

- Werner P. Genetic aspects of adenomatosis of endocrine glands. *Am J Med* 1954;16:363–371.
- Kouvaraki MA, Lee JE, Shapiro SE, et al. Genotype-phenotype analysis in multiple endocrine neoplasia type 1. *Arch Surg* 2002;137:641–647.
- Lévy-Bohbot N, Merle C, Goudet P, et al. Prevalence, characteristics and prognosis of MEN 1-associated glucagonomas, VIPomas, and somatostatinomas: study from the GTE (Groupe des Tumeurs Endocrines) registry. *Gastroenterol Clin Biol* 2004;28:1075–1081.
- Triponez F, Dosses D, Goudet P, et al. Epidemiology data on 108 MEN 1 patients from the GTE with isolated nonfunctioning tumors of the pancreas. *Ann Surg* 2006;243:265–272.
- Verges B, Boureille F, Goudet P, et al. Pituitary disease in MEN type 1 (MEN1): data from the France-Belgium MEN1 multicenter study. *J Clin Endocrinol Metab* 2002;87:457–465.
- Goudet P, Bonithon-Kopp C, Murat A, et al. Gender-related differences in MEN1 lesion occurrence and diagnosis: a cohort study of 734 cases from the Groupe d'étude des Tumeurs Endocrines. *Eur J Endocrinol* 2011;165:97–105.
- Thevenon J, Bourredjem A, Faivre L, et al. Higher risk of death among MEN1 patients with mutations in the JunD interacting domain: a Groupe d'étude des Tumeurs Endocrines (GTE) cohort study. *Hum Mol Genet* 2013;22:1940–1948.
- Chandrasekharappa SC, Guru SC, Manickam P, et al. Positional cloning of the gene for multiple endocrine neoplasia-type 1. *Science* 1997;276:404–407.
- Manickam P, Vogel AM, Agarwal SK, et al. Isolation, characterization, expression and functional analysis of the zebrafish ortholog of MEN1. *Mamm Genome* 2000;11:448–454.
- Huang SC, Zhuang Z, Weil RJ, et al. Nuclear/cytoplasmic localization of the multiple endocrine neoplasia type 1 gene product, menin. *Lab Invest* 1999;79:301–310.
- Crabtree JS, Scacheri PC, Ward JM, et al. A mouse model of multiple endocrine neoplasia, type 1, develops multiple endocrine tumors. *Proc Natl Acad Sci U S A* 2001;98:1118–1123.
- Lemos MC, Thakker RV. Multiple endocrine neoplasia type 1 (MEN1): analysis of 1336 mutations reported in the first decade following identification of the gene. *Hum Mutat* 2008;29:22–32.
- Huang J, Gurung B, Wan B, et al. The same pocket in menin binds both MLL and JUND but has opposite effects on transcription. *Nature* 2012;482:542–546.
- Gurung B, Hua X, Menin/PRMT5/hedgehog signaling: a potential target for the treatment of multiple endocrine neoplasia type 1 tumors. *Epigenomics* 2013;5:469–471.
- Agarwal SK. Multiple endocrine neoplasia type 1. *Front Horm Res* 2013;41:1–15.
- Thakker RV. Multiple endocrine neoplasia type 1 (MEN1). *Best Pract Res Clin Endocrinol Metab* 2010;24:355–370.
- Bassett JH, Forbes SA, Pannett AA, et al. Characterization of mutations in patients with multiple endocrine neoplasia type 1. *Am J Hum Genet* 1998;62:232–244.
- Knudson AG Jr, Strong LC, Anderson DE. Heredity and cancer in man. *Prog Med Genet* 1973;9:113–158.
- Agarwal SK, Debelenko LV, Kester MB, et al. Analysis of recurrent germline mutations in the MEN1 gene encountered in apparently unrelated families. *Hum Mutat* 1998;12:75–82.
- Owens M, Ellard S, Vaidya B. Analysis of gross deletions in the MEN1 gene in patients with multiple endocrine neoplasia type 1. *Clin Endocrinol* 2008;68:350–354.
- Schussheim DH, Skarulis MC, Agarwal SK, et al. Multiple endocrine neoplasia type 1: new clinical and basic findings. *Trends Endocrinol Metab* 2001;12:173–178.
- Brown EM, Gamba G, Riccardi D, et al. Cloning and characterization of an extracellular Ca(2+)-sensing receptor from bovine parathyroid. *Nature* 1993;366:575–580.
