ELENCO COMPLETO DELLE PUBBLICAZIONI – *aggiornato Ottobre 2019*

(1) Gaudino G., Fasolo A., Merlo G., Lazarus L.H., Renda T., D'Este L. and Vandesande F. (1985) Active peptides from amphibian skin are also amphibian neuropeptides. **Peptides** 6, supp.3: 209-213.

(2) De Bortoli M., Theillet C., Escot C., Perroteau I., Merlo G., Lidereau R. and Callahan R. (1986) *ras* proto-oncogene in mammary cancer. In: **Endocrinology and** **Malignancy**. Iacobelli S. (ed), Parthenon Press, United Kingdom, pp. 104-111.

(3) Ohuchi N., Hand P.H., Merlo G., Fujita J., Mariani-Costantini R., Thor A., Nose M., Callahan R. and Schlom J. (1987) Enhanced expression of c-Ha-*ras*-1 p21 in stomach adeno-carcinomas defined by immunoassays using monoclonal antibodies and *in situ* hybridization. **Cancer Res.**, 47: 1413-1420.

(4) Lynch, H.T., Watson, P., Marcus, J.N., Callahan, R., Schlom, J., Merlo, G., Conway, T., Fitzsimmons, M.L. and Lynch, J. (1987) Hereditary breast cancer: a search for biomarkers **J. Tum. Marker Onc.** 2: 153-159.

(5) Mariani-Costantini, R., Escot, C., Theillet, C., Gentile, A., Merlo, G., Lidereau, R. and Callahan, R. (1988) *In situ* c-*myc* expression and genomic status of the c-*myc* locus in infiltrating ductal carcinomas of the breast. **Cancer Res.**, 48: 199-205.

(6) Ally S., Tortora G., Clair T., Grieco D., Merlo G., Katsaros D., Ogreid D., Doskeland S., Jahnsen T. and Cho-Chung Y.S. (1988) Selective modulation of protein kinase isozymes by site-selective 8-Cl-cAMP provides a biological means for control of human colon cancer cell growth. **Proc. Natl. Acad. Sci.** (USA) 85: 6319-6322.

(7) Ciardiello F., Kim N., Hynes N., Jaggi R., Redmond S., Liscia D.S., Sanfilippo B., Merlo G., Callahan R. and Salomon D. (1988) Induction of transforming growth factor-a expression in mouse mammary cells after transformation with a point-mutated c-Ha-*ras* proto-oncogene. **Mol. Endocrin.** 2: 1202-1216.

(8) Mariani-Costantini R., Theillet C., Hutzel P., Merlo G., Schlom J. and Callahan R. (1989) *In situ* detection of c-*myc* RNA in adenocarcinomas, adenomas and mucosa of human colon. **J. Histochem.** **Cytochem.**, 37: 293-298.

(9) Ali I.U., Merlo G., Lidereau R. and Callahan R. (1989) Amplification unit on chromosome 11q13 in aggressive primary human breast tumors entails *bcl*-1, *int*-2 and *hst* oncogenes. **Oncogene**, 4: 89-92.

(10) Shankar V., Ciardiello F., Kim N., Derynck R., Liscia D.S., Merlo G., Langton B.C., Sheer D., Callahan R., Bassin R., Lippman M., Hynes N. and Salomon D. (1989) Transformation of normal mouse mammary epithelial cells following transfection with a human transforming growth factor-a cDNA. **Mol.** **Carcinogenesis**, 2: 1-11.

(11) Ciardiello F., Kim N., Liscia D.S., Bianco C., Lidereau R., Merlo G., Callahan R., Greiner J., Szpak C., Kidwell W., Schlom J. and Salomon D.S. (1989) Transforming growth factor alpha (TGF-) mRNA expression in human breast and colon carcinomas and TGF- activity in the effusions of cancer patients. **J. Natl. Cancer** **Inst.**, 81: 1165-1171

(12) Mednieks M.I., Yokozaki H., Merlo G.R., Tortora G., Clair T., Ally S., Tahara E. and Cho-Chung Y.S. (1989) Site-selective 8-Cl-cAMP which causes growth inhibition and differentiation increases DNA (CRE)-binding activity in cancer cells. **FEBS Lett.**, 254: 83-88

(13) Liscia D.S., Merlo G., Garrett C., French D., Mariani-Costantini R. and Callahan R. (1989) Expression of *int*-2 mRNA in human tumors amplified for *int*-2 gene. **Oncogene**, 4: 1219-1224

(14) Ali I.U., Campbell G., Merlo G., Smith G., Callahan R. and Lidereau R. (1989) Multiple genetic alterations in human breast cancer and their possible prognostic significance. In: **Cancer Cell**, Vol 7, Cold Spring Harbor Publ., pp. 399-403

(15) Merlo G., Siddiqui J., Cropp C., Liscia D.S., Lidereau R., Callahan R. and Kufe D. (1989) Frequent alterations of the DF3 tumor-associated antigen gene in primary human breast carcinomas. **Cancer Res.**, 49: 6966-6971

(16) Mariani-Costantini R., Merlo G., and Frati L. (1989) Genomic alterations in human breast cancer: a review. **Tumori**, 75: 311-320

(17) Liscia D.S., Merlo G., Ciardiello F., Smith G., Callahan R. and Salomon D.S. (1990) Transforming growth factor-alpha (TGF-a) messanger RNA expression in the development of the rodent and human mammary gland as detected by *in situ* hybridization. **Develop. Biol.,** 140: 123-131

(18) Bieche I., Champeme M.-H., Merlo G., Larsen C.-J., Callahan R. & Lidereau R. (1990) Loss of heterozygosity of the L-*myc* proto-oncogene in human breast tumors. **Hum. Genet.**, 85: 101-105

