

## *CURRICULUM VITAE*

<b>NAME</b> Valeria Marigo	<b>POSITION TITLE</b> Associate Professor University of Modena and Reggio Emilia	<b>BIRTHDATE</b> 12/13/1963	
<b>INSTITUTION AND LOCATION</b>	<b>DEGREE</b>	<b>YEAR CONFERRED</b>	<b>FIELD OF STUDY</b>
University of Padua, Italy	Degree in Biology	1987	Biological Sciences
University of Padua and University La Sapienza (Rome), Italy	Ph.D.	1993	Morpho- genetic and Cytological Sciences

### Research and Professional Experience

03/1986-10/1988	Predocctoral Fellow, Institute of Anatomy, University of Padua, Italy, Supervisor: Prof. M. Bortolussi
03/1987	<b>Degree in Biology</b> (Laurea), Summa cum laude
11/1988-03/1993	PhD student, Institute of Histology and Embryology, University of Padua, Italy; Supervisors: Profs. D. Volpin and G. M. Bressan
06/1993	<b>Ph.D. in Morphogenetic and Cytological Sciences</b>
04/1993-11/1996	<b>Postdoctoral Fellow</b> , Department of Genetics, Harvard Medical School, Boston, MA; Supervisor: Prof. Clifford J. Tabin
12/1996- 09/2000	<b>Researcher</b> , Telethon Institute of Genetics and Medicine (TIGEM), Milan, Italy; Director: Prof. A. Ballabio
10/2000- 10/2005	<b>Group Leader</b> , Telethon Institute of Genetics and Medicine (TIGEM), Naples, Italy; Director: Prof. A. Ballabio
06/2000-06/2005	<b>Supervisor of the RNA In Situ Hybridization Core</b> , TIGEM, Naples, Italy
11/2001- current	Member of the Scientific Advisory Board of “The Vision of Children Foundation” (San Diego, CA)
11/2004- 10/2005	Member of the New York Academy of Sciences.
11/2005- current	<b>Associate Professor of Molecular Biology</b> at Department of Biomedical Sciences University of Modena and Reggio Emilia, Modena, Italy
2007- current	Member of the Editorial Board of the journal “Pigment Cell & Melanoma Research”

### Honors

April 1988     Award: Centro Regionale Specializzato per la Epidemiologia e la Prevenzione dell' Arteriosclerosi (University of Padua)

### Fellowships

03/1993 – 08/1994: Borsa di Studio per Attività di Perfezionamento all'Estero (University of Padua, Italy) at the Department of Genetics, Harvard Medical School, (USA) in the laboratory of Prof. Clifford J. Tabin.

07 –09/1998: Deutscher Akademischer Austauschdienst (DAAD) at the Institute of Genetics, GSF-Neuherberg (Germany) in the laboratory of Prof. Jochen Graw.

03/2003: EMBO Short term Fellowship at Department of Anatomy and Cell Biology, University of Toronto, Canada in the laboratory of Dr. Derek van der Kooy.

### **Grants**

- Fondazione Telethon, numero T02 (Luglio 2000 – Giugno 2003)
- Fondazione Telethon, numero T03 (Luglio 2000 – Giugno 2003)
- Vision of Children Foundation (Giugno 2001 – Maggio 2004)
- March of Dimes #1-FY01-117 (Giugno 2001 – Maggio 2004)
- Fondazione Telethon (Luglio 2003 – Giugno 2006)
- Co-PI in NIH grant 1R01EY015136-01 (Ottobre 2003 – Settembre 2008)
- European Retinal Research Training Network RET-NET MRTN-CT-2003-504003 (Gennaio 2004 – Dicembre 2007)
- VI European framework IP, EVI-GENORET LSHG-CT-2005-512036 (Aprile 2005 - Marzo 2008)
- Progetti di cooperazione scientifica e tecnologica - Accordo Italia-USA, RBIN042ARX (2006-2009)
- Fondazione Telethon GGP06096 (Novembre 2006 - Ottobre 2009)
- COFIN PRIN 2006053302\_003 (2007-2008)