- Foley TP Jr, Harrison HC, Arnaud CD, et al. Familial benign hypercalcemia. *J Pediatr* 1972;81:1060–1067.
- Pollak MR, Brown EM, Chou YH, et al. Mutations in the human Ca(2+)-sensing receptor gene cause familial hypocalcemic hypercalcemia and neonatal severe hyperparathyroidism. *Cell* 1993;75:1297–1303.
- Heath H 3rd, Jackson CE, Otterud B, et al. Genetic linkage analysis in familial benign (hypocalcemic) hypercalcemia: evidence for locus heterogeneity. *Am J Hum Genet* 1993;53:193–200.
- Lloyd SE, Pannett AA, Dixon PH, et al. Localization of familial benign hypercalcemia, Oklahoma variant (FBHOk), to chromosome 19q13. *Am J Hum Genet* 1999;64:189–195.
- Nesbit MA, Hannan FM, Graham U, et al. Identification of a second kindred with familial hypocalcemic hypercalcemia type 3 (FHH3) narrows localization to a <3.5 megabase pair region on chromosome 19q13.3. *J Clin Endocrinol Metab* 2010;95:1947–1954.
- Alon US, VandeVoerde RC. Beneficial effect of cinacalcet in a child with familial hypocalcemic hypercalcemia. *Pediatr Nephrol* 2010;25:1747–1750.
- Pollak MR, Brown EM, Estep HL, et al. Autosomal dominant hypocalcaemia caused by a Ca(2+)-sensing receptor gene mutation. *Nat Genet* 1994;8:303–307.
- Kinoshita Y, Hori M, Taguchi M, et al. Functional activities of mutant calcium-sensing receptors determine clinical presentations in patients with autosomal dominant hypocalcemia. *J Clin Endocrinol Metab* 2014;99:E363–E368.
- Warner J, Epstein M, Sweet A, et al. Genetic testing in familial isolated hyperparathyroidism: unexpected results and their implications. *J Med Genet* 2004;41:155–160.
- Warner JV, Nyholt DR, Busfield F, et al. Familial isolated hyperparathyroidism is linked to a 1.7 Mb region on chromosome 2p13.3-14. *J Med Genet* 2006;43:e12.
- Jackson CE, Norum RA, Boyd SB, et al. Hereditary hyperparathyroidism and multiple ossifying jaw fibromas: a clinically and genetically distinct syndrome. *Surgery* 1990;108:1006–1012; discussion 1012–1013.
- Cavaco BM, Guerra L, Bradley KJ, et al. Hyperparathyroidism-jaw tumor syndrome in Roma families from Portugal is due to a founder mutation of the HRPT2 gene. *J Clin Endocrinol Metab* 2004;89:1747–1752.
- Guarnieri V, Scillitani A, Muscarella LA, et al. Diagnosis of parathyroid tumors in familial isolated hyperparathyroidism with HRPT2 mutation: implications for cancer surveillance. *J Clin Endocrinol Metab* 2006;91:2827–2832.
- Carpten JD, Robbins CM, Villablanca A, et al. HRPT2, encoding parafibromin, is mutated in hyperparathyroidism-jaw tumor syndrome. *Nat Genet* 2002;32:676–680.
- Lin L, Czapiga M, Nini L, et al. Nuclear localization of the parafibromin tumor suppressor protein implicated in the hyperparathyroidism-jaw tumor syndrome enhances its proapoptotic function. *Mol Cancer Res* 2007;5:183–193.
- Bradley KJ, Cavaco BM, Bowl MR, et al. Parafibromin mutations in hereditary hyperparathyroidism syndromes and parathyroid tumors. *Clin Endocrinol* 2006;64:299–306.
- Newey PJ, Bowl MR, Cranston T, et al. Cell division cycle protein 73 homolog (CDC73) mutations in the hyperparathyroidism-jaw tumor syndrome (HPT-JT) and parathyroid tumors. *Hum Mutat* 2010;31:295–307.
- Howell VM, Haven CJ, Kahnoski K, et al. HRPT2 mutations are associated with malignancy in sporadic parathyroid tumours. *J Med Genet* 2003;40:657–663.
- Shattuck TM, Valimaki S, Obara T, et al. Somatic and germ-line mutations of the HRPT2 gene in sporadic parathyroid carcinoma. *N Engl J Med* 2003;349:1722–1729.
- Krebs LJ, Shattuck TM, Arnold A. HRPT2 mutational analysis of typical sporadic parathyroid adenomas. *J Clin Endocrinol Metab* 2005;90:5015–5017.