(19) Blondel B.J., Talbot N., Merlo G.R., Wychowski C., Yokozaki H., Valverius E.M., Salomon D.S. and Bassin R.H. (1990) Efficient induction of focus formation in NIH 3T3 cells by c-*myc* and its inhibition by serum and by growth factors. **Oncogene** 5: 857-865

(20) Merlo G.R., Blondel B.J., Deed R., MacAllan D., Peters G., Dickson C., Liscia D.S., Ciardiello F., Valverius E., Salomon D.S. and Callahan R. (1990) The mouse *int*-2 gene exhibits basic fibroblast growth factor (bFGF) activity in a bFGF-responsive cell line. **Cell Growth & Diff.,** 1: 463-472

(21) Valverius E.M., Ciardiello F., Heldin N-H., Blondel B., Merlo G.R., Smith G.H., McGeady M., Stampfer M., Lippman M.E., Salomon D.S. & Dickson R. (1990) Stromal influence on transformation of human mammary epithelial cells overexpressing c-*myc* and SV40-T. **J. Cell. Physiol.**, 145: 207-216

(22) Callahan R., Cropp C.S., Merlo G., Campbell G., & Lidereau R. (1990) Mutations in breast cancer. In: "**The therapeutic implications of the molecular biology of breast cancer**" (E. Mihich and M. Lippman, eds.) J. Libby Publ. Rome. pp 57-67

(23) Merlo G.R., Cropp C.S., Callahan R. and Takahashi T. (1991) Detection of loss of heterozygosity in primary tumor DNA samples by PCR. **Biotechniques**, 11: 166-169

(24) Osborne R.J., Merlo G., Mitsudomi T., Venesio T., Liscia D.S., Cappa A.P.M., Chiba I., Takahashi T., Nau M., Callahan R. & Minna J.D. (1991) Mutations in the p53 gene in primary human breast cancers. **Cancer Res.**, 51: 6194-6198

(25) Merlo G.R., Venesio T., Bernardi A., Canale L., Gaglia P., Lauro D., Cappa A.P.M., Callahan R., Liscia D.S. (1992) Loss of heterozygosity on chromosome 17p13 in breast carcinomas identifies tumours with a high proliferation index. **Am. J. Pathol.**, 140: 215-223

(26) Venesio T., Taverna D., Hynes N., Deed R., MacAllan D., Ciardiello F., Valverius E.M., Salomon D.S., Callahan R. & Merlo G.R. (1992) The *int*-2 gene product acts as a growth factor and substitutes for basic fibroblast growth factor in promoting the differentiation of a normal mouse mammary epithelial cell line. **Cell Growth & Diff.**, 3: 63-71

(27) Callahan R., Cropp C.S., Merlo G., Liscia D., Cappa A.P.M., & Lidereau R. (1992) Somatic mutation and human breast cancer: a status report. **Cancer**, 69: 1582-1588

(28) Callahan R., Cropp C.S., Gallahan D., Liscia D.S., Merlo G., Smith G.H., & Lidereau R. (1992) The genetic pathology of breast cancer. In**: Comparative Molecular Carcinogenesis**, **Prog. Clin. Biol. Res**. 376: 117-136

(29) Callahan R., Gallahan D., Smith G., Cropp C., Merlo G., Venesio T., Liscia D., and Lidereau R. (1992) Common genetic pathways in breast oncogenesis. **Pathol. Biol**. (Paris) 39: 910-911

(30) Hynes N., NicMhuiris C., Stiefel U., Taverna D., Ball R., Happ B., Schmitt-Ney M., Groner B., Venesio T., & Merlo G. (1993) The v-*raf* and Ha-*ras* oncogenes inhibit transcription from the b-casein gene promoter by suppression of a mammary gland specific transcription factor. In: "**Hormonal Carcinogenesis**" (J.J. Li, S. Nandi and S.A. Li, eds.) Springer Verlag Publ., pp. 164-171

(31) Marchetti A., Buttitta F., Merlo G., Diella F., Pellegrini S., Pepe S., Macchiarini P., Angeletti C.A., Callahan R., Bistocchi M., & Squartini F. (1993) p53 alterations in non small cell lung cancers correlate with metastatic involvement of hilar/mediastinal lymph-nodes and advanced stages of disease. **Cancer Res.**, 53: 2846-2851

(32) Merlo G.R., Bernardi A., Diella F., Venesio T., Cappa A.P.M., Callahan R., & Liscia D.S. (1993) In primary human breast cancer mutations in exons 5 and 6 of the p53 gene are associated with a high S-phase index. **Int. J. Cancer**, 54: 531-535

(33) Merlo G.R., Venesio T., Taverna D., Callahan R., and Hynes N. (1993) Growth suppression of normal mammary epithelial cells by wild-type p53. **Ann. N.Y. Acad. Sci.** (USA) 698: 108-113

(34) Liscia D.S., Venesio T., Diella F., Canale L., Bernardi A., Cappa A.P.M., Callahan R. and Merlo G.R. (1993) A locus on chromosome 17p13.3 associated with a high S-phase index is distinct from the p53 gene in breast cancer. **Ann. N.Y. Acad. Sci.** (USA) 698: 120-125

(35) Callahan R., Gallahan D., Smith G., Cropp C.S., Merlo G.R., Diella F. & Lidereau R. (1993) Frequent mutations in breast cancer. **Ann. N.Y. Acad. Sci.** (USA) 698: 21-31