### **Invited speaker at:**

- “Santa Cruz Summer Conference: Developmental Biology” July 1996, title: “Hedgehog signaling”
- Biotech Company Ontogeny (Boston, USA) October 1996, title: “The Sonic hedgehog signaling pathway”
- “Pediatric Academic Society Meeting”, Washington, USA May 1997, title: “The Sonic hedgehog signaling pathway in development and human diseases”
- “European Society of Human Genetics”, Genoa May 1997, title: “The Sonic hedgehog signaling pathway in development and human diseases “
- “XIV International Congress of Eye Research” Santa Fe, USA October 2000 title: “Study of RP pathogenesis by regulation of P23H rhodopsin allele expression”
- “The Association for research in Vision and Ophthalmology”, Fort Lauderdale, USA May 2001; title: “A mouse model for Ocular albinism type 1 (OA1)”
- “Concerted Action of the European Union, New therapeutic approaches in hereditary eye disease-from gene to cure”, Prague, Czech Rep. July 2001; title: “Animal models of eye diseases for pathogenetic studies and phenotype rescue attempts”
- “European Society for Pigment Cell Research”, Rome September 2001; title: “The mouse model for Ocular albinism type 1 (OA1) as a tool to study OA1 function and to develop therapeutic approaches”
- University of Genoa April 2002; title: “The mouse model for Ocular albinism type 1 (OA1) as a tool to study OA1 function”
- University of Geneva; October 2002; title: “The mouse model for Ocular albinism type 1 (OA1) as a tool to study OA1 function and to develop therapeutic approaches”
- “15th IIGB meeting From Genome Sequence to Functional Analysis and Medical Applications” October 2002; title: “Human chromosome 21 gene expression atlas in the mouse”
- Functional genomics and disease”, Prague, Czech Rep. May 2003; title: “Human chromosome 21 gene expression atlas in the mouse”
- University of Padua June 2003; title: “Atlante dei geni del cromosoma 21 umano”
- University of Modena April 2004; title: “Albinismo oculare di tipo 1: identificazione degli elementi regolatori che conferiscono tessuto specificità pre approcci terapeutici”
- The Scripps Research Institute, San Diego, USA August 2004; title: "Function and regulation of the OA1 gene"
- Jules Stein Eye Institute UCLA School of Medicine Los Angeles, USA, August 2004; title: "Function and regulation of the OA1 gene"

- Department of Human Genetics, UCLA School of Medicine Los Angeles, USA 9 August 2004; title: “Large scale RNA in situ hybridization as a tool for functional studies of human diseases”
- “17th IGB meeting The Biology and development of the Eye in health and Disease” Capri, October 2004; title: “Morpholino correction of a retinal pigment epithelium defect”
- “The Association for research in Vision and Ophthalmology”, Fort Lauderdale, USA May 2005; title: “Functional studies of mutations found in PRPF3 pre-mRNA splicing factor causino RP18”
- 13th meeting of the European Society for Pigment cell research, ESPCR06” Barcelona, Spain; title: “Albinism as a defect of melanosome number and maturation and not of melanin production: a lesson from the OA1 gene”
- “EVER 2006”, Faro, Portugal October 2006; title: “Stem cell differentiation: the importance of functional genomics”
- “International Congress of Eye Research”, Buenos Aires, Argentina November 2006; title: “Defect of melanosome number and maturation and not of melanin production as a cause of albinism: a lesson from the OA1 gene”.
- EVI-GENORET Symposium “Models, mechanisms and treatment strategies for retinal degeneration” Paris, October 2007; title: “Cross-talk between AIF and caspase-12 in retinal degeneration mouse models”.
- “European Vision Summit 2007”, Interlaken, CH, December 2007; title: “Common apoptotic mechanisms in different forms of retinitis pigmentosa”.

