics	Universitat de Barcelona	1995-2020

A.3. Education

BSc, PhD, post-doctoral	University	Year
Biology, BSc	Universitat de Barcelona	1987
Biology, PhD	Universitat de Barcelona	1991

Part B. CV SUMMARY *(max. 5000 characters, including spaces)*

Gemma Marfany obtained the Biology BSc at University of Barcelona (1986). Her PhD (1991) was focused on the *Adh* locus molecular evolution in several *Drosophila* species, publishing 6 articles and obtaining the maximum qualification of Excellent cum laude, with extraordinary PhD prize. She was a post-doc at the Dept of Biochemistry (University of Oxford, UK, 199294) working on the molecular genetics of transposable genetic elements, gaining experience in protein techniques and mammalian cell culture. She then started long-standing and fruitful collaborations with other scientists. In 1995, she was appointed as Associate Professor in Genetics (UB) and joined the Human Molecular Genetics research group at the Dept of Genetics, where she started new lines of research in human genetics. In 2020, she was

appointed as Full Professor in Genetics. Her teaching involves a large number of subjects of theory and practical courses, actively contributing to the Master of Genetics and Developmental Biology (now Master in Genetics and Genomics), being the coordinator in 2010-11 (awarded as Master of Excellence by Fundació La Pedrera).

Since being a PI, her research is focused on human molecular genetics, initially, on identifying novel human genes on chromosome 21 involved in the Down syndrome, as well as research in Alzheimer's disease and diabetes. For the **last 20 years, her research is centered in inherited retinal dystrophies**, focusing on both genetic-molecular diagnosis and identification of new genes, and functional gene analysis in cell cultures and *in vivo* by generating *knockout* (mouse) and *knockdown* (zebrafish) animal models, using cre/loxP and CRISPR techniques. Currently, she is also interested in retinal organoids as a disease model. Besides generating a large number of articles highly cited in the molecular genetics of vision field, she submitted a patent application for the design of a high-throughput chip for retinal dystrophy genetic diagnosis (2006-2009).

Gemma Marfany has been granted **17 projects in competitive calls as a PI**, and participated in 40. She is author of **more than 125 publications**, including articles, books and book chapters, and more than 160 communications to international and national congresses. Up to 92 of her publications are cited in PubMed, with 112 contributions in the Web of Science and 100 entries in ORCID. She defends the accessibility of science and 75% of her articles (last decade) are in open access. The **h cumulative index is 33**, being the total number of citations 3446 (Google Scholar), with an average of 22 citations per article, overall supporting the constant contribution and value of her scientific contributions. She has been granted **six research sexenios** (last 31/12/2023), recognized by the CNEAI and AQU (Catalan University System), **seven quinquenios in teaching** (last 31/12/2021), recognized by both the Ministry and the Catalan university system, and **one sexenio of transference** (2020).

Concerning managements tasks, she is **head of the U718 unit of CIBERER**, member of Institut de Biomedicina UB (IBUB-IRSJD), member of Observatorio de Bioética y Derecho and the Committee of Bioethics at Universitat de Barcelona (being the Secretary in 2020-21) and was a Board Member of SEG (Sociedad Española de Genética, 2014-2018). She was the LERU Expert from UB participating in the Group of Experts of Animals used for Scientific Purposes (League of European Research Universities) 2013-2015. She is the **co-founder and scientific assessor of a spin-off company of genetic diagnosis** of inherited visual disorders (DBGen Ocular Genomics, www.dbgen.com, since 2018).

She also believes in being a mentor of younger scientists. She has **supervised 16 PhD Thesis** (5 in the last 5 years); 22 Master thesis and 21 final degree Thesis. She has also mentored three postdoctoral trainees: Alejandro Garanto is now tenured Assistant Professor and PI at Radboudumc (The Netherlands); Serena Mirra is now a senior PI in the Stazione Zoologia Anton Dohrn (Naples, Italy); and Vasileios Toulis, a senior CIBERER postdoctoral researcher, who is now applying for his own research projects.

Dr. Marfany is also extremely active in scientific divulgation: she has written 2 books of divulgative genetics and writes weekly scientific divulgation contributions to digital newspapers and journals (more than 300 articles), and collaborates with generalistic radio and TV programs. She performs conferences in primary and secondary schools, libraries and cultural centres, promoting informed debate on the advances of genetics, and is now coordinator of the Genetics course in Universitat de l'Experiència (addressed to students over 55 years old). She has been awarded the Scientific Divulgation Prize of the University of Barcelona in 2019 and the Lluís Companys Award in 2023, and she was also involved in University management, as Delegate of the rector for scientific divulgation from 2021-2024.

Part C. RELEVANT MERITS

C.1. Ten selected publications of the last 5 years (* corresponding author).