(36) Diella F., Normanno N., Merlo G.R., Salomon D.S. & Callahan R. (1993) Absence of p53 point mutations in non transformed human mammary epithelial cell lines. **Life Sci Adv-Biochem.,** 12: 47-51

(37) Callahan R., Cropp C.S., Merlo G.R., Diella F., Venesio T., Lidereau R., Cappa A.P.M. and Liscia D.S. (1993) Genetic and molecular heterogeneity of breast cancer cells. **Clin. Chim. Acta**, 217: 63-73

(38) Callahan R., Cropp C., Sheng Z.M., Merlo G., Steeg P., Liscia D. and Lidereau R. (1993) Definition of regions of the human genome affected by loss of heterozygosity in primary human breast tumors. **J. Cell Biochem**. Suppl. 17G: 167-172

(39) Thor A.D., Salomon D.S., Merlo G., Liscia D.S., Lidereau R., Callahan R., Schlom J. and Ali I.U. (1994) Genetic abnormalities in breast carcinoma. **Surg. Pathol.**, 5: 331-348

(40) Merlo G.R., Venesio T., Taverna D., Marte B., Callahan R., and Hynes N. (1994) Growth suppression of normal mammary epithelial cells by wild-type p53. **Oncogene**, 9: 443-453

(41) Bianco C., Tortora G., Basolo F., Fiore L., Fontanini G., Merlo G., Salomon D.S., Bianco A.R., & Ciardiello F. (1994) Effect of mutant p53 genes on transformation of human mammary epithelial cells. **Int. J. Oncol.**, 4: 1077-1082

(42) Basolo F., Venesio T., Calvo S., Fiore L., Fontanini G., Toniolo A., Liscia D.S. & Merlo G.R. (1994) The effect of *Fgf*-3/*int*-2 on growth and transformation of MCF-10A normal human mammary epithelial cells is distinct from FGF-1 and FGF-2. **Int. J. Oncol.**, 4: 1365-1370

(43) Qi, C.-F., Liscia, D.S., Normanno, N., Merlo, G.R., Johnson, G.R., Gullick, W.J., Ciardiello, F., Saeki, T., Brandt, R., Kim, N. & Salomon, D.S. (1994) Expression of transforming growth factor-a, amphiregulin and cripto-1 in human breast carcinomas. **Br. J. Cancer**, 69: 903-910

(44) Tortora G., Budillon A., Yokozaki H., Clair T., Pepe S., Merlo G., Rohlff C., and Cho-Chung Y.S. (1994) Retroviral vector-mediated overexpression of the RIIb subunit of the cAMP-dependent protein kinase induces differentiation in human leukemia cells and reverts the transformed phenotype of mouse fibroblasts. **Cell Growth & Diff.**, 5: 753-759

(45) Merlo G.R., Venesio T., Bernardi A., Cropp C.S., Diella F., Cappa A.P.M., Liscia D.S. & Callahan R. (1994) Evidence for a second tumor suppressor gene on chromosome 17p linked to high S-phase index in primary human breast carcinomas. **Cancer Genet. & Cytogenet.**, 76: 106-111

(46) Fontanini G., Vignati S., Bigini D., Ribecchini A., Angeletti C.A., Merlo G.R., Basolo F., Pingitore R., and Bevilacqua G. (1994) over-expression of the p53 protein is maintained throughout progression of human non small cell lung cancer: relation with the proliferative activity. **J. Pathol.**, 174: 23-31

(47) Merlo G.R. and Hynes N.E. (1994) Cooperation between mutant p53 and the *ras*, *raf*, *erb*B-2 and *fgf*-3 oncogenes for transformation of mammary epithelial cells. **Int. J. Oncol.**, 5: 1141-1150

(48) Fontanini G., Fiore L., Bigini D., Vignati S., Calvo S., Mussi A., Angeletti C.A., Merlo G.R. and Basolo F. (1994) Levels of p53 antigen in the serum of non small cell lung cancer patients correlate with positive p53 immunohistochemistry on tumor sections, tumor necrosis and nodal involvement. **Int. J. Oncol.** 5: 553-558

(49) Merlo G.R., Basolo F., Fiore L., Duboc L. and Hynes N.H. (1995) p53-dependent and p53-independent activation of apoptosis in mammary epithelial cells reveals a survival function for EGF and insulin. **J. Cell Biol.** 128: 1185-1196

(50) Marchetti A., Merlo G.R., Buttitta F., Callahan R., Bistocchi M. and Squartini F. (1995) Detection of DNA mutations in acid formalin-fixed paraffin-embedded archival specimens by polymerase chain reaction-single strand conformation polymorphism analysis. **Cancer Detect. Prev.** 19: 278-281

(51) Budillon A., Cereseto A., Kondrashin A., Nesterova M., Merlo G., Clair T. and Cho-Chung Y. S. (1995) Point mutation of the autophosphorylation site or in the nuclear location signal causes protein kinase A RIIb regulatory subunit to lose its ability to revert transformed fibroblasts. **Proc. Natl. Acad. Sci.** USA, 92: 10634-10638

(52) Damiano V, Diisernia G, Ciardiello F, Pepe S, Bianco A, Bianco C, Ruggiero A, Baldassarre G, Merlo G, Tortora G. (1995) Overexpression of wild-type p53 overrides the mitogenic effect of RI-alpha subunit of protein-kinase-a in human breast cells. **Int J Oncol.** 7(2): 331-336. PubMed PMID: 21552843.

