

PRODUZIONE SCIENTIFICA – ARTICOLI SELEZIONATI

: Romeo G, Ronchetto P, Luo Y, Barone V, Seri M, Ceccherini I, Pasini B, Bocciardi R, Lerone M, Kääriäinen H, and Martucciello G. Point mutations affecting the tyrosine kinase domain of the RET proto-oncogene in Hirschsprung's disease. *Nature*. 1994 Jan 27;367(6461):377-8.

: Avolio L, Rinaldi A, Serafini G, Martucciello G. Endocavitary Electrocardiography during central vein catheter positioning in a newborn with persistent left superior vena cava. *J Vasc Access*. 2009 Jul-Sep;10(3):212-3.

: Avolio L, Martucciello G. Images in clinical medicine. Ingested magnets. *N Engl J Med*. 2009 Jun 25;360(26):2770.

: Borghini S, Duca MD, Pini Prato A, Lerone M, Martucciello G, Jasonni V, Ravazzolo R, Ceccherini I. Search for pathogenetic variants of the SPRY2 gene in intestinal innervation defects. *Intern Med J*. 2009 May;39(5):335-7.

: Martucciello G. Hirschsprung's disease, one of the most difficult diagnoses in pediatric surgery: a review of the problems from clinical practice to the bench. *Eur J Pediatr Surg*. 2008 Jun;18(3):140-9. Review.

: Pini Prato A, Gentilino V, Giunta C, Avanzini S, Mattioli G, Parodi S, Martucciello G, Jasonni V. Hirschsprung disease: do risk factors of poor surgical outcome exist? *J Pediatr Surg*. 2008 Apr;43(4):612-9.

: Romano P, Avolio L, Martucciello G, Steyaert H, Valla JS. Adrenal masses in children: the role of minimally invasive surgery. *Surg Laparosc Endosc Percutan Tech.* 2007 Dec;17(6):504-7.

: Pini Prato A, Gentilino V, Giunta C, Avanzini S, Parodi S, Mattioli G, Martucciello G, Jasonni V. Hirschsprung's disease: 13 years' experience in 112 patients from a single institution. *Pediatr Surg Int.* 2008 Feb;24(2):175-82.

: Martucciello G, Brizzolara A, Favre A, Lombardi L, Bocciardi R, Sanguineti M, Pini Prato A, Jasonni V. Neural crest neuroblasts can colonise aganglionic and ganglionic gut in vivo. *Eur J Pediatr Surg.* 2007 Feb;17(1):34-40.

: Martucciello G, Luinetti O, Romano P, Magrini U. [Molecular biology, basic research and diagnosis of Hirschsprung's disease]. *Pathologe.* 2007 Mar;28(2):119-24. Review. German.

: Pini-Prato A, Avanzini S, Gentilino V, Martucciello G, Mattioli G, Coccia C, Parodi S, Bisio GM, Jasonni V. Rectal suction biopsy in the workup of childhood chronic constipation: indications and diagnostic value. *Pediatr Surg Int.* 2007 Feb;23(2):117-22.

: Pini-Prato A, Martucciello G, Jasonni V. Rectal suction biopsy in the

diagnosis of intestinal dysganglionoses: 5-year experience with Solo-RBT in 389 patients. *J Pediatr Surg*. 2006 Jun;41(6):1043-8.

: Avolio L, Karmarkar SJ, Martucciello G. Complete penoscrotal transposition. *Urology*. 2006 Jun;67(6):1287. Epub 2006 May 12.

: Martucciello G, Lombardi L, Savasta S, Gibbons RJ. Gastrointestinal phenotype of ATR-X syndrome. *Am J Med Genet A*. 2006 Jun 1;140(11):1172-6.

: Merello E, De Marco P, Mascelli S, Raso A, Calevo MG, Torre M, Cama A, Lerone M, Martucciello G, Capra V. HLXB9 homeobox gene and caudal regression syndrome. *Birth Defects Res A Clin Mol Teratol*. 2006 Mar;76(3):205-9. Erratum in: *Birth Defects Res A Clin Mol Teratol*. 2006 Jul;76(7):568.

: Torre M, Martucciello G, Barabino A, Bertagnon M, Brizzolara A, Repetto P, Favre A, Briano S, Nobili F, Mattioli G, Gambini C, Jasonni V. [Constipation and chronic intestinal pseudoobstruction as a clinical expression of intestinal neuronal dysplasia (IND)]. *Minerva Gastroenterol Dietol*. 1998 Dec;44(4):179-86. Italian.