The **criterium for the selection** of these articles is the relevance and impact of the work in the progress of my group's research, by identification of novel genes and mutations causative of genetic diseases, the generation and charactrization of animal models, and substantial contribution to the functional characterization of disease-causative genes. No reviews have been included.

1.- Aísa-Marín I, Rovira Q, Díaz N, Calvo-López L, Vaquerizas JM,* **Marfany G*.** Specific photoreceptor cell fate pathways are differentially altered in NR2E3-associated diseases. ***Neurobiol Dis.*** 194:106463 (2024) doi: 10.1016/j.nbd.2024.106463.

2.- Izquierdo-Villalba I, Mirra S, Manso Y,… **Marfany G**, Auladell C, Navarro X, Enríquez JA, López de Munain A, Soriano E, Aragay AM. A mammalian-specific Alex3/Gαq protein complex regulates mitochondrial trafficking, dendritic complexity, and neuronal survival. ***Sci Signal.*** 17:eabq1007. (2024) doi: 10.1126/scisignal.abq1007.

3.- García-Arroyo R, Domènech EB, Herrera-Úbeda C, … , Mirra S*, **Marfany G*.** Exacerbated response to oxidative stress in the Retinitis Pigmentosa CerklKD/KO mouse model triggers retinal degeneration pathways upon acute light stress. ***Redox Biol.*** 66:102862 (2023). doi: 10.1016/j.redox.2023.102862.

4.- Toulis V, Casaroli-Marano R, Camós-Carreras A… **Marfany G***, Costa MDC* **(10/11).** Altered retinal structure and function in Spinocerebellar ataxia type 3. ***Neurobiol Dis.*** 170:105774 (2022). doi:10.1016/j.nbd.2022.105774.

5.- Sánchez-Bellver, L.; Férriz-Gordillo, A.; Carrillo-Pz,... **Marfany G.* (6/6).**The Deubiquitinating Enzyme USP48 Interacts with the Retinal Degeneration-Associated Proteins UNC119a and ARL3. **Int J Mol Sci.** 23(20):12527 (2022). doi: 10.3390/ijms232012527.

6.- Mirra S*, García-Arroyo R, Domènech EB,… **Marfany G* (10/10).** *CERKL*, a retinal dystrophy gene, regulates mitochondrial function and dynamics in the mammalian retina. ***Neurobiol Dis.*** 156:105405. (2021) doi:10.1016/j.nbd.2021.105405.

7.- Toulis T, García-Monclús S, de la Peña-Ramírez C … Costa MDC* **Marfany G* (10/10).** The deubiquitinating enzyme ataxin-3 regulates retinal ciliogenesis and phagocytosis. ***Cell Reports*** 33:108360. (2020). doi:10.1016/j.celrep.2020.108360.

8.- Aísa-Marín I, Lopez-Iniesta MJ, Milla S, Lillo J, Navarro G, de la Villa P, **Marfany G* (7/7).** *Nr2e3* functional domain ablation by CRISPR-Cas9D10A identifies a new isoform and generates retinitis pigmentosa and enhanced S-cone syndrome models. ***Neurobiol Dis***. 146:105122. (2020). doi:10.1016/j.nbd.2020.105122.

9.- Domènech EB, Andrés R, Lopez-Iniesta MJ,…**Marfany G* (11/11).** A new *Cerkl* mouse model generated by CRISPR-Cas9 shows progressive retinal degeneration and altered morphological and electrophysiological phenotype. ***Invest. Ophthalmol. Vis. Sci.*** 61:14 (2020). doi:10.1167/iovs.61.8.14.

10.- Bolinches-Amorós A, … **Marfany G**, Gonzàlez-Duarte R, Erceg S, Lukovic D. **(4/7).** Generation of an iPSC line from a retinitis pigmentosa patient carrying a homozygous mutation in *CERKL* and a healthy sibling. ***Stem Cell Res.*** 38:101455 **(**2019). doi:10.1016/j.scr.2019.101455

C.2. Invited & oral communications to international and national congresses (selection)

1.- Bringing light to darkness: *CERKL*, a retinal resilience gene against oxidative stress**. Marfany, G.** Invited speaker. ***WORKSHOP "REDOX BIOLOGY IN HEALTH AND DISEASE" (Barcelona)***

2.- CRISPR/Cas9 editing in iPSCs and generation of 3D retinal organoids as inherited retinal dystrophy models. **Marfany, G.** Invited speaker in the session **"**Exploring Cellular and Organoid Platforms for Advancing Disease Understanding and Therapeutic Development in Retinal Disorders". ***ARVO SIG 2024 (online)***

3.- Impairment of light-induced stress response in the Retinitis Pigmentosa mouse model CerkIKD/KO. García-Arroyo, R., Domènech, E., Herrera-Úbeda, C., Asensi, M.A., Pallardó, F.V., Garcia-Fernández, J., **Marfany, G**., Mirra, S. Oral communication. ***ESHG 2023, Glasgow***.