(53) Marchetti A., Buttitta F., Pellegrini S., Merlo G., Chella A., Angeletti A., Bistocchi M. and Bevilacqua G. (1995) *mdm*-2 gene amplification and overexpression in non-small cell lung carcinomas with accumulation of the p53 protein in the absence of p53 gene mutations. **Diagnostic Mol. Pathol.** 4: 93-97

(54) Marchetti A., Doglioni C., Barbareschi M., Buttitta F., Pellegrini S., Bertacca G., Chella A., Merlo G., Angeletti C.A., Dalla Palma P., and Bevilacqua G. (1996) p21 RNA and protein expression in non-small cell lung carcinomas: evidence of p53-independent expression and association with tumoral differentiation. **Oncogene**, 12: 1319-1324

(55) Merlo G.R., Graus-Porta D., Cella N., Marte B., Taverna D. and Hynes N.E. (1996) Growth, differentiation and survival of HC11 mammary epithelial cells: diverse effects of receptor tyrosine kinase-activating peptide growth factors. **Eur. J. Cell Biol.,** 70: 97-105

(56) Basolo, F., Fiore, L., Calvo, S., Falcone, V., Conaldi, P.G., Fontanini, G., Caligo, A.M., Merlo, G., Gluzman, Y., Toniolo, A. (1996) Defective interleukin six expression and responsiveness in human mammary cells transformed by an adeno 5/SV40 hybrid virus. **Br. J. Cancer**. 73: 1356-1361

(57) Merlo G.R., Cella N. and Hynes N.E. (1997) Apoptosis is accompanied by changes in Bcl-2 and Bax expression, induced by loss of attachment, and inhibited by specific extracellular matrix proteins in mammary epithelial cells. **Cell Growth & Diff.**, 8: 251-260

(58) Merlo G.R., Fiore L., Basolo F., Woods-Cook K. and Hynes N.E. (1997) In mammary epithelial cells p53-mediated apoptosis in response to DNA damage is dependent on the agent and can be influenced by growth factors. **Endocrine-Related Cancer**, 4: 55-66

(59) Cipollini G., Beri A., Fiore L., Rainaldi G., Basolo F., Merlo G., Bevilacqua G., and Caligo M.A. (1997) Down regulation of nm23 H1 gene inhibits cell proliferation. **Int. J. Cancer**, 73: 297-2302

(60) Topilko P., Levi G., Merlo G., Mantero S., Desmarquet C., Mancardi G. and Charnay P. (1997) Differential regulation of the zinc finger genes *Krox-20* and *Krox-24* (*Egr-1*) suggests antagonistic roles in Schwann cells. **J. Neurosci. Res**., 50: 702-712

(61) Marchetti A., Buttitta F., Carnicelli V., Pellegrini S., Bertacca G., Merlo G., and Bevilacqua G. (1997) Enriched SSCP: a highly sensitive method for the detection of unknown mutations. Application to the molecular diagnosis of lung cancer in sputum samples. **Diagn. Mol. Pathol**., 6(4): 185-191

(62) Goula D., Benoist C., Mantero S., Merlo G., Levi G., and Demeneix B. (1998) Polyethyleneimine-based intravenous delivery of transgene to mouse lung**. Gene Therapy**, 5: 1291-1295

(63) Marchetti A. Doglioni C., Barbareschi M., Buttitta F., Pellegrini S., Gaeta P., La Rocca R., Merlo G., Chella A., Angeletti C.A., Dalla Palma P. and Bevilacqua G. (1998) Cyclin D1 and retinoblastoma susceptibility gene alterations in non-small cell lung cancer. **Int. J. Cancer**, 75: 187-192

(64) Basolo F., Fiore L., Fusco A., Giannini R., Albini A., Merlo G.R., Fontanini G., Conaldi P.G. and Toniolo A. (1999) Potentiation of the malignant phenotype of the undifferentiated ARO thyroid cell line by insertion of the bcl-2 gene. **Int. J. Cancer**, 81: 956-962

(65) Acampora D., Merlo G.R., Paleari L., Zerega B., Mantero S., Barbieri O., Postiglione M.P., Simeone A. & Levi G. (1999) Craniofacial, vestibular and bone defects in mice lacking the *Distal-less*-related gene *Dlx*5. **Development** 126: 3795-3809

(66) Merlo G., Zerega B., Paleari L., Trombino S., Mantero S., and Levi G. (2000) Multiple function of *Dlx* genes. **Int. J. Develop. Biol.** 44 (6° num. spec.): 619-626

(67) Pfeffer U., Ferro P., Pavia V., Trombino S., Dell'Eva R., Merlo G.R. and Levi G. (2000) The coding region of the human *DLX6* gene contains a polymorphic CAG/CCG repeat. **Int. J. Oncol**. 18: 1293-1297

(68) Charite J., McFadden D.G., Merlo G.R., Levi G., Clouthier, D.E., Yanagisawa, M., Richandson, J.A., and Olson, E. (2001) Role of Dlx6 in regulation of an endothelin-1-dependent, dHAND branchial arch enhancer. **Genes Develop**. 15: 3039-3049

(69) Merlo G.R., Paleari L., Mantero S., Genova F., Beverdam A., Palmisano G.L., Barbieri O. and Levi G. (2002) A mouse model of Split Hand/Foot Malformation Type I. **Genesis** 33: 97-101

(70) Merlo G.R., Paleari L., Mantero S., Zerega B.,Adamska M., Rinkwitz S., BoberE. and Levi G. (2002) The *Dlx5* homeobox gene is essential for vestibular morphogenesis in the mouse embryo through a BMP4-mediated pathway. **Develop. Biol**. 248: 157-169

(71) Beverdam, A., Merlo, G.R., Paleari, L., Mantero, S., Genova, F., Barbieri, O., Janvier, P. and Levi, G. (2002) Jaw transformation with gain of symmetry after *Dlx5/Dlx6* inactivation: mirror of the past ? **Genesis,** 34: 221-227