: Lantieri F, Griseri P, Puppo F, Campus R, Martucciello G, Ravazzolo R, Devoto M, Ceccherini I. Haplotypes of the human RET proto-oncogene associated with Hirschsprung disease in the Italian population derive from a single ancestral combination of alleles. *Ann Hum Genet*. 2006 Jan;70(Pt 1):12-26.

: Martucciello G, Pini Prato A, Puri P, Holschneider AM, Meier-Ruge W, Jasonni V, Tovar JA, Grosfeld JL. Controversies concerning diagnostic guidelines for anomalies of the enteric nervous system: a report from the fourth International Symposium on Hirschsprung's disease and related neurocristopathies. *J Pediatr Surg*. 2005 Oct;40(10):1527-31.

: Holschneider A, Hutson J, Peña A, Beket E, Chatterjee S, Coran A, Davies M, Georgeson K, Grosfeld J, Gupta D, Iwai N, Kluth D, Martucciello G, Moore S, Rintala R, Smith ED, Sripathi DV, Stephens D, Sen S, Ure B, Grasshoff S, Boemers T, Murphy F, Söylet Y, Dübbers M, Kunst M. Preliminary report on the International Conference for the Development of Standards for the Treatment of Anorectal Malformations. *J Pediatr Surg*. 2005 Oct;40(10):1521-6.

: Pini Prato A, Martucciello G, Torre M, Jasonni V. Feasibility of perineal sagittal approaches in patients without anorectal malformations. *Pediatr Surg Int*. 2004 Oct;20(10):762-7.

: Mattioli G, Castagnetti M, Martucciello G, Jasonni V. Results of a mechanical Duhamel pull-through for the treatment of Hirschsprung's disease and intestinal neuronal dysplasia. *J Pediatr Surg*. 2004 Sep;39(9):1349-55.

: Martucciello G, Torre M, Belloni E, Lerone M, Pini Prato A, Cama A, Jasonni

V. Currarino syndrome: proposal of a diagnostic and therapeutic protocol. *J Pediatr Surg.* 2004 Sep;39(9):1305-11. Review. PubMed PMID: 15359381.

: Brizzolara A, Torre M, Favre A, Pini Prato A, Bocciardi R, Martucciello G.  
Histochemical study of Dom mouse: A model for Waardenburg-Hirschsprung's phenotype. *J Pediatr Surg.* 2004 Jul;39(7):1098-103.

: Sementa AR, Martucciello G, Gambini C. [Histochemical techniques in the morphologic diagnosis of Hirschsprung disease and related diseases]. *Pathologica.* 2003 Oct;95(5):243-4. Italian.

: Sancandi M, Griseri P, Pesce B, Patrone G, Puppo F, Lerone M, Martucciello G, Romeo G, Ravazzolo R, Devoto M, Ceccherini I. Single nucleotide polymorphic alleles in the 5' region of the RET proto-oncogene define a risk haplotype in Hirschsprung's disease. *J Med Genet.* 2003 Sep;40(9):714-8.

: Torre M, Favre A, Pini Prato A, Brizzolara A, Martucciello G. Histologic study of peritoneal adhesions in children and in a rat model. *Pediatr Surg Int.* 2002 Dec;18(8):673-6. Epub 2002 Sep 24.

: Brizzolara A, Favre A, Schäfer KH, Michelazzi A, Sanguineti M, Martucciello G, Jasonni V. Mouse-isolated plexus differentiates neural crest precursors into enteric neuroblasts. *Eur J Pediatr Surg.* 2002 Dec;12(6):391-6.

: Torre M, Martucciello G, Ceccherini I, Lerone M, Aicardi M, Gambini C, Jasonni V. Diagnostic and therapeutic approach to multiple endocrine neoplasia type 2B in pediatric patients. *Pediatr Surg Int.* 2002 Sep;18(5-6):378-83.

: Fava M, Borghini S, Cinti R, Cusano R, Seri M, Lerone M, De Giorgio R, Stanghellini V, Martucciello G, Ravazzolo R, Ceccherini I. HOX11L1: a promoter study to evaluate possible expression defects in intestinal motility disorders. *Int J Mol Med.* 2002 Jul;10(1):101-6.

: Martucciello G, Torre M, Pini Prato A, Lerone M, Campus R, Leggio S, Jasonni V. Associated anomalies in intestinal neuronal dysplasia. *J Pediatr Surg.* 2002 Feb;37(2):219-23.

