

## 分子医療部門 変異遺伝子解析研究分野（原研遺伝）

### A 欧 文

#### A-a

1. Mizugichi T, Furuta I, Watanabe Y, Tsukamoto K, Tomita H, Tsujihata M, Ohta T, Kishino T, Matsumoto N, Minakami H, Niikawa N, Yoshiura K: LRP5, low density lipoprotein receptor-related protein 5, is a determinant for bone mineral density (BMD). *J Hum Genet* 49 (2): 80-86, 2004 \* ◇
2. Harada N, Hatchwell E, Okamoto N, Tsukahara M, Kurosawa K, Kawame H, Kondoh T, Ohashi H, Tsukino R, Kondoh Y, Shimokawa O, Ida T, Nagai T, Fukushima Y, Niikawa N, Matsumoto N: Subtelomere specific microarray based comparative genomic hybridisation: a rapid detection system for cryptic rearrangements in idiopathic mental retardation. *J Med Genet* 41 (2): 130-136, 2004 \*
3. Masuzaki H, Miura K, Yoshiura K, Yoshimura S, Ishimaru T: A monozygotic conjoined twin pregnancy discordant for laterality of cleft lip. *Gynecol Obstet Invest* 57 (2): 100-102, 2004 \* ◇
4. Yamada T, Mitsuya K, Kayashima T, Yamasaki K, Ohta T, Yoshiura K, Matsumoto N, Yamada H, Minakami H, Oshimura M, Niikawa N, Kishino T: Imprinting analysis of 10 genes and/or transcripts in a 1.5-Mb MEST-flanking region at human chromosome 7q32. *Genomics* 83 (3): 402-412, 2004 \*
5. Iwata K, Takamura N, Nakashima M, Alipov G, Mine M, Matsumoto N, Yoshiura K, Prouglo Y, Sekine I, Katayama I, Yamashita S: Loss of heterozygosity on chromosome 9q22.3 in microdissected basal cell carcinoma around Semipalatinsk nuclear testing site, Kazakhstan. *Hum Pathol* 35 (4): 460-464, 2004 \*
6. Masuzaki H, Miura K, Yoshiura K, Yoshimura S, Niikawa N, Ishimaru T: Detection of cell-free placental DNA in maternal plasma: Direct evidence from three cases of confined placental mosaicism. *J Med Genet* 41 (4): 289-292, 2004 \* ◇
7. Matsuzawa N, Yoshiura K, Machida J, Nakamura T, Niimi T, Furukawa H, Toyoda T, Natsume N, Shimozato K, Niikawa N: Two missense mutations in the IRF6 gene in two Japanese families with Van der Woude syndrome. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 98 (4): 414-417, 2004 \*
8. Ghadami M, Majidzadeh-A K, Morovvati S, Damavandi E, Nishimura G, Kinoshita A, Pasalar P, Komatsu K, Najafi MT, Niikawa N, Yoshiura K: An isolated congenital anosmia locus maps to 18p11.23-q12.2. *J Med Genet* 41 (4): 299-303, 2004 \*
9. Iwakoshi M, Okamoto N, Harada N, Nakamura T, Yamamori S, Fujita H, Niikawa N, Matsumoto N: 9q34.3 deletion syndrome in three unrelated children. *Am J Med Genet* 126A (3): 278-283, 2004 \*
10. Miyake N, Tonoki H, Gallego M, Harada N, Shimokawa O, Yoshiura K, Ohta T, Kishino T, Niikawa N, Matsumoto N: Phenotype-genotype correlation in two patients with 12q proximal deletion. *J Hum Genet* 49 (5): 282-284, 2004 \*
11. Kinoshita A, Shirahama A, Miyahara A, Nishimura G, Haga N, Namba A, Ueda H, Hayashi H, Seidel J, Fukumaki Y, Ikegawa S, Niikawa N, Yoshiura K: *TGFB1* mutations in four new families with Camurati-Engelmann disease: Confirmation of independently arising LAP-domain-specific mutations. *Am J Med Genet* 127A (1): 104-107, 2004 \*
12. Kato R, Kawamura J, Sugawara H, Niikawa N, Matsumoto N: A rapid diagnostic method for a retrotransposal insertional mutation into the *FCMD* gene in Japanese patients with Fukuyama congenital muscular dystrophy. *Am J Med Genet* 127A (1): 54-57, 2004 \*
13. Clayton EW, Knoppers BM, Qiu R, Kent A, Dunston GM, Kato K, Niikawa N, Foster MW, Adewole IF, Watkin J, Zhang H, Zeng C, Matsuda I, Fukushima Y, Macer DR, Suda E, Rotimi CN, Adebamowo CA, Aniagwu T, Marshall PA, Matthew O, Nkwodimma C, Royal CDM, Leppert MF, Dixon M, Valle DL, Jorde LB, Belmont JW, Chakravarti A, Cho MK, Duster T, Foster MW, Jasperse M, Kwok PW, Licinio J, Long JC, Ossorio PN, Wang VO, Rotimi CN, Spallone P, Terry SF, Gibbs RA, Belmont JW, Sodergren E, Weinstock GM, Birren BW, Wilson RK, Fulton LL, Rogers J, Feolo M, Altshuler D, Daly MJ, Schaffner SF, Stein LD, Cunningham F, Kanani A, Thorisson GA, Chakravarti A, Chen PE, Cutler DJ, Kashuk CS, Lin S, Donnelly P, Marchini J, McVean GAT, Myers SR, Hardenbol P, Willis TD, Yu F, Altshuler D, Yang H, Ch'ang LY, Huang W, Liu B, Yan S, Tam PKH, Tsui L-C, Waye MMY, Wong JT-F, Zeng C, Zhang Q, Chee MS, Galver LM, Kruglyak S, Murray SS, Oliphant AR, Montpetit A, Chagnon F, Bellemare V, Todani K, Fujita T, Tanaka S, Holden AL, Lai EH, Collins FS, McEwen JE, Brooks LD, Guyer MS, Jordan E, Peterson JL, Spiegel J, Sung LM, Zacharia LF, Dunn MG, Seabrook R, Shillito M, Skene B, Stewart JG, Lander E, Nickerson DA, Abecasis GR, Altshuler D, Bentley DR, Boehnke M, Cardon LR, Daly MJ, Deloukas P, Douglas JA, Gabriel SB, Hudson RR, Hudson TJ, Kruglyak L, Kwok PY, Nakamura Y, Nussbaum RL, Royal CDM, Sherry ST, Tanaka T (The International HapMap Consortium): Integrating ethics and science in the International HapMap Project. *Nat Rev Genet* 5 (6): 467-475, 2004 \*
14. Ghadami M, Majidzadeh-A K, Morovvati S, Damavandi E, Nishimura G, Kinoshita A, Najafi M-T, Niikawa N, Yoshiura K: Isolated congenital anosmia with morphologically normal olfactory bulb in two unrelated Iranian families: A new clinical entity? *Am J Med Genet* 127A (3): 307-309, 2004 \*
15. Okubo A, Miyoshi O, Baba K, Takagi M, Tsukamoto K, Kinoshita A, Yoshiura K, Kishino T, Ohta T, Niikawa N,