(72) Levi G., Puche A.C., Mantero S., Barbieri O., Trombino S., Paleari L., Egeo A. and Merlo, G.R. (2003) The *Dlx5* homeodomain gene is essential for normal olfactory development and connectivity in the mouse. **Mol. Cell. Neurosci**., 22: 530-543

(73) Merlo G.R, Beverdam A. and Levi G. (2003) *Dlx* genes in craniofacial and limb morphogenesis. in: "Murine Homeobox Gene Control of Embryonic Patterning and Organogenesis" Chap.4. (T. Lufkin, ed). **Adv. Develop. Biol. Biochem**. 13: 107-132

(74) Perera M., Merlo G.R., Verardo S., Paleari L., Corte G. and Levi, G. (2004)Defective neurogenesis in the absence of *Dlx5.* **Mol. Cell. Neurosci.** 25: 153-161

(75) Levi G., Mantero S., Barbieri O., Cantatore D., Paleari L., Beverdam A., Genova F., Robert B. and Merlo G.R. (2006) *Msx1* and *Dlx5* act independently in development of craniofacial skeleton, but converge on the regulation of Bmp signaling in palate formation. **Mech. Develop**. 123: 3-16

(76) Meneghini V., Odent S., Platonova N., Egeo A. and Merlo G.R. (2006) Novel *TBX3* mutation data in families with Ulnar-Mammary syndrome indicate a genotype-phenotype relationship: mutations that do not disrupt the T-domain are associated with less severe limb defects. **Eur. J. Med. Genet**. 49: 151-158

(77) Zaghetto A.A., Paina S., Mantero S., Peretto P., Bovetti, S., Puche A., Piccolo S., and Merlo G. (2006) A *Wnt*-beta-catenin responsive cell population that participates in the formation of olfactory axon connections in the mouse embryo. **Int. J. Dev. Neurosci**. 24: 584-585

(78) Radoja N., Guerrini L., LoIacono N., Merlo G.R., Costanzo A., Weinberg W.C., LaMantia G., Calabrò V., and Morasso M.I. (2007) Homeobox gene *Dlx3* is regulated by *p63* during ectoderm development: relevance in the pathogenesis of ectodermal dysplasias. **Development** 134: 13-18

(79) Platonova N., Scotti M., Babich P., Bertoli G., Zucchi I., and Merlo G. (2007) Tbx3, the Ulnar-Mammary Syndrome gene, plays role in mammary cell proliferation independently of p19ARF and p53. **Eur. J. Hum. Genet**. 14 (suppl 1): 281

(80) Platonova N., Scotti M., Babich P., Bertoli G, Mento E., Meneghini V., Egeo A., Zucchi I. and Merlo G.R. (2007) The *TBX3* gene, mutated in Ulnar-Mammary syndrome, promotes growth of mammary epithelial cells independently of *ARF* and *p53*. **Cell Tissue Res.** 328: 301-316

(81) Vieux-Rochas M., Coen L., Sato T., Kurihara Y., Gitton Y., Barbieri O., Le Blay K., Merlo G.R., Ekker M., Kurihara H., Janvier P. and Levi G. (2007) Molecular dynamics of retinoic acid-induced craniofacial malformations: implications for the origin of the gnatostome jaw. **PLoS One**, Vol 2 (issue 6): e510

(82) Merlo G.R., Mantero S., Zaghetto A.A., Peretto P., Paina S. and Gozzo M. (2007) The role of Dlx homeogenes in early development of the olfactory pathway. **J. Mol. Histol.** (special issue) 38(6): 612-623 *corrected and republished from J. Mol. Histol 38(4): 347-358*

(83) Zaghetto A.A., Paina S., Mantero S., Platonova N., Peretto P., Bovetti S., Puche A.C., Piccolo S. and Merlo G.R. (2007) Activation of the *Wnt*-catenin pathway in a cell population on the surface of the forebrain is essential for the establishment of olfactory axon connections. **J. Neurosci.** 27: 9757-9768 PMID: 17804636; PubMed Central PMCID: PMC1986640.

 (84) LoIacono N., Mantero S., Chiarelli A., Garcia E., Mills A.A., Morasso M.I., Costanzo, A., Levi, G., Guerrini L. and Merlo G.R. (2008) Regulation of Dlx5 and Dlx6 gene expression by p63 is involved in EEC and SHFM congenital limb defects. **Development** 135: 1377-1388

(85) Lopardo T., LoIacono N., Marinari B., Giustizieri M.L., Cyr D.G., Merlo G., Crosti F., Costanzo A. and Guerrini L. (2008) Claudin-1 is a p63 target gene with a crucial role in epithelial development. **PLoS One**, Vol 3 (issue 7): e2715.

(86) Merlo G.R., Paina S., Moiana A., Cattaneo E., DeMarchis S., Corte G. (2008) A Dlx5-Wnt5a regulation involved in the control of GABAergic differentiation. **Int. J. Dev. Neurosci.** 26(8): 877

(87) Moretti F., Marinari B., LoIacono N., Botti E., Giunta A., Spallone G., Garaffo G., Vernersson-Lindhal E., Merlo G.R., Mills A.A., Ballarò C., Alemà S., Chimenti S., Guerrini L. and Costanzo A. (2010) A regulatory feed-back loop involving p63 and IRF6 links the pathogenesis of two genetically different ectodermal dysplasias. **J. Clin. Invest**. 120(5): 1570–1577

(88) Vieux-Rochas M., Mantero S., Heude E., Barbieri O., Astigiano S., Couly G., Kurihara H., Levi G. and Merlo G.R. (2010) Spatio-temporal dynamics of gene expression of the Edn1-Dlx5/6 pathway during development of the lower jaw. **Genesis**, 48: 362-373

(89) Paina S., Garzotto D., DeMarchis S., Moiana, A., Cattaneo, E., Conti L., Perera M., Corte, G., Calautti E. and Merlo G.R. (2011) *Wnt5a* is a transcriptional target of *Dlx* genes and promotes differentiation of olfactory interneuron progenitors. **J. Neurosci.** 31: 2675-2687 PubMed PMID: 21325536.

