MACARENA GOMEZ LIRA, Bsc PhD

Biological Sciences degree obtained in January 1981 at the Universidad de Chile, Santiago, Chile
Doctoral degree obtained in2009 at the University of Verona, Italy

1981-1984 working at the Society for Biotechnological Research, Department of Molecular Biology, Braunschweig, Germany. Main interest is the study of the role of histone acetylation and phosphorylation in relation to events of cell cycle gene activation and cell transformation
-**April 1989-until now**: performs research at the University of Verona, DMIBG, Section Biology and Genetics.
Currently (2010- until now),
- Study of candidate genes that predisposeto atherosclerosis based on association and expression studies in human atherosclerotic plaques and peripheral blood.
-Study of candidate genes that predispose to cutaneous melanoma based on association studies using DNA of patients and controls, and gene expression using RNA from culture fibroblasts.
History
-**Until 1994**, carries out studies on the molecular genetic of Osteogenesis Imperfecta,
-**From 1994** -1999 is responsible for organizing and coordinating Molecular genetic studies on Sandhoff disease, metachromatic leukodystrophy and adrenoleukodystrophy of (ALD).
1999 to 2011 Analysis of genetic risk factors of various diseases such as idiopathic pancreatitis (search for gene mutations in SPINK1 and CFTR), multiple sclerosis (MOG genes, CD45, PLA2G7, COX-2), in IgA nephropathy. (Genes of the RAS and TGF beta1), atherosclerosis (PLA2G7 genes and COX-2) and predisposition to skin cancer after transplantation (GSTs, CYP1A1, PTCH1 and COX-2 genes).
2011-today.-Professor assistant at the UNIVR; is responsible for the organization and coordination of several candidate genes association studies in complex diseaseases including non-melanoma skin cancer, melanoma, atherosclerosis, and arthritis reumatoide, and functional studies on the effect of mutations on gene expression
Grants

From 1999 to 2002 grants and research contracts for the study of various genetic diseases such as Adrenoleukodistrophy, the metachromatic leukodystrophy, multiple sclerosis and idiopathic pancreatitis.

2002 -2010 research grants from the University of Verona on the project "Molecular genetic analysis of multifactorial diseases

2010-2011, research contract for the study of gene expression in human vulnerable atherosclerotic plaques.

2011-today,Professor Assistant in Medice-Genetics the University of Verona

PUBBLICATIONS

H index= 14

1: Baldan A, Ferronato S, Olivato S, Malerba G, Scuro A, Veraldi GF, Gelati M, Ferrari S, Mariotto S, Pignatti PF, Mazzucco S, **Gomez-Lira M.** Cyclooxygenase 2, Toll-like receptor 4 and interleukin 1β mRNA expression in atherosclerotic plaques of type 2 diabetic patients. Inflamm Res. 2014 Oct;63(10):851-8. PubMed PMID: 25095741.

2: **Gomez-Lira M,** Ferronato S, Malerba G, Santinami M, Maurichi A, Sangalli A, Turco A, Perego P, Rodolfo M. Association of promoter polymorphism -765G>C in the PTGS2 gene with malignant melanoma in Italian patients and its correlation to gene expression in dermal fibroblasts. Exp Dermatol. 2014 Oct;23(10):766-8. PubMed PMID: 25060715.

3: Ferronato S, **Gomez-Lira M**, Menegazzi M, Diani E, Olivato S, Sartori M, Scuro A, Malerba G, Pignatti PF, Romanelli MG, Mazzucco S. Polymorphism -2604G>A variants in TLR4 promoter are associated with different gene expression level in peripheral blood of atherosclerotic patients. J Hum Genet. 2013 Dec;58(12):812-4. PubMed PMID: 24108365.

4: Gajofatto A, Stefani A, Turatti M, Bianchi MR, **Lira MG**, Moretto G, Salviati A, Benedetti MD. Prevalence of multiple sclerosis in Verona, Italy: an epidemiological and genetic study. Eur J Neurol. 2013 Apr;20(4):697-703. PubMed PMID: 23279712.