- Carney JA, Gordon H, Carpenter PC, et al. The complex of myxomas, spotty pigmentation, and endocrine overactivity. *Medicine (Baltimore)* 1985;64:270–283.
- Daly AF, Jaffrain-Rea ML, Ciccarelli A, et al. Clinical characterization of familial isolated pituitary adenomas. *J Clin Endocrinol Metab* 2006;91:3316–3323.
- Igreja S, Chahal HS, King P, et al. Characterization of aryl hydrocarbon receptor interacting protein (AIP) mutations in familial isolated pituitary adenoma families. *Hum Mutat* 2010;31:950–960.
- Dull AB, Carlson DB, Petrusis JR, et al. Characterization of the phosphorylation status of the hepatitis B virus X-associated protein 2. *Arch Biochem Biophys* 2002;406:209–221.
- Vierimaa O, Georgitsi M, Lehtonen R, et al. Pituitary adenoma predisposition caused by germline mutations in the AIP gene. *Science* 2006;312:1228–1230.
- Daly AF, Tichomirowa MA, Petrossians P, et al. Clinical characteristics and therapeutic responses in patients with germ-line AIP mutations and pituitary adenomas: an international collaborative study. *J Clin Endocrinol Metab* 2010;95:E373–E383.
- Leontiou CA, Gueorguiev M, van der Spuy J, et al. The role of the aryl hydrocarbon receptor-interacting protein gene in familial and sporadic pituitary adenomas. *J Clin Endocrinol Metab* 2008;93:2390–2401.
- Steiner AL, Goodman AD, Powers SR. Study of a kindred with pheochromocytoma, medullary thyroid carcinoma, hyperparathyroidism and Cushing's disease: multiple endocrine neoplasia, type 2. *Medicine (Baltimore)* 1968;47:371–409.
- Williams ED, Pollock DJ. Multiple mucosal neuromata with endocrine tumours: a syndrome allied to von Recklinghausen's disease. *J Pathol Bacteriol* 1966;91:71–80.
- Famdon JR, Leight GS, Dille WG, et al. Familial medullary thyroid carcinoma without associated endocrinopathies: a distinct clinical entity. *Br J Surg* 1986;73:278–281.
- Wells SA Jr, Santoro M. Targeting the RET pathway in thyroid cancer. *Clin Cancer Res* 2009;15:7119–7123.
- Lairmore TC, Ball DW, Baylin SB, et al. Management of pheochromocytomas in patients with multiple endocrine neoplasia type 2 syndromes. *Ann Surg* 1993;217:595–601; discussion 601–603.
- Lips CJ, Landsvater RM, Hoppener JW, et al. Clinical screening as compared with DNA analysis in families with multiple endocrine neoplasia type 2A. *N Engl J Med* 1994;331:828–835.
- Takahashi M, Ritz J, Cooper GM. Activation of a novel human transforming gene, ret, by DNA rearrangement. *Cell* 1985;42:581–588.
- Tahira T, Ishizaka Y, Itoh F, et al. Characterization of ret proto-oncogene mRNAs encoding two isoforms of the protein product in a human neuroblastoma cell line. *Oncogene* 1990;5:97–102.

58. Myers SM, Eng C, Ponder BA, et al. Characterization of RET proto-oncogene 3' splicing variants and polyadenylation sites: a novel C-terminus for RET. *Oncogene* 1995;11:2039–2045.
59. de Graaff E, Srinivas S, Kilkenny C, et al. Differential activities of the RET tyrosine kinase receptor isoforms during mammalian embryogenesis. *Genes Dev* 2001;15:2433–2444.
60. Baloh RH, Tansey MG, Lampe PA, et al. Artemin, a novel member of the GDNF ligand family, supports peripheral and central neurons and signals through the GFRalpha3-RET receptor complex. *Neuron* 1998;21:1291–1302.
61. Creedon DJ, Tansey MG, Baloh RH, et al. Neurturin shares receptors and signal transduction pathways with glial cell line-derived neurotrophic factor in sympathetic neurons. *Proc Natl Acad Sci U S A* 1997;94:7018–7023.
62. Sanicola M, Hession C, Worley D, et al. Glial cell line-derived neurotrophic factor-dependent RET activation can be mediated by two different cell-surface accessory proteins. *Proc Natl Acad Sci U S A* 1997;94:6238–6243.
63. Donis-Keller H, Dou S, Chi D, et al. Mutations in the RET proto-oncogene are associated with MEN 2A and FMTC. *Hum Mol Genet* 1993;2:851–856.