(90) Guerrini L., Costanzo A. and Merlo G.R. (2011) A symphony of regulations centered on p63 to control development of ectoderm-derived structures. **J. Biomed. Biotech**. Vol 2011, pag. 1-13 (invited review).

(91) Merlo GR, Altruda F. and Poli V. (2012) Mice as Experimental Organisms. In: **Encyclopedia of Life Science,** John Wiley & Sons, Ltd: Chichester. UK. DOI: 10.1002/9780470015902.a0002029.pub2

(92) Chiabrando D., Marro S., Mercurio S., Giorgi C., Petrillo S., Vinchi F., Fiorito V., Fagoonee S., Camporeale A., Turco E., Merlo G.R., Silengo L., Altruda F., Pinton P. and Tolosano E. (2012) The mitochondrial isoform of the heme exporter FLVCR1 controls erythroid differentiation by exporting heme from mitochondria. **J. Clin. Invest**. 122(12): 4569-4579. doi: 10.1172/JCI62422.

(93) Vieux-Rochas M, Bouhali K, Mantero S, Garaffo G, Provero P, Barbieri O, Caratozzolo M, Tullo A, Lallemand Y, Robert B, Levi G, Merlo G.R. (2013) Bmp-mediated functional cooperation between *Dlx5;Dlx6* and *Msx1;Msx2* during mammalian limb development. **PLoS One**, 8: e51700. doi:10.1371/journal.pone.0051700

(94) Garaffo G, Provero P., Molineris I, Pinciroli P, Peano C., Battaglia C., Tomaiuolo D., Etzion T., Gothilf Y., Santoro M., Merlo G.R. (2013) Profiling, bioinformatic and functional data on the developing olfactory/GnRH system reveal cellular and molecular pathways essential for this process and potentially relevant for the Kallmann syndrome**. Front. Endocrinol**. 4:203. doi: 10.3389/fendo.2013.00203. PMID: 24427155

(95) Franco I., Gulluni F., Campa C.,Costa C., Margaria J.P., Ciraolo E., Martini M, Monteyne D., De Luca E., Germena G., Posor Y.,Maffucci T., Marengo S., Haucke V., Falasca M., Perez-Morga P., Boletta A., Merlo G.R., Hirsch E. (2014) PI3K class II α controls spatially restricted endosomal PtdIns3P and Rab11 activation to promote primary cilium function. **Develop. Cell,** 28(6): 647-658.

 doi: 10.1016/j.devcel.2014.01.022. PubMed PMID: 24697898.

(96) Restelli, M., Lopardo T., Lo Iacono N., Garaffo G., Conte D., Rustighi A., Napoli M., Del Sal G., Perez-Morga D., Costanzo A., Merlo G.R. and Guerrini L. (2014) *DLX5, FGF8* and the *Pin1* isomerase control Np63 protein stability during limb development: a regulatory loop at the basis of the SHFM and EEC congenital malformations. **Hum. Mol. Genet.** 23: 3830-3842. doi: 10.1093/hmg/ddu096 PubMed PMID: 24569166

(97) [Bonomi M](http://www.ncbi.nlm.nih.gov/pubmed/?term=Bonomi%20M), [Cappa M](http://www.ncbi.nlm.nih.gov/pubmed/?term=Cappa%20M), [Cariboni A](http://www.ncbi.nlm.nih.gov/pubmed/?term=Cariboni%20A), [Di Schiavi E](http://www.ncbi.nlm.nih.gov/pubmed/?term=Di%20Schiavi%20E), [Fabbri A](http://www.ncbi.nlm.nih.gov/pubmed/?term=Fabbri%20A), [Ferlin A](http://www.ncbi.nlm.nih.gov/pubmed/?term=Ferlin%20A), [Foresta C](http://www.ncbi.nlm.nih.gov/pubmed/?term=Foresta%20C), [Ghizzoni L](http://www.ncbi.nlm.nih.gov/pubmed/?term=Ghizzoni%20L), [Jannini E](http://www.ncbi.nlm.nih.gov/pubmed/?term=Jannini%20E), [Krausz C](http://www.ncbi.nlm.nih.gov/pubmed/?term=Krausz%20C), [Loche S](http://www.ncbi.nlm.nih.gov/pubmed/?term=Loche%20S), [Lombardo F](http://www.ncbi.nlm.nih.gov/pubmed/?term=Lombardo%20F), [Maggi M](http://www.ncbi.nlm.nih.gov/pubmed/?term=Maggi%20M), [Maggi R](http://www.ncbi.nlm.nih.gov/pubmed/?term=Maggi%20R), [Maghnie M](http://www.ncbi.nlm.nih.gov/pubmed/?term=Maghnie%20M), [Mancini A](http://www.ncbi.nlm.nih.gov/pubmed/?term=Mancini%20A), [Merlo G](http://www.ncbi.nlm.nih.gov/pubmed/?term=Merlo%20G), [Panzica G](http://www.ncbi.nlm.nih.gov/pubmed/?term=Panzica%20G), [Radetti G](http://www.ncbi.nlm.nih.gov/pubmed/?term=Radetti%20G), [Russo G](http://www.ncbi.nlm.nih.gov/pubmed/?term=Russo%20G), [Simoni M](http://www.ncbi.nlm.nih.gov/pubmed/?term=Simoni%20M), [Sinisi AA](http://www.ncbi.nlm.nih.gov/pubmed/?term=Sinisi%20AA), [Persani L](http://www.ncbi.nlm.nih.gov/pubmed/?term=Persani%20L). (2014) Kallmann’s syndrome and normosmic isolated hypogonadotropic hypogonadism: two largely overlapping manifestations of one rare disorder. **J. Endocrin Invest**. 37(5): 499-500 doi: 10.1007/s40618-014-0063-z