5: **Gomez-Lira M,** Tessari G, Mazzola S, Malerba G, Rugiu C, Naldi L, Nacchia F, Valerio F, Anna B, Forni A, Boschiero L, Sandrini S, Faggian G, Girolomoni G,

Turco A. Analysis of the 3'UTR of the prostaglandin synthetase-2 (PTGS-2/COX-2) gene in non-melanoma skin cancer after organ transplantation. Exp Dermatol. 2011 Dec;20(12):1025-7.
PubMed PMID: 21995456.
6: Steiner B, Rosendahl J, Witt H, Teich N, Keim V, Schulz HU, Pfützer R, Löhr M, Gress TM, Nickel R, Landt O, Koudova M, Macek M Jr, Farre A, Casals T, Desax MC, Gallati S, **Gomez-Lira M**, Audrezet MP, Férec C, des Georges M, Claustres M, Truninger K. Common CFTR haplotypes and susceptibility to chronic pancreatitis and congenital bilateral absence of the vas deferens. Hum Mutat. 2011 Aug;32(8):912-20. doi: 10.1002/humu.21511. Epub 2011 Jun 7. Erratum in: Hum Mutat. 2012 Feb;33(2):456. Lühr, Matthias [corrected to Löhr, Matthias]. PubMed PMID: 21520337.
7: Begnini A, Tessari G, Turco A, Malerba G, Naldi L, Gotti E, Boschiero L, Forni A, Rugiu C, Piaserico S, Fortina AB, Brunello A, Cascone C, Girolomoni G, **Gomez Lira M**. PTCH1 gene haplotype association with basal cell carcinoma after transplantation. Br J Dermatol. 2010 Aug;163(2):364-70.
8: **Lira MG**, Provezza L, Malerba G, Naldi L, Remuzzi G, Boschiero L, Forni A, Rugiu C, Piaserico S, Alaibac M, Turco A, Girolomoni G, Tessari G. Glutathione S-transferase and CYP1A1 gene polymorphisms and non-melanoma skin cancer risk in Italian transplanted patients. Exp Dermatol. 2006 Dec;15(12):958-65. Erratum in: Exp Dermatol. 2011 Apr;20(4):375-6. PubMed PMID: 17083362.
9: Ferronato S, **Lira MG,** Olivato S, Scuro A, Veraldi GF, Romanelli MG, Patuzzo C, Malerba G, Pignatti PF, Mazzucco S. Upregulated expression of Toll-like receptor 4 in peripheral blood of ischaemic stroke patients correlates with cyclooxygenase 2 expression. Eur J Vasc Endovasc Surg. 2011 Mar;41(3):358-63.
PubMed PMID: 21236709.
10: Brezzi B, Del Prete D, Lupo A, Magistroni R, **Gomez-Lira M,** Bernich P, Anglani F, Mezzabotta F, Turco A, Furci L, Ceol M, Antonucci F, Abaterusso C, Bonfante L, D'Angelo A, Albertazzi A, Gambaro G. Primary IgA nephropathy is more severe in TGF-beta1 high secretor patients. J Nephrol. 2009 Nov-Dec;22(6):747-59. PubMed PMID: 19967654.
11: **Gomez Lira M**, Provezza L, Terranova C, Martinelli N, Bozzini C, Pignatti PF. Two new highly polymorphic markers in the 3' UTR region of the PLA2G7 gene. Int J