64. Mulligan LM, Kwok JB, Healey CS, et al. Germ-line mutations of the RET proto-oncogene in multiple endocrine neoplasia type 2A. *Nature* 1993;363:458–460.
65. Carlson KM, Bracamontes J, Jackson CE, et al. Parent-of-origin effects in multiple endocrine neoplasia type 2B. *Am J Hum Genet* 1994;55:1076–1082.
66. Imai T, Uchino S, Okamoto T, et al. High penetrance of pheochromocytoma in multiple endocrine neoplasia 2 caused by germ line RET codon 634 mutation in Japanese patients. *Eur J Endocrinol* 2013;168:683–687.
67. Frank-Raue K, Rybicki LA, Erlic Z, et al. Risk profiles and penetrance estimations in multiple endocrine neoplasia type 2A caused by germline RET mutations located in exon 10. *Hum Mutat* 2011;32:51–58.
68. Herfarth KK, Bartsch D, Doherty GM, et al. Surgical management of hyperparathyroidism in patients with multiple endocrine neoplasia type 2A. *Surgery* 1996;120:966–973; discussion 973–974.
69. Gagel RF, Levy ML, Donovan DT, et al. Multiple endocrine neoplasia type 2a associated with cutaneous lichen amyloidosis. *Ann Intern Med* 1989;111:802–806.
70. Borst MJ, VanCamp JM, Peacock ML, et al. Mutational analysis of multiple endocrine neoplasia type 2A associated with Hirschsprung's disease. *Surgery* 1995;117:386–391.
71. Eng C, Smith DP, Mulligan LM, et al. Point mutation within the tyrosine kinase domain of the RET proto-oncogene in multiple endocrine neoplasia type 2B and related sporadic tumours. *Hum Mol Genet* 1994;3:237–241.
72. Smith DP, Houghton C, Ponder BA. Germline mutation of RET codon 883 in two cases of de novo MEN 2B. *Oncogene* 1997;15:1213–1217.
73. Miyauchi A, Futami H, Hai N, et al. Two germline missense mutations at codons 804 and 806 of the RET proto-oncogene in the same allele in a patient with multiple endocrine neoplasia type 2B without codon 918 mutation. *Jpn J Cancer Res* 1999;90:1–5.
74. Iwashita T, Murakami H, Kurokawa K, et al. A two-hit model for development of multiple endocrine neoplasia type 2B by RET mutations. *Biochem Biophys Res Commun* 2000;268:804–808.
75. Kameyama K, Okinaga H, Takami H. RET oncogene mutations in 75 cases of familial medullary thyroid carcinoma in Japan. *Biomed Pharmacother* 2004;58:345–347.
76. Nakao KI, Usui T, Ikeda M, et al. Novel tandem germline RET proto-oncogene mutations in a patient with multiple endocrine neoplasia type 2B: report of a case and a literature review of tandem RET mutations with in silico analysis. *Head Neck* 2013;35:E363–E368.
77. Elisei R, Cosci B, Romei C, et al. Prognostic significance of somatic RET oncogene mutations in sporadic medullary thyroid cancer: a 10-year follow-up study. *J Clin Endocrinol Metab* 2008;93:682–687.
78. Moura MM, Cavaco BM, Pinto AE, et al. High prevalence of RAS mutations in RET-negative sporadic medullary thyroid carcinomas. *J Clin Endocrinol Metab* 2011;96:E863–E868.
79. Elisei R, Romei C, Cosci B, et al. RET genetic screening in patients with medullary thyroid cancer and their relatives: experience with 807 individuals at one center. *J Clin Endocrinol Metab* 2007;92:4725–4729.
80. Skinner MA, Moley JA, Dilley WG, et al. Prophylactic thyroidectomy in multiple endocrine neoplasia type 2A. *N Engl J Med* 2005;353:1105–1113.
81. Brandi ML, Gagel RF, Angeli A, et al. Guidelines for diagnosis and therapy of MEN type 1 and type 2. *J Clin Endocrinol Metab* 2001;86:5658–5671.
82. American Thyroid Association Guidelines Task Force, Kloos RT, Eng C, et al. Medullary thyroid cancer: management guidelines of the American Thyroid Association. *Thyroid* 2009;19:565–612.
83. Chen H, Sippel RS, O'Dorisio MS, et al. The North American Neuroendocrine Tumor Society consensus guideline for the diagnosis and management of neuroendocrine tumors: pheochromocytoma, paraganglioma, and medullary thyroid cancer. *Pancreas* 2010;39:775–783.