: Torre M, Martucciello G, Jasonni V. Sacral development in anorectal malformations and in normal population. *Pediatr Radiol.* 2001 Dec;31(12):858-62.

: Martucciello G, Favre A, Torre M, Pini Prato A, Jasonni V. A new rapid acetylcholinesterase histochemical method for the intraoperative diagnosis of Hirschsprung's disease and intestinal neuronal dysplasia. *Eur J Pediatr Surg.* 2001 Oct;11(5):300-4.

: Pini Prato A, Martucciello G, Jasonni V. Solo-RBT: a new instrument for rectal suction biopsies in the diagnosis of Hirschsprung's disease. *J Pediatr Surg.* 2001 Sep;36(9):1364-6.

: Martucciello G, Ceccherini I, Lerone M, Jasonni V. Pathogenesis of Hirschsprung's disease. *J Pediatr Surg.* 2000 Jul;35(7):1017-25. Review.

: Costa M, Fava M, Seri M, Cusano R, Sancandi M, Forabosco P, Lerone M, Martucciello G, Romeo G, Ceccherini I. Evaluation of the HOX11L1 gene as a candidate for congenital disorders of intestinal innervation. *J Med Genet.* 2000 Jul;37(7):E9.

: Belloni E, Martucciello G, Verderio D, Ponti E, Seri M, Jasonni V, Torre M, Ferrari M, Tsui LC, Scherer SW. Involvement of the HLXB9 homeobox gene in Currarino syndrome. *Am J Hum Genet.* 2000 Jan;66(1):312-9.

: Favre A, Briano S, Mazzola C, Brizzolara A, Torre M, Cilli M, Sanguineti M, Martucciello G. Anorectal malformations associated with enteric dysganglionosis in Danforth's short tail (Sd) mice. *J Pediatr Surg.* 1999 Dec;34(12):1818-21.

: Karmarkar SJ, Martucciello G, Repetto P, Karande TP. Primary anastomosis in esophageal atresia type I without a gap. *Pediatr Surg Int.* 1999 Jul;15(5-6):376-7.

: Martucciello G, Mazzola C, Favre A, Negri F, Bertagnon M, Morando A, Torre M,

Gambini C, Jasonni V. Preoperative enzyme-histochemical diagnosis of dysganglionoses associated with anorectal malformations (ARM) with recto-vestibular and recto-perineal fistula. *Eur J Pediatr Surg.* 1999 Apr;9(2):96-100.

: Seri M, Martucciello G, Paleari L, Bolino A, Priolo M, Salemi G, Forabosco P, Caroli F, Cusano R, Tocco T, Lerone M, Cama A, Torre M, Guys JM, Romeo G, Jasonni V. Exclusion of the Sonic Hedgehog gene as responsible for Currarino syndrome and anorectal malformations with sacral hypodevelopment. *Hum Genet.* 1999 Jan;104(1):108-10.

: Jasonni V, Martucciello G. Total colonic aganglionosis. *Semin Pediatr Surg.* 1998 Aug;7(3):174-80. Review.

: Romeo G, Ceccherini I, Celli J, Priolo M, Betsos N, Bonardi G, Seri M, Yin L, Lerone M, Jasonni V, Martucciello G. Association of multiple endocrine neoplasia type 2 and Hirschsprung disease. *J Intern Med.* 1998 Jun;243(6):515-20.

: Martucciello G, Holschneider AM. Update on basic research on Hirschsprung's disease. *Eur J Pediatr Surg.* 1998 Jun;8(3):131-2.

: Martucciello G, Thompson H, Mazzola C, Morando A, Bertagnon M, Negri F, Brizzolara A, Rocchetti L, Gambini C, Jasonni V. GDNF deficit in Hirschsprung's disease. *J Pediatr Surg.* 1998 Jan;33(1):99-102.

: Jasonni V, Lelli-Chiesa PL, Repetto P, Torre M, Nobili F, Mazzola C, Martucciello G. [Congenital deformities of the chest wall. Surgical treatment]. *Minerva Pediatr.* 1997 Sep;49(9):407-13. Italian.

: Lerone M, Bolino A, Martucciello G. The genetics of anorectal malformations: a complex matter. *Semin Pediatr Surg.* 1997 Nov;6(4):170-9. Review.