4.- *CERKL*, a retinal dystrophy gene, regulates mitochondrial function and dynamics in the mammalian retina. **Marfany G.** Invited Speaker. (2022). ***16th PRO RETINA (Potsdam, Germany)***

5.- NR2E3 transcription factor and photoreceptor fate: identification of gene regulatory networks causing retinal remodeling in NR2E3-associated diseases. Aísa-Marin I, Rovira Q, Díaz N, Vaquerizas JM, **Marfany G.** Oral communication. ***ESHG 2021 (Vienna, Austria)***

6.- Gene-edited mouse models and human retinal organoids to study the function of *CERKL*, *NR2E3* and *ATXN3* genes in inherited retinal dystrophies. **Marfany G.** Oral communication. ***ARVO 2019 (Vancouver).***

C.3. Selected Research projects and grants last 10 years

1.- PID2022-140957OB-I00. Modelizacion de enfermedades hereditarias de retina en raton y organoides humanos para explorar estrategias de terapia genica. Ministerio de Ciencia e Innovación (MICINN). **PI: Gemma Marfany Nadal**, 2023-2026.

2.- Ref: 101017229 - Transforming Open Responsible Research and Innovation through CHARM (TORCH), European University Alliance. **G. Marfany. Work Package Leader WP10 & WP11.** 2021-2023.

3.- 2021SGR1093. Genètica Molecular Humana. Grupos Consolidados. Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). **PI:** Raquel Rabionet Janssen. (**G. Marfany** is a senior member of the team) 2023-2025.

4.- PID2019-108578RB-I00. Explorando las vías de señalización neurosensoriales en enfermedades hereditarias de retina como posible aproximación a la terapia. Ministerio de Ciencia e Innovación. **co-PIs: Gemma Marfany Nadal**, Serena Mirra 2020-2023.

5.- ACCI2020. Regulación de la proteostasis en la formación del cilio: una diana terapéutica en ciliopatías (CILIOPAT). CIBERER. **Co-PIs: Gemma Marfany Nadal,** F. Garcia-Gonzalo.

6.- SAF2016-80937-R. Modelos Animales para las distrofias de retina: estrategias de edición genómica, fenotipado y nanoterapia. Ministerio de Economia y Competitividad. **coPIs: Gemma Marfany Nadal**, Roser Gonzalez Duarte 2016-2019.

7.- REF: 20141730- Fundació La Marató de TV3. Dissecting protein trafficking in retinal neurodegeneration by super-resolution imaging on animal models and human iPSCs. IP: Roser Gonzalez Duarte. (**G. Marfany** was a senior member of the team) 2015-2017.

8.- BM1307. PROTEOSTASIS: European network to integrate research on intracellular proteolysis pathways in health and disease. (Comission of the European Communities - Directorate General Joint Research Centre, JRC). PI: Rosa Barrio Olano. 2014-2019. (**G. Marfany** was the Management Core Member and Coordinator of the Dissemination Activities).

9.- ACCI2015- MODCELANI_CRISPR. Nuevos modelos animales de enfermedades raras neurosensoriales generados mediante la tecnología CRISPR-Cas9. U718- Modelos animales en ratón de retinosis pigmentaria asociados a mutaciones en los genes CERKL y NR2E3. CIBERER. **PI: Gemma Marfany.** 2016–2017.

10.- SAF2013-49069-C2-1-R. Identificación de dianas terapéuticas para distrofias de retina:caracterización funcional in vivo e in vitro de genes causantes de ceguera hereditaria. Ministerio de Economia y Competitividad. **co-PIs:** Roser Gonzalez Duarte, **Gemma Marfany.** 2014-2016.

C.4. Knowledge transference, partnership contract

1.- Generation of a new spin-off company: DBGen Ocular Genomics. Participation in R&D and Innovation contract between the Universitat of Barcelona and DBGEN Ocular Genomics, S.L **Gemma Marfany Nadal** (co-founder and scientific assessor).19/01/2018-01/06/2024.

2.- Patent Application Registry: Método de análisis de haplotipos para el diagnóstico o pronóstico de patologías que cursen con distrofia retiniana Inventores/autores/obtentores: Gonzalez-Duarte, R.; **Marfany, G**.; Pomares, E.; Carracedo, A.; Brión, M. Entidad titular de derechos: Universidade de Santiago de Compostela, Universitat de Barcelona. Nº de solicitud: P200601038. País de inscripción: España. Fecha de registro: 2006.