Immunogenet. 2007 Dec;34(6):465-8. PubMed PMID: 18001304.
12: D'Alessandro M, Coats SE, Morley SM, Mackintosh L, Tessari G, Turco A, Gerdes AM, Pichert G, Whittaker S, Brandrup F, Broesby-Olsen S, **Gomez-Lira M,** Girolomoni G, Maize JC, Feldman RJ, Kato N, Koga Y, Ferguson-Smith MA, Goudie DR, Lane EB. Multiple self-healing squamous epithelioma in different ethnic groups: more than a founder mutation disorder? J Invest Dermatol. 2007 Oct;127(10):2336-44. PubMed PMID: 17554363.
13: Mazzola S, **Lira MG**, Benedetti MD, Salviati A, Ottaviani S, Malerba G, Ortombina M, Pignatti PF. COX-2 promoter region polymorphisms in multiple sclerosis: lack of association of -765G>C with disease risk. Int J Immunogenet. 2007 Apr;34(2):71-4. PubMed PMID: 17373929.
14: **Lira MG**, Mazzola S, Tessari G, Malerba G, Ortombina M, Naldi L, Remuzzi G, Boschiero L, Forni A, Rugiu C, Piaserico S, Girolomoni G, Turco A. Association of functional gene variants in the regulatory regions of COX-2 gene (PTGS2) with nonmelanoma skin cancer after organ transplantation. Br J Dermatol. 2007 Jul;157(1):49-57. PubMed PMID: 17578436
15: **Gomez-Lira M**, Liguori M, Magnani C, Bonamini D, Salviati A, Leone M, Andreoli V, Trojano M, Valentino P, Savettieri G, Quattrone A, Pignatti PF, Momigliano-Richiardi P, Giordano M. CD45 and multiple sclerosis: the exon 4 C77G polymorphism (additional studies and meta-analysis) and new markers. J Neuroimmunol. 2003 Jul;140(1-2):216-21. PubMed PMID: 12864992.
16: **Gomez-Lira M**, Bonamini D, Castellani C, Unis L, Cavallini G, Assael BM, Pignatti PF. Mutations in the SPINK1 gene in idiopathic pancreatitis Italian patients. Eur J Hum Genet. 2003 Jul;11(7):543-6. PubMed PMID: 12825076.
17: Patuzzo C, Castellani C, Sagramoso C, **Gomez-Lira M,** Bonamini D, Belpinati F, Dechecchi MC, Assael BM, Pignatti PF. Cationic trypsinogen and pancreatic secretory trypsin inhibitor gene mutations in neonatal hypertrypsinaemia. Eur J Hum Genet. 2003 Jan;11(1):93-6. PubMed PMID: 12529713.
18: **Gomez-Lira M,** Moretto G, Bonamini D, Benedetti MD, Pignatti PF, Rizzuto N, Salviati A. Myelin oligodendrocyte glycoprotein polymorphisms and multiple sclerosis. J Neuroimmunol. 2002 Dec;133(1-2):241-3. PubMed PMID: 12446029.
19: **Gomez Lira M,** Patuzzo C, Castellani C, Bovo P, Cavallini G, Mastella G, Pignatti PF. CFTR and cationic trypsinogen mutations in idiopathic pancreatitis