: Roggero P, Mazzola C, Fava G, Clerici Bagozzi D, Martucciello G, Luzzani S. Intestinal microvillous atrophy and transient neuronal dysplasia. *Transplant Proc.* 1997 May;29(3):1868-9.

: Seri M, Yin L, Barone V, Bolino A, Celli I, Bocciardi R, Pasini B, Ceccherini I, Lerone M, Kristoffersson U, Larsson LT, Casasa JM, Cass DT, Abramowicz MJ, Vanderwinden JM, Kravcenkiene I, Baric I, Silengo M, Martucciello G, Romeo G. Frequency of RET mutations in long- and short-segment Hirschsprung disease. *Hum Mutat.* 1997;9(3):243-9.

: Martucciello G. Hirschsprung's disease as a neurochristopathy. *Pediatr Surg Int.* 1997;12(1):2-10. Review.

: Tam PK, Gould SJ, Martucciello G, Biddolph S, Takahashi M, Jasonni V. Ret protein in the human fetal rectum. *J Pediatr Surg.* 1996 Apr;31(4):568-71.

: Martucciello G, Favre A, Takahashi M, Jasonni V. Immunohistochemical localization of RET protein in Hirschsprung's disease. *J Pediatr Surg.* 1995 Mar;30(3):433-6.

: Martucciello G, Caffarena PE, Lerone M, Mattioli G, Barabino A, Bisio G, Jasonni V. Neuronal intestinal dysplasia: clinical experience in Italian patients. *Eur J Pediatr Surg.* 1994 Oct;4(5):287-92.

: Jasonni V, Lelli Chiesa P, Martinelli M, Mattioli G, Martucciello G. [Surgical treatment of thoracic wall malformations]. *Cir Pediatr.* 1994 Apr;7(2):83-7.

: Caffarena PE, Mattioli G, Bisio G, Martucciello G, Ivani G, Jasonni V. Long-gap oesophageal atresia: a combined endoscopic and radiologic evaluation. *Eur J Pediatr Surg.* 1994 Apr;4(2):67-9.

: Mattioli G, Cagnazzo A, Martucciello G, Jasonni V. Use of staplers in congenital lobar emphysema. *Eur J Pediatr Surg.* 1993 Dec;3(6):359-61.

: Silengo M, Lerone M, Romeo G, Calcagno E, Martucciello G, Jasonni V. Uncombable hair, retinal pigmentary dystrophy, dental anomalies, and brachydactyly: report of a new patient with additional findings. *Am J Med Genet.*

1993 Nov 1;47(6):931-3.

: Lyonnet S, Bolino A, Pelet A, Abel L, Nihoul-Fékété C, Briard ML, Mok-Siu V, Kaariainen H, Martucciello G, Lerone M, et al. A gene for Hirschsprung disease maps to the proximal long arm of chromosome 10. *Nat Genet.* 1993 Aug;4(4):346-50.

: Puliti A, Covone AE, Biccocchi MP, Bolino A, Lerone M, Martucciello G, Jasonni V, Romeo G. Deleted and normal chromosome 10 homologs from a patient with Hirschsprung disease isolated in two cell hybrids through enrichment by immunomagnetic selection. *Cytogenet Cell Genet.* 1993;63(2):102-6.

: Lerone M, Soliani M, Corea D, Romeo G, Martucciello G, Silengo MC. Congenital diaphragmatic hernia associated with ipsilateral upper limb reduction defects: report of a case with thumb hypoplasia. *Am J Med Genet.* 1992 Dec 1;44(6):827-9.

: Silengo M, Lerone M, Martinelli M, Martucciello G, Caffarena PE, Jasonni V, Romeo G. Autosomal recessive microcephaly with early onset seizures and spasticity. *Clin Genet.* 1992 Sep;42(3):152-5.

: Doderio P, Martucciello G, Bisio G. [Intraoperative histochemical diagnosis of Hirschsprung disease]. *Cir Pediatr.* 1992 Apr;5(2):72-4.

: Fratino G, Granata C, Dodero P, Lonati L, Martucciello G, Taccone A, Jasonni V. [An association between anorectal malformations and Down's syndrome]. *Pediatr Med Chir.* 1992 Mar-Apr;14(2):157-9. Review.

: Taccone A, Marzoli A, Martucciello G, Dodero P. Intraabdominal calcifications in the newborn: an unusual case with anorectal malformation and other anomalies. *Pediatr Radiol.* 1992;22(4):309-10.