#### Chairman at:

- “Transgenic models of inducible disease” International Congress of Eye Research. October 2000 Santa Fe, New Mexico, USA.
- “Animal models of retinal disease I” Association for Research in Vision and Ophthalmology (ARVO), May 2001 Fort Lauderdale, Florida, USA.

#### Publications

**Marigo, V.**, Sitta, A., Volpin, D., and Bressan, G. M. (1992). Mapping of binding sites for monoclonal antibodies to chick tropoelastin by recombinant DNA techniques. *Connective Tissue Research* 28: 13-28.

**Marigo, V.**, Daga-Gordini, D., Sitta, A., Volpin, D., and Bressan, G. M. (1992). Effect of monoclonal antibodies to defined regions of tropoelastin on elastogenesis in vitro. *European Journal of Cell Biology* 57: 254-264.

Bonaldo, P., Piccolo, S., Marvulli, D., Volpin, D., **Marigo, V.**, and Bressan, G. M. (1993). Murine  $\alpha 1(\text{VI})$  collagen chain. Complete amino acid sequence and identification of the gene promoter region. *Matrix* 13: 223-233.

**Marigo, V.**, Volpin, D., and Bressan, G. M. (1993). Regulation of the human elastin promoter in chick embryo cells. Tissue-specific effect of TGF- $\beta$ . *Biochimica Biophysica Acta* 1172: 31-36.

Bressan, G.M., Daga-Gordini, D., Colombatti, A., Castellani, I., **Marigo, V.**, and Volpin, D. (1993). Emilin, a component of elastic fibers preferentially located at the elastin-microfibrils interface. *The Journal of Cell Biology* 121: 201-212.

**Marigo, V.**, Volpin, D., Vitale, G., and Bressan, G.M. (1994). Identification of a TGF- $\beta$  responsive element in the human elastin promoter. *Biochemical and Biophysical Research Communications* 199: 1049-1056.

- Laufer, E., and **Marigo, V.** (1994). Evolution in developmental biology: of morphology and molecules. *Trends in Genetics* 10: 261-263.
- Marigo, V.**, Roberts, D.J., Lee, S.M.K., Tsukurov, O., Levi, T., Gastier, J.M., Epstein, D.J., Gilbert, D.J., Copeland, N.G., Seidman, C.E., Jenkins, N.A., Seidman, J.G., McMahon, A.P., and Tabin C. (1995). Cloning, expression, and chromosomal location of SHH and IHH: two human homologues of the *Drosophila* segment polarity gene hedgehog. *Genomics* 28: 44-51.
- Piccolo, S., **Marigo, V.**, Giroto, D., Volpin, D., and Bressan, G.M. (1995). Identification of a recognition element for CAAT-enhancer binding proteins (C/EBPs) in the elastin promoter. *Biochimica Biophysica Acta* 1264: 40-44.
- Wang, M.Z., Jin, P., Bumcrot, D.A., **Marigo, V.**, McMahon, A.P., Wang, E.A., Woolf, T., and Pang, K. (1995). Induction of dopaminergic neuron phenotype in the midbrain by Sonic hedgehog protein. *Nature Medicine* 1: 1184-1188.
- Marigo, V.**, Scott, M.P., Johnson, R.L., Goodrich, L.V., and Tabin, C.J. (1996). Conservation in hedgehog signaling: induction of a chicken patched homolog by Sonic hedgehog in the developing limb. *Development* 122: 1225-1233.
- Marigo, V.**, Laufer, E., Nelson, C.E., Riddle, R.D., Johnson, R.L., and Tabin, C. (1996). Sonic hedgehog regulates patterning in early embryos. *Biochemical Society Symposium* 62: 51-60.
- Marigo, V.**, and Tabin C. (1996). Regulation of Patched by Sonic hedgehog in the developing neural tube. *Proceedings of the National Academy of Sciences USA* 93: 9346-9351.
- Marigo, V.**, Davey, R., Zuo, Y., Cunningham, J.M., and Tabin C. (1996). Biochemical evidence that Patched is the Hedgehog receptor. *Nature* 384: 176-179.
- Marigo, V.**, Johnson, R.L., Vortkamp, A., and Tabin, C.J. (1996). Sonic hedgehog differentially regulates expression of GLI and GLI3 during limb development. *Developmental Biology* 180: 273-283.
- Quirk, J., van den Heuvel, M., Henrique, D., **Marigo, V.**, Jones, T.A., Tabin, C., and Ingham, P.W. (1997). The Smoothed gene and Hedgehog signal transduction in *Drosophila* and vertebrate development. *Cold Spring Harbor Symposia on Quantitative Biology* LXII: 217-226.
- Bulfone, A., Martinez, S., **Marigo, V.**, Campanella, M., Basile, A., Quaderi, N., Gattuso, C., Rubenstein, J.L.R., and Ballabio, A. (1999). Expression pattern of the *Tbr2* (Eomesodermin) gene during mouse and chick brain development. *Mechanisms of Development* 84: 133-138.
- Incerti, B., Cortese, K., Pizzigoni, A., Surace, E.M., Varani, S., Coppola, M., Jeffery, G., Seeliger, M., Jaissle, G., Bennett, D.C., **Marigo, V.**, Schiaffino, M.V., Tacchetti, C., Ballabio, A. (2000). Oa1 knock-out: new insights on the pathogenesis of Ocular Albinism type 1. *Human Molecular Genetics* 9: 2781-2788.
- Surace, E.M., Angeletti, B., Ballabio, A., **Marigo, V.** (2000). Expression pattern of the ocular albinism type 1 (OA1) gene in the murine retinal pigment epithelium. *Investigative Ophthalmology & Visual Science* 41: 4333-4337.
- Smith, D.M., Collins-Racie, L.A., **Marigo, V.**, Roberts, D.J., Davis, N.M., Hartmann, C., Schweitzer, R., LaVallie, E.R., Gamer, L., McCoy, J., Tabin C.J. (2001). Cloning and expression of a novel Cysteine-Rich Secreted Protein (CRISP) family member expressed in thyroid and pancreatic mesoderm within the chicken embryo. *Mechanisms of Development* 102: 223-226.