- Matsumoto N: A novel *GATA4* mutation completely segregated with atrial septal defect in a large Japanese family. *J Med Genet* 41 (7): e97, 2004 \* ○◇
16. Kamimura J, Wakui K, Kadokawa H, Watanabe Y, Miyake K, Harada N, Sakamoto M, Kinoshita A, Yoshiura K, Ohta T, Kishino T, Ishikawa M, Kasuga M, Fukushima Y, Niikawa N, Matsumoto N: The IHPK1 gene is disrupted at the 3p21.31 breakpoint of t(3;9) in a family with type 2 diabetes mellitus. *J Hum Genet* 49 (7): 360-365, 2004 \* ○◇
  17. Miyake N, Harada N, Shimokawa O, Ohashi H, Kurosawa K, Matsumoto T, Fukushima Y, Nagai T, Shotelersuk Y, Yoshiura K, Ohta T, Kishino T, Niikawa N, Matsumoto N: On the reported 8p22-p23.1 duplication in Kabuki make-up syndrome (KMS) and its absence in patients with typical KMS. *Am J Med Genet* 128A (2): 170-172, 2004 \* ○○◇
  18. Shimokawa O, Kurosawa K, Ida T, Harada N, Kondoh T, Miyake N, Yoshiura K, Kishino T, Ohta T, Niikawa N, Matsumoto N: Molecular characterization of inv dup del(8p): analysis of five cases. *Am J Med Genet* 128A (2): 133-137, 2004 \* ○
  19. Zuccheri TM, Cooper ME, Maher BS, Daack-Hirsch S, Nepomuceno B, Ribeiro L, Caprau D, Christensen K, Suzuki Y, Machida J, Natsume N, Yoshiura K, Vieira AR, Orioli IM, Castilla EE, Moreno L, Arcos-Burgos M, Lidral AC, Field LL, Liu YE, Ray A, Goldstein TH, Schultz RE, Shi M, Johnson MK, Kondo S, Schutte BC, Marazita ML, Murray JC: Interferon regulatory factor 6 (IRF6) gene variants and the risk of isolated cleft lip or palate. *N Engl J Med* 351 (8): 769-780, 2004 \*
  20. Mizuguchi T, Collod-Beroud G, Akiyama T, Harada N, Morisaki T, Abifadel M, Allard D, Varret M, Claustres M, Ihara M, Kinoshita A, Yoshiura K, Junien C, Kajii T, Jondeau G, Niikawa N, Boileau C, Matsumoto N: Heterozygous *TGFBR2* mutations in Marfan syndrome. *Nat Genet* 36 (8): 855-860, 2004 \* ○○◇
  21. Harada N, Visser R, Dawson A, Fukamachi M, Iwakoshi M, Okamoto N, Kishino T, Niikawa N, Matsumoto N: A 1-Mb critical region in six patients with 9q34.3 terminal deletion syndrome. *Hum Genet* 49 (8): 440-444, 2004 \* ○
  22. Masuzaki H, Miura K, Miura S, Yoshiura K, Mapendano CK, Nakayama D, Yoshimura S, Niikawa N, Ishimaru T: Labor increases maternal DNA contamination in cord blood. *Clin Chem* 50 (9): 1709-1711, 2004 \* ○◇
  23. Kurosawa K, Harada N, Sosonkina N, Niikawa N, Matsumoto N, Saitoh S: Unmasking 15q12 deletion using microarray-based comparative genomic hybridization in a mentally retarded boy with r(Y). *Am J Med Genet* 130A (3): 322-324, 2004 \* ○○◇
  24. Masuzaki H, Miura K, Yoshimura S, Yoshiura K, Ishimaru T: A monozygotic twin pregnancy discordant for acardia and X-inactivation pattern. *Eur J Obstet Gynecol Reprod Biol* 117 (1): 102-104, 2004 \* ○◇
  25. Matsumoto T, Niikawa N: Eight novel microsatellite markers in the 3' region of the dystrophin gene useful for diagnosis of Duchenne muscular dystrophy. *Prenat Diagn* 24 (12): 1014-1015, 2004 \*
  26. Niikawa N: Molecular basis of Sotos syndrome. *Horm Res* 62 (Suppl 3): 60-65, 2004 \*