: Taccone A, Martucciello G, Dodero P, Delliacqua A, Marzoli A, Salomone G, Jasonni V. New concepts in preoperative imaging of anorectal malformation. New concepts in imaging of ARM. *Pediatr Radiol.* 1992;22(3):196-9.

: Dodero P, Martinelli M, Cuneo GP, Martucciello G, Granata C, Fratino G, Bisio G, Michelazzi A, Arioni C, Ivani G. [Foreign bodies in childhood: general considerations and the report of an exceptional case of splenic abscess caused by an ingested foreign body]. *Pediatr Med Chir.* 1991 Nov-Dec;13(6):623-4.

: Taccone A, Marzoli A, Martucciello G, Salomone G, Magnano GM, Dodero P, Jasonni V. [Computed tomography vs. magnetic resonance in the diagnosis of anorectal anomalies]. *Radiol Med.* 1991 Nov;82(5):638-43.

: Martucciello G, Taccone A, Cheli M, Lonati L, Oddone M, Dodero P, Pelizza A,

Jasonni V. [Blocked transcolostomy colonography in the identification of anorectal malformations]. *Radiol Med*. 1991 Nov;82(5):635-7.

: Dodero P, Caffarena PE, Martinelli M, Fratino G, Martucciello G, Ivani G, Taccone A, Pallecchi AE. [Unusual pelvic pathology treated with posterior sagittal anorectoplasty]. *Pediatr Med Chir*. 1991 May-Jun;13(3):289-91

: Lerone M, Dodero P, Romeo G, Martucciello G, Caffarena PE, Brisigotti M, Toma P, Taccone A, Silengo M. Leiomyomatosis of oesophagus, congenital cataracts and hematuria. Report of a case with rectal involvement. *Pediatr Radiol*. 1991;21(8):578-9.

: Ivani G, Fossa S, Della Rocca M, De Negri P, Rizzo A, Podestà E, Martucciello G, Fratino G. [Propofol for the induction and maintenance of anesthesia in children undergoing operations for more than 2 hours]. *Minerva Anesthesiol*. 1990 Sep;56(9):885. Italian.

: De Negri P, Ivani G, Della Rocca M, Dodero P, Martucciello G, Fratino P, Tamisani AM. [Anesthesiologic approach in surgery of malformations in infants]. *Minerva Anesthesiol*. 1990 Sep;56(9):663.

: Rizzo A, Lerone M, Martucciello G, Perez Aytes A, Calcagno E, Romeo G. [Lip pits and Van der Woude syndrome. Description of a new familial case]. *Pediatr Med*

Chir. 1990 Jan-Feb;12(1):99-103.

: Pisano F, Pallecchi AE, Tuo P, Salomone G, Martucciello G, Dodero P. [Apple peel small bowel: surgical correction and post-operative treatment]. *Pediatr Med Chir.* 1989 Nov-Dec;11(6):707-11.

: Fondelli P, Taccone A, Martucciello G, Dodero P, Caffarena P. [Computed tomography and anorectal malformations. Their postoperative evaluation]. *Radiol Med.* 1989 Apr;77(4):361-4.

: Taccone A, Fondelli P, Martucciello G, Dodero P, Caffarena P. [Computed tomography and anorectal malformations. Their preoperative evaluation]. *Radiol Med.* 1989 Apr;77(4):356-60.

: Fondelli P, Martucciello G, Taccone A, Dodero P. [The radiologic picture of total colonic aganglionosis]. *Radiol Med.* 1989 Mar;77(3):207-10.

: Taccone A, Martucciello G, Fondelli P, Dodero P, Ghiorzi M. CT of anorectal malformation--a postoperative evaluation. *Pediatr Radiol.* 1989;19(6-7):375-8.

: Martucciello G, Dodero P, Romańczuk W. Hirschsprung's disease: alpha-naphthylesterase activity in the enzyme-histochemical evaluation of the extent of the aganglionic segment during surgery. *Mater Med Pol.* 1988

Oct-Dec;20(4):245-51.

: Dodero P, Martucciello G, Morán Penco JM. [Hirschsprung disease: histochemical technic for alpha-naphthyl-esterase in the intraoperative diagnosis of the proximal extent of the aganglionosis]. An Esp Pediatr. 1988 May;28(5):429-32.

: Giampalmo A, Berardi L, Martucciello G, Musso GL, Rossi F. Enzymatic activation of lymphoid population following inflammatory reactions in the human appendix. Z Mikrosk Anat Forsch. 1983;97(5):785-96..