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- Bolino, A., **Marigo, V.**, Ferrara, F., Loader, J., Romio, L., Leoni, A., Di Duca, M., Cinti, R., Cecchi, C., Feltri, M.L., Wrabetz, L., Ravazzolo, R., Monaco A. (2002) Molecular characterization and expression analysis of *Mtmr2*, mouse homologue of *MTMR2*, the myotubularin-related 2 gene, mutated in CMT4B. *Gene* 283: 17-26.
- Reymond, A. \*, **Marigo, V.** \*, Yaylaoglu, M.B. \*, Leoni, A., Ucla, C., Scamuffa, N., Caccioppoli, C., Dermitzakis, E.T., Lyle, R., Banfi, S., Eichele, G., Antonarakis, S.E., Ballabio A. (2002) Human chromosome 21 gene expression atlas in the mouse. *Nature* 420: 582-586. (\* these authors equally contributed to this work).
- Angeletti, B., Löster, J., Auricchio, A., Gekeler, F., Shinoda, K., Ballabio, A., Graw, J., and **Marigo, V.** (2003). An *in vivo* doxycycline-controlled expression system for functional studies of the retina. *Investigative Ophthalmology & Visual Science* 44:755-760.
- Buniello, A., Montanaro, D., Volinia, S., Gasparini, P., **Marigo, V.** (2004). An expression atlas of *connexin* genes in the mouse. *Genomics* 83: 812-820.
- Marigo, V.**, Nigro, A., Pecci, A., Montanaro, D., Di Stazio, M.T., Balduini, C.L., Savoia, A. (2004). Correlation between the clinical phenotype of *MYH9*-related disease and tissue distribution of class II non-muscle myosin heavy chains. *Genomics* 83: 1125-1133.
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- Surace E.M., Domenici L., Cortese K., Cotugno G., Di Vicino U., Venturi C., Cellerino A., **Marigo V.**, Tacchetti C., Ballabio A., Auricchio A. (2005). Amelioration of both functional and morphological abnormalities in the retina of a mouse model of ocular albinism following AAV-mediated gene transfer. *Molecular Therapy* 12: 652-658.
- Bocciardi R., Giorda R., **Marigo V.**, Zordan P., Montanaro D., Gimelli S., Seri M., Lerone M., Ravazzolo R., Gimelli G. (2005) Molecular characterization of a T(2;6) balanced translocation associated with complex phenotype and leading to the truncation of the *TCBA1* gene. *Human Mutation* 26: 426-36.
- Cortese K., Giordano F., Surace E.M., Venturi C., Ballabio A., Tacchetti C., **Marigo V.** (2005) The Ocular albinism type 1 (*OAI*) gene controls melanosome maturation and size. *Investigative Ophthalmology & Visual Science* 46: 4358-4364.
- Vetrini F., Tammaro R., Bondanza S., Surace E.M., Auricchio A., De Luca M., Ballabio A., **Marigo V.** (2006)