## B 邦 文

### B-b

1. 新川詔夫：メンデル遺伝とその他の遺伝. 母子保健情報 49 : 16-20, 2004
2. 新川詔夫：単一遺伝子による内分泌疾患の発症機構. 小児科診療 10(13) : 1585-1590, 2004
3. 水口 剛, 松本直通：ヘテロ接合性TGFBR2変異がMarfan症候群2型の原因である. 医学のあゆみ 210(12) : 1010-1011, 2004
4. 霜川 修, 新川詔夫：染色体検査. 臨床精神医学(増刊号) : 484-489, 2004
5. 新川詔夫：ゲノム医学における倫理的課題. 科学 74(5) : 596-601, 2004

### B-c

1. 副島英伸, 太田 亨, 木住野達也, 新川詔夫, 向井常博：インプリンティングドメインの異常と疾患. (佐々木裕之(編)：エビジェネティクス, Springer Reviews 所収) 2004

### B-d

1. 吉浦孝一郎, 夏目長門：顔面奇形の発生抑制遺伝子の解析. 平成15年度科学研究費補助金(特定領域研究(2)「ゲノム医科学」)研究成果報告書, 2004
2. 新川詔夫：染色体転座・微細欠失からの疾病遺伝子の単離と解析. 平成15年度日本科学技術振興機構・戦略的基礎科学研究事業(CREST)研究成果報告書, 2004
3. 新川詔夫, 木住野達也：骨粗鬆症およびゲノム刷り込み関連遺伝子群の解明. 平成15年度科学研究費補助金(特定領域研究(2)「ゲノム医科学」)研究成果報告書, 2004
4. 木下 晃：骨リモデリング研究のためのモデルマウス作製と確立. 平成15年度科学研究費補助金(若手研究(B))

研究実績報告書, 2004

**原著論文数一覧**

	A-a	A-b	A-c	A-d	合計	SCI	B-a	B-b	B-c	B-d	合計	総計
2004	26	0	0	0	26	26	0	5	1	4	10	36

**学会発表数一覧**

	A-a	A-b		合計	B-a	B-b		合計	総計
		シンポジウム	学会			シンポジウム	学会		
2004	1	1	4	6	2	1	11	14	20

**原著論文総数に係る教員生産係数一覧**

	欧文論文総数	教員生産係数	SCI掲載論文数	教員生産係数
	論文総数	(欧文論文)	欧文論文総数	(SCI掲載論文)
2004	0.722	6.5	1	6.5

**Impact factor 値一覧**

	Impact factor	教員当たり Impact factor	論文当たり Impact factor
2004	156.048	39.012	6.002