84. Tuttle RM, Ball DW, Byrd D, et al. Medullary carcinoma. *J Natl Compr Canc Netw* 2010;8:512–530.
85. Matuszczyk A, Petersenn S, Bockisch A, et al. Chemotherapy with doxorubicin in progressive medullary and thyroid carcinoma of the follicular epithelium. *Horm Metab Res* 2008;40:210–213.
86. Druker BJ, Guilhot F, O'Brien SG, et al. Five-year follow-up of patients receiving imatinib for chronic myeloid leukemia. *N Engl J Med* 2006;355:2408–2417.
87. Wells SA Jr, Gosnell JE, Gagel RF, et al. Vandetanib for the treatment of patients with locally advanced or metastatic hereditary medullary thyroid cancer. *J Clin Oncol* 2010;28:767–772.
88. Robinson BC, Paz-Ares L, Krebs A, et al. Vandetanib (100 mg) in patients with locally advanced or metastatic hereditary medullary thyroid cancer. *J Clin Endocrinol Metab* 2010;95:2664–2671.
89. Wells SA Jr, Robinson BC, Gagel RF, et al. Vandetanib in patients with locally advanced or metastatic medullary thyroid cancer: a randomized, double-blind phase III trial. *J Clin Oncol* 2012;30:134–141.
90. Elisei R, Schlumberger MJ, Muller SP, et al. Cabozantinib in progressive medullary thyroid cancer. *J Clin Oncol* 2013;31:3639–3646.
91. Fagin JA. The Jeremiah Metzger Lecture: intelligent design of cancer therapy: trials and tribulations. *Trans Am Clin Climatol Assoc* 2007;118:253–261.
92. Howell GM, Hodak SP, Yip L. RAS mutations in thyroid cancer. *Oncologist* 2013;18:926–932.
93. Lee J, Hwang JA, Lee EK. Recent progress of genome study for anaplastic thyroid cancer. *Genomics Inform* 2013;11:68–75.
94. Fusco A, Grieco M, Santoro M, et al. A new oncogene in human thyroid papillary carcinomas and their lymph-nodal metastases. *Nature* 1987;328:170–172.
95. Santoro M, Melillo RM, Fusco A. RET/PTC activation in papillary thyroid carcinoma: European Journal of Endocrinology Prize Lecture. *Eur J Endocrinol* 2006;155:645–653.
96. Klugbauer S, Lengfelder E, Demidchik EP, et al. High prevalence of RET rearrangement in thyroid tumors of children from Belarus after the Chernobyl reactor accident. *Oncogene* 1995;11:2459–2467.
97. Nikiforov YE, Rowland JM, Bove KE, et al. Distinct pattern of ret oncogene rearrangements in morphological variants of radiation-induced and sporadic thyroid papillary carcinomas in children. *Cancer Res* 1997;57:1690–1694.
98. Ho AL, Grewal RK, Leboeuf R, et al. Selumetinib-enhanced radioiodine uptake in advanced thyroid cancer. *N Engl J Med* 2013;368:623–632.
99. Pennell NA, Daniels GH, Haddad RI, et al. A phase II study of gefitinib in patients with advanced thyroid cancer. *Thyroid* 2008;18:317–323.
100. Brose MS, Nutting C, Jarzab B, et al. Sorafenib in locally advanced or metastatic patients with radioactive iodine-refractory differentiated thyroid cancer: The phase III Decision trial. *J Clin Oncol* 2013;31:4.
101. Kroll TG, Sarraf P, Pecciarini L, et al. PAX8-PPARgamma1 fusion oncogene in human thyroid carcinoma [corrected]. *Science* 2000;289:1357–1360.
102. Nikiforova MN, Lynch RA, Biddinger PW, et al. RAS point mutations and PAX8-PPAR gamma rearrangement in thyroid tumors: evidence for distinct molecular pathways in thyroid follicular carcinoma. *J Clin Endocrinol Metab* 2003;88:2318–2326.
103. Zhu GR, Ji O, Ji JM, et al. Combining nilotinib and imatinib improves the outcome of imatinib-resistant blast phase CML. *Acta Haematol* 2012;127:152–155.
104. Gomez-Almaguer D, Tarin-Arza L, Cantu-Rodriguez O, et al. More about imatinib and nilotinib combination therapy in chronic myeloid leukemia. *Acta Haematol* 2013;129:18–19.