- Aberrant splicing in the ocular albinism type 1 gene (*OAI/GPR143*) is corrected *in vitro* by morpholino antisense oligonucleotides. *Human Mutation* 27: 420-426.
- Sanges D., **Marigo V.** (2006) Cross-talk between two apoptotic pathways activated by endoplasmic reticulum stress: differential contribution of Caspase-12 and AIF. *Apoptosis* 11: 1629-1641.
- Sanges D., Comitato A., Tammaro R., **Marigo V.** (2006) Apoptosis in retinal degeneration involves cross-talk between AIF and caspase-12 and is blocked by calpain inhibitors. *Proceedings of the National Academy of Sciences USA* 103: 17366-17371.
- Karali M., Peluso I., **Marigo V.**, Banfi S. (2007) Identification and characterization of microRNAs expressed in the mouse eye. *Investigative Ophthalmology & Visual Science* 48: 509-515.
- Marigo V.** (2007) Programmed Cell Death in Retinal Degeneration: Targeting Apoptosis in Photoreceptors as Potential Therapy for Retinal Degeneration. *Cell Cycle* 6: 652-655.
- Comitato A., Spanpanato C., Chakarova C., Sanges D., Bhattacharya S.S., **Marigo V.** (2007) Mutations in splicing factor PRPF3, causing retinal degeneration, form detrimental aggregates in photoreceptor cells. *Human Molecular Genetics* 16: 1699-1707.
- Allocca, M., Mussolino, C., Hoyos, M. G., Sanges, D., Iodice, C., Petrillo, M., Vandenberghe, L. H., Wilson, J. M., **Marigo, V.**, Surace, E. M., Auricchio, A. (2007). Novel AAV serotypes efficiently transduce murine photoreceptors. *Journal of Virology* 81: 11372- 11380.
- Chakarova C.F., Papaioannou M.G., Khanna H., Lopez I., Waseem N., Shah A., Theis T., Friedman J. , Maubaret C., Bujakowska K., Veraitch B., Abd El-Aziz M.M., Prescott D.Q., Parapuram S., Bickmore W.A., Munro P.M.G., Gal A., Hamel C., **Marigo V.**, Ponting C.P., Wissinger B, Zrenner E., Matter K., Swaroop A., Koenekoop R.K. Bhattacharya S.S. (2007) Mutations in *TOPORS* Cause Autosomal Dominant Retinitis Pigmentosa with Perivascular Retinal Pigment Epithelium Atrophy. *The American Journal of Human Genetics* 81: 1098-1103.
- Giordano F., De Marzo A. Vetrini F., **Marigo V.** (2007) FGF and EGF differently affect differentiation of murine retinal stem cells in vitro. *Molecular Vision* 13: 1842-1850.
- Trifunović D., Karali M., Camposampiero D., Ponzin D., Banfi S., Marigo V. (2008) A high-resolution RNA expression atlas of Retinitis Pigmentosa genes in the human and mouse retinas. *Investigative Ophthalmology & Visual Science* in press.