(98) Molineris I., Santoro R., Garaffo G., Provero P. and Merlo G.R. (2014) The contribution of developmental biology to human genetics in the era of next-gen genome sequencing: what are we learning from studying the Kallmann syndrome? **Trends Dev Biol.** 8: 97-127

(99) Garaffo G., Conte D., Provero P., Tomaiuolo D., Luo Z., Pinciroli P, Peano C., D’Atri I., Gitton Y., Etzion E., Gothilf Y., Gays D., Santoro M.M., Merlo G.R. (2015) The Dlx5 and Foxg1 transcription factors, linked via miRNA-9 and -200, are required for the development of the olfactory and GnRH system. **Mol. Cell. Neurosci.**  68: 103-119. doi: 10.1016/j.mcn.2015.04.007PubMed PMID: 25937343; PubMed Central PMCID: PMC4604252.

(100) Restelli M., Marinari B., Gnesutta N., Conte D., Merlo G.R., Costanzo A., Guerrini L. (2015) FGF8, c-Abl and p300 participate in a pathway that controls stability and function of the ΔNp63α protein. **Hum** **Mol Genet**, 24(15): 4185-4197. doi: 10.1093/hmg/ddv151. PMID: 25911675

(101) Conte D., Guerrini L. and Merlo G.R. (2015) Novel cellular and molecular interactions during limb development, revealed from studies on the Split Hand-Foot congenital malformation. Book Chapter **InTech, Open Science**. Embryology, ISBN 978-953-51-4214-0. *in press.*

# (102) Franco I., Margaria J.P., De Santis M.C., Ranghino A., Monteyne D., Chiaravalli M., Pema M., Campa C.C., Ratto E., Gulluni F., Perez-Morga P., Somlo S., Merlo G.R., Boletta A. and Hirsch E. (2015) Phosphoinositide 3-Kinase-C2α Regulates Polycystin-2 Ciliary Entry and Protects against Kidney Cyst Formation. J. Am Soc. Nephrol., pii: ASN.2014100967*.*

(103) Grosso A, Cambiaghi M, Renna A, Milano L, Merlo GR, Sacco T, Sacchetti B. (2015) The higher order auditory cortex is involved in the assignment of affective value to sensory stimuli. **Nat. Commun.** 6:8886. doi: 10.1038/ncomms9886. PubMed PMID: 26619940

(104) Merlo GR., Armentano M, Zamboni V, Sarò G, Berto G, Ciraolo E, Ghigo A, Passafaro M, Carabelli V, Gavello D, El-Assawi N, Mauro A, Priano L, Hirsch E (2015) Hyper-activation of the Rho-GTPase Rac1 via disruption of ArhGAP15 results in reduced architectural and functional complexity. **Int J Dev Neurosci** 47(Pt A): 28-29. doi: 10.1016/j.ijdevneu.2015.04.083

(105) Conte D, Garaffo G, Lo Iacono N, Mantero S, Piccolo S, Cordenonsi D, Perez-Morga D, Orecchia V, Poli V and Merlo GR (2015) The Apical Ectodermal Ridge of *Dlx5;Dlx6*-/- ectrodactylous limbs shows altered Wnt5a expression and planar-cell polarity pathway, rescued by exogenous Wnt5a ligand. **Hum Mol Genet.** 25(4): 740-754. doi: 10.1093/hmg/ddv514. PubMed PMID: 26685160; PubMed Central PMCID: PMC4743692

 (106) Bassi I., Andre` V, Marelli F., Vezzoli V, Merlo G.R, Cariboni A, Persani L, Gothilf Y, Bonomi M. (2016)Zebrafish as an emerging animal model for investigating the hypothalamic regulation of reproduction. **Minerva Medica**, 41(2): 250-265 Epub 2016 Mar 2. PubMed PMID: 26934719

(107) Grosso A., Cambiaghi M., Renna A., Milano L., Merlo G.R., Sacco T., Sacchetti B. (2015) Value coding neurons in lateral amygdala are recruited by both unconditioned and learned emotional stimuli. **45th annual meeting of the Society for Neuroscience**, Chicago USA, 17-21/10/2015 http://sfn15.hubbian.com/id\_11327

# (108) Zamboni V., Armentano M., Sarò G., Ciraolo E., Ghigo A., Germena G., Umbach A., Valnegri P., Passafaro M., Carabelli V., Gavello D., Bianchi V., D’Adamo P, DeCurtis I., El-Assawi N., Mauro A., Priano L., Hirsch E. and Merlo G.R. (2016) Disruption of *ArhGAP15* results in hyperactive Rac1, affects the architecture and function of hippocampal interneurons and causes cognitive deficits. Scientific Report 6: 34877. doi: 10.1038/srep34877