and neonatal hypertrypsinemia. Pancreatology. 2001;1(5):538-42. PubMed PMID: 12120234.
20: Benedetti MD, Salviati A, Filipponi S, Manfredi M, De Togni L, **Gomez Lira M,** Stenta G, Fincati E, Pampanin M, Rizzuto N, Danti G. Prevalence of dementia and apolipoprotein e genotype distribution in the elderly of buttapietra, verona province, Italy. Neuroepidemiology. 2002 Mar-Apr;21(2):74-80. PubMed PMID: 11901276.
21: Primorac D, Rowe DW, Mottes M, Barisić I, Anticević D, Mirandola S, **Gomez Lira M,** Kalajzić I, Kusec V, Glorieux FH. Osteogenesis imperfecta at the beginning of bone and joint decade. Croat Med J. 2001 Aug;42(4):393-415. Review. PubMed PMID: 11471191.
22: Castellani C, **Gomez Lira M**, Frulloni L, Delmarco A, Marzari M, Bonizzato A, Cavallini G, Pignatti P, Mastella G. Analysis of the entire coding region of the cystic fibrosis transmembrane regulator gene in idiopathic pancreatitis. Hum Mutat. 2001 Aug;18(2):166. PubMed PMID: 11462247.
23: **Gomez-Lira M,** Mottes M, Perusi C, Pignatti PF, Rizzuto N, Gatti R, Salviati A. A novel 4-bp deletion creates a premature stop codon and dramatically decreases HEXB mRNA levels in a severe case of Sandhoff disease. Mol Cell Probes. 2001 Apr;15(2):75-9. PubMed PMID: 11292324.
24: **Gomez-Lira M**, Marzari MG, Uziel G, Pignatti P, Rizzuto N, Salviati A. Myelin oligodendrocyte glycoprotein (MOG) polymorphisms and adrenoleukodystrophy. J Neuroimmunol. 2000 Nov 1;111(1-2):245-7. PubMed PMID: 11063846.
25: Felice KJ, **Gomez Lira M,** Natowicz M, Grunnet ML, Tsongalis GJ, Sima AA, Kaplan RF. Adult-onset MLD: a gene mutation with isolated polyneuropathy. Neurology. 2000 Oct 10;55(7):1036-9. PubMed PMID: 11061266.
26: **Gomez Lira M**, Benetazzo MG, Marzari MG, Bombieri C, Belpinati F, Castellani C, Cavallini GC, Mastella G, Pignatti PF. High frequency of cystic fibrosis transmembrane regulator mutation L997F in patients with recurrent idiopathic pancreatitis and in newborns with hypertrypsinemia. Am J Hum Genet. 2000
27: **Lira MG**, Mottes M, Pignatti PF, Medica I, Uziel G, Cappa M, Bertini E, Rizzuto N, Salviati A. Detection of mutations in the ALD gene (ABCD1) in seven Italian families: description of four novel mutations. Hum Mutat. 2000 Sep;16(3):271. PubMed PMID: 10980539.

Jun;66(6):2013-4. PubMed PMID: 10801389; PubMed Central PMCID: PMC1378065.
28: Mottes M, **Gomez Lira M**, Zolezzi F, Valli M, Lisi V, Freising P. Four new cases of lethal osteogenesis imperfecta due to glycine substitutions in COL1A1 and genes. Mutations in brief no. 152. Online. Hum Mutat. 1998;12(1):71-2. PubMed PMID: 10627137.
29**: Gomez-Lira M**, Perusi C, Mottes M, Pignatti PF, Uziel G, Rizzuto N, Salviati A. Two novel frameshift mutations in the adrenoleukodystrophy gene in Italian patients. J Neurol Sci. 1999 May 1;165(1):62-5. PubMed PMID: 10426149.
30: Perusi C, **Gomez-Lira M,** Mottes M, Pignatti PF, Bertini E, Cappa M, Vigliani MC, Schiffer D, Rizzuto N, Salviati A. Two novel missense mutations causing adrenoleukodystrophy in Italian patients. Mol Cell Probes. 1999 Jun;13(3):179-82. PubMed PMID: 10369742.
31: Perusi C, *Lira MG*, Duyff RF, Weinstein HC, Pignatti PF, Rizzuto N, Salviati A. Mutations associated with very late-onset metachromatic leukodystrophy. Clin Genet. 1999 Feb;55(2):130. PubMed PMID: 10189092.
32: **Gomez-Lira M**, Perusi C, Mottes M, Pignatti PF, Manfredi M, Rizzuto N, Salviati A. Molecular genetic characterization of two metachromatic leukodystrophy patients who carry the T799G mutation and show different phenotypes; description of a novel null-type mutation. Hum Genet. 1998 Apr;102(4):459-63. Erratum in: Hum Genet 1998 May;102(5):602. PubMed PMID: 9600244.
33: Perusi C, **Gomez-Lira M**, Mottes M, Pignatti PF, Rizzuto N, Salviati A. A novel mutation which represents the fifth non-pathogenic polymorphism in the coding sequence of the arylsulfatase A gene. Mol Cell Probes. 1997 Dec;11(6):449-51. PubMed PMID: 9500813.
34: **Gomez-Lira M,** Perusi C, Mottes M, Pignatti PF, Rizzuto N, Gatti R, Salviati A. Splicing mutation causes infantile Sandhoff disease. Am J Med Genet. 1998 Jan 23;75(3):330-3. PubMed PMID: 9475608.
35: **Gomez-Lira M,** Sangalli A, Mottes M, Perusi C, Pignatti PF, Rizzuto N, Salviati A. A common beta hexosaminidase gene mutation in adult Sandhoff disease patients. Hum Genet. 1995 Oct;96(4):417-22. PubMed PMID: 7557963.
36: **Gomez-Lira M**, Perusi C, Brutti N, Farnetani MA, Margollicci MA, Rizzuto N, Pignatti PF, Salviati A. A 48-bp insertion between exon 13 and 14 of the HEXB