##### (109) Palagano E., Zuccarini G., Mantero S., Conte D., Angius A., Uva P., Prontera P., Vezzoni P., Villa A., Merlo G., Sobacchi C. (2016) NBAS is the gene mutated in two patients affected by Acrofrontofacionasal Dysostosis type 1 04/2016; DOI:10.1530/boneabs.5.P244

(110) Cottone E., D'Atri I., Conte D., Pomatto V., Gothilf Y., Santoro M., Merlo G., and Bovolin P. (2016) The endocannabinoid system regulates zebrafish GnRH neuronal development. 28th Conference of European Society for Comparative Endocrinology ESCE, Leuven, Belgio 21-25 August 2016, pag 273

(111) Petrillo S, Chiabrando D, Genova T, Fiorito V, Ingoglia G, Vinchi F, Mussano F., Carossa S, Silengo L, Altruda F, Merlo G.R., Munaron L, Tolosano E. (2018) Heme accumulation in endothelial cells impairs angiogenesis by triggering paraptosis. **Cell Death Differ.** 25(3): 573-588. doi: 10.1038/s41418-017-0001-7. PMID:29229999

(112) Zamboni V, Armentano M, Berto G, Ciraolo E, Ghigo A, Garzotto D, Umbach A, DiCunto F, Parmigiani E, Boido M, Vercelli A, El-Assawi N, Mauro A, Priano L, Ponzoni L, Murru L, Passafaro M, Hirsch E, Merlo GR. (2018) Hyperactivity of Rac1-GTPase pathway impairs neuritogenesis by altering actin dynamics. **Scientific Report,** 8: 7254. doi: 10.1038/s41598-018-25354-3*.* PMID 29740022

(113) Palagano E, Zuccarini G, Prontera P, Borgatti R, Stangoni G, Elisei S, Mantero S, Menale C, Forlino A, Uva P, Oppo M, Vezzoni P, Villa A, Merlo GR, Sobacchi C. (2018) Mutations in the Neuroblastoma Amplified Sequence gene in a family affected by Acrofrontofacionasal Dysostosis type 1. **Bone** 114:125-136. doi: 10.1016/j.bone.2018.06.013

(114) Zamboni V, Jones R, Umbach A, Ammoni A, Passafaro M, Hirsch E and Merlo GR (2018) Rho GTPases in intellectual disability: from genetics to therapeutic opportunities. **Int. J. Molecular Sciences** 19(6) pii: E1821. doi: 10.3390/ijms19061821*.* PubMed PMID: 29925821

(115) Grassi E, Santoro R, Umbach A, Grosso A, Oliviero S, Neri F, Conti L, Ala U, Provero P, DiCunto F, Merlo GR (2019) Choice of alternative polyadenylation sites, mediated by the RNA-binding protein Elavl3, plays a role in differentiation of inhibitory neuronal progenitors. **Front. Cell. Neurosci.** 12:518. doi: 10.3389/fncel.2018.00518

(116) Frisina F., Valetti G., Zuccarini G., Conti L., Merlo G.R. (2019)Advances in the use of GABAergic interneurons for the treatment of epilespia. **J. Stem Cells Ther. Transplant.** 3: 009-022. DOI:10.29328/journal.jsctt.1001014

(117) Messina A., Pulli K., Santini S., Acierno J., Känsäkoski J., Cassatella D., Xu C., Casoni F., Malone S.A., Ternier G., Conte D., Sidis Y., Tommiska J., Vaaralahti K., Dwyer A., Gothilf Y., Merlo G.R., Santoni F., Niederländer N.J., Giacobini P., Raivio T., Pitteloud N. (2019) Neuron-derived neurotrophic factor (*NDNF*) is mutated in patients with Congenital Hypogonadotropic Hypogonadism, **Am. J. Hum Genet**, *in press.*

# (1118) Zuccarini G, D’Atri I, Cottone E, Mackie K., Gothilf Y, Provero P, Bovolin P and Merlo GR (2019) Interference with the cannabinoid receptor CB1 results in miswiring of GnRH+ and AgRP1+ axons in zebrafish embryos. Int. J. Molecular Sci, special issue “Endocannabinoid System in Health and Disease: Current Situation and Future Perspective*s”, submitted.*

(119) Russo I, Zamboni V, Ammoni A, Morellato A, Cimino I, Alfieri A, Angelini C, Giacobini P, Merlo G.R., Turco E, and Defilippi P (2019) The scaffold protein p140Cap controls female fertility in mice, acting early postnatally on the maturation and survival of GnRH neurons. **NeuroEndocrinology**, *in preparation.*

(120) Conti L., Zassi, Merlo GR, Umbach A. (2019) Human progenitor cells to model differentiation of GABAergic inhibitory neurons, *in preparation.*

 (121) Zamboni V, Umbach A, Ghigo A, Sarò G., Parmigiani E, Yanagawa Y, Obata K, Ponzoni L, Murru L, Passafaro M, Hirsch E. and Merlo GR. (2020) Hyperactive Rac1 GTPase affects the tangential-to-radial switch of migrating interneurons and results in reduced inhibition and epilepsy. *in preparation.*

(122) Villa C, Umbach A, Riganti C, Legato M., MonguzziA, Boido M., Merlo G.R., Torrente Y. (2020) Treatment with ROS detoxifying gold quantum clusters alleviates the progressive functional decline in the mouse model of Friedreich’s Ataxia. *in preparation.*

(123) Ammoni A, Zamboni V, Camacho-Leal P, Morello N, Giacobini P, Cabodi S. and Merlo G.R. (2020) The adaptor protein *p130cap* is required for the migration of GnRH neurons via the sialylation of the adhesion molecule NCAM on olfactory axons. *in preparation.*