gene causes infantile-onset Sandhoff disease. Hum Mutat. 1995;6(3):260-2. PubMed PMID: 8535449.
37: **Gomez-Lira M**, Sangalli A, Pignatti PF, Digilio MC, Giannotti A, Carnevale E, Mottes M. Determination of a new collagen type I alpha 2 gene point mutation which causes a Gly640 Cys substitution in osteogenesis imperfecta and prenatal diagnosis by DNA hybridisation. J Med Genet. 1994 Dec;31(12):965-8. PubMed PMID: 7891382; PubMed Central PMCID: PMC1016701.
38: Mottes M, Sangalli A, Valli M, Forlino A, **Gomez-Lira M**, Antoniazzi F, Constantinou-Deltas CD, Cetta G, Pignatti PF. A base substitution at IVS-19 3'-end splice junction causes exon 20 skipping in pro alpha 2(I) collagen mRNA and produces mild osteogenesis imperfecta. Hum Genet. 1994 Jun;93(6):681-7. PubMed PMID: 8005592.
39: Cohen-Solal L, Zylberberg L, Sangalli A, **Gomez Lira M**, Mottes M. Substitution of an aspartic acid for glycine 700 in the alpha 2(I) chain of type I collagen in a recurrent lethal type II osteogenesis imperfecta dramatically affects the mineralization of bone. J Biol Chem. 1994 May 20;269(20):14751-8.
40: Mottes M, **Gomez Lira MM**, Valli M, Scarano G, Lonardo F, Forlino A, Cetta G, Pignatti PF. Paternal mosaicism for a COL1A1 dominant mutation (alpha 1 Ser-415) causes recurrent osteogenesis imperfecta. Hum Mutat. 1993;2(3):196-204.
41: Mottes M, Sangalli A, Valli M, **Gomez Lira M**, Tenni R, Buttitta P, Pignatti PF, Cetta G. Mild dominant osteogenesis imperfecta with intrafamilial variability: the cause is a serine for glycine alpha 1(I) 901 substitution in a type-I collagen gene. Hum Genet. 1992 Jul;89(5):480-4..
42: Valli M, Mottes M, Tenni R, Sangalli A, **Gomez Lira M**, Rossi A, Antoniazzi F, Cetta G, Pignatti PF. A de novo G to T transversion in a pro-alpha 1 (I) collagen gene for a moderate case of osteogenesis imperfecta. Substitution of cysteine for glycine 178 in the triple helical domain. J Biol Chem. 1991 Jan 25;266(3):1872-8.
43: Bertrand E, Erard M, **Gómez-Lira M**, Bode J. Influence of histone hyperacetylation on nucleosomal particles as visualized by electron microscopy. Arch Biochem Biophys. 1984 Feb 15;229(1):395-8.
44: Bode J, **Gómez-Lira MM**, Schröter H. Nucleosomal particles open as the histone core becomes hyperacetylated. Eur J Biochem. 1983 Feb 15;130(3):437-45.
45: Schröter H, **Gómez-Lira MM**, Plank KH, Bode J. The extent of histone

acetylation induced by butyrate and the turnover of acetyl groups depend on the nature of the cell line. Eur J Biochem. 1981 Nov;120(1):21-8.
46: **Gómez-Lira MM**, Bode J. Effects of butyrate upon the metaphase-specific deacetylation of histone H4. FEBS Lett. 1981 May 18;127(2):228-32.

.