

## 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. Amani D, Dehaghani SA, Zolghadri J, Ravangard F, Niikawa N, Yoshiura K, Ghaderi A: Lack of association between the TGF- $\beta$ 1 gene polymorphisms and recurrent spontaneous abortion. *J Reprod Immunol* 68(1-2): 91-103, 2005 \*
2. Visser R, Shimokawa O, Harada N, Niikawa N, Matsumoto N: Non-hotspot-related breakpoints of common deletions in Sotos syndrome are located within destabilized DNA regions. *J Med Genet* 42(11): e66, 2005 \*
3. Nomura M, Hamasaki Y, Ktayama I, Niikawa N, Yoshiura K: Eosinophil infiltration and amyloidosis in three patients with generalized atrophic benign epidermolysis bullosa from a Japanese family. *J Hum Genet* 50(9): 483-489, 2005 \*
4. Gibbs RA, Belmont JW, Boudreau A, Leal S, Hardenbol P, Pasternak S, Wheeler DA, Willis TD, Yu F, Yang H, Gao Y, Hu H, Hu W, Li C, Lin W, Liu S, Pan H, Tang X, Wang J, Wang W, Yu J, Zeng C, Zhang B, Zhang Q, Zhao H, Zhao H, Zhou J, Gabrie SB, Barry R, Blumenstiel B, Camargo A, Defelice M, Faggart M, Goyette M, Gupta S, Moore J, Nguyen H, Onofrio RC, Parkin M, Roy J, Stahl E, Winchester E, Ziaugra L, Altshuler D, Shen Y, Yao Z, Huang W, Chu X, He W, Jin L, Liu Y, Shen Y, Sun W, Wang H, Wang Y, Wang Y, Xiong X, Xu L, Tsui SKW, Xue H, Wong JT-F, Galver LM, Fan J-B, Murray SS, Oliphant AR, Chee MS, Montpetit A, Chagnon F, Ferretti V, Leboeuf M, Oliver JF, Phillips MS, Roumy S, Sall S, Verner A, Hudson TJ, Frazer KA, Ballinger DG, Cox DR, Hinds DA, Stuve LL, Kwok R-Y, Cai D, Koboldt DC, Miller RD, Pawlikowska L, Taillon-Miller P, Xiao M, Tsui LC, Mak W, Sham PC, Song YQ, Tam PKH, Nakamura Y, Kawaguchi T, Kitamoto T, Morizono T, Nagashima A, Ohnishi Y, Sekine A, Tanaka T, Tsunoda T, Deloukas P, Delgado M, Gwilliam R, Hunt S, Morrison J, Powell D, Whittake P, Bentley DR, Daly MJ, de Bakker PIW, Barrett J, Fry B, McCarroll S, Patterson N, Pe'er I, Purcell S, Richter DJ, Sabeti P, Saxena R, Schaffner SF, Varilly P, Altshuler D, Stein LD, Krishnan L, Vernon Smith A, Thorisson GA, Chakravarti A, Chen PE, Cutler DJ, Kashuk CS, Lin S, Abecasis GR, Guan W, Munro HM, Qin ZS, Thomas DJ, Bottolo L, Eyheramendy S, Freeman C, Marchini J, McVean G, Myers S, Spencer C, Stephens M, Donnelly P, Cardon LR, Clarke G, Evans DM, Morris AP, Weir BS, Mullikin JC, Sherry ST, Feolo M, Dermitzakis ET, Bird CP, Stranger BE, Zhang H, Zeng C, Zhao H, Matsuda I, Fukushima Y, Macer DR, Suda E, Rotimi CN, Adebamowo CA, Ajayi I, Aniagwu T, Marshall PA, Nkwodimma C, Royal CDM, Leppert MF, Dixon M, Peiffer A, Qiu R, Kent A, Kato K, Niikawa N, Adewole IF, Knoppers BM, Foster MW, Clayton EW, Watkin J, Muzny B, Nazareth L, Pasternak S, Sodergren E, Weinstock GM, Wheeler DA, Gabriel SB, Onofrio RC, Richter DJ, Ziaugra L, Daly MJ, Altshuler D, Wilson RK, Fulton LL, Rogers J, Burton J, Carter NP, Clee CM, Griffiths M, Jones MC, McLay K, Plumb RW, Ross MT, Sims SK, Willey DL, Birren BW, Chen Z, Han H, Kang L, Godbout M, Wallenburg JC, L'Archeveue P, Bellemare G, Saeki K, An D, Li Q, Wang H, Qiu H, Wang R, Holden AL, Brooks LD, Guyer MS, McEwen JE, Nailer PJ, Ota Wang V, Peterson JL, Shi M, Spiegel J, Sung LM, Witonsky J, Zacharia LF, Collins FS, Kennedy K, Jamieson R, Stewart J: A haplotype map of the human genome. *Nature* 437(7063): 1299-1320, 2005 \*
5. Matsumoto T, Miyake N, Watanabe Y, Yamanaka G, Oana S, Ogiwara M, Hoshika A, Miyahara H, Niikawa N: X-linked adrenoleukodystrophy with partial deletion of *ALD* due to fusion with the neighbor gene, *PLXNB*. *Am J Med Genet A* 138(3): 300-302, 2005 \*
6. Saitoh S, Wada T, Okajima M, Takano K, Sudo A, Niikawa N: Uniparental disomy and imprinting defects in Japanese patients with Angelman syndrome. *Brain Develop* 27(5): 389-391, 2005 \*
7. Machida H, Tsukamoto K, Wen C-Y, Narumi Y, Shikuwa S, Isomoto H, Takeshima F, Mizuta Y, Niikawa N, Murata I, Kohno S: Association of polymorphic alleles of CTLA4 with inflammatory bowel disease in the Japanese. *World J Gastroenterol* 11(27): 4188-4193, 2005 ○○◇
8. Machida H, Tsukamoto K, Wen C-Y, Shikuwa S, Isomoto H, Mizuta Y, Takeshima F, Murase K, Matsumoto N, Murata I, Kohno S: Crohn's disease in the Japanese is associated with a SNP-haplotype of the N- acetyltransferase 2 gene. *World J Gastroenterol* 11(31): 4833-4837, 2005 ○◇
9. Yamasaki Y, Kayashima T, Soejima H, Kinoshita A, Yoshiura K, Matsumoto N, Ohta T, Urano T, Masuzaki H, Ishimaru T, Mukai T, Niikawa N, Kishino T: Neuron-specific relaxation of Igf2r imprinting is associated with neuron-specific histone modifications and lack of its antisense transcript Air. *Hum Mol Genet* 14(17): 2511-2530, 2005 \*★◇
10. Shimokawa O, Miyake N, Yoshimura T, Sosonkina N, Harada N, Mizuguchi T, Kondoh S, Kishino T, Ohta T, Remco V, Takashima T, Kinoshita A, Yoshiura K, Niikawa N, Matsumoto N: Molecular characterization of del(8) (p23.1p23.1) in a case of congenital diaphragmatic hernia. *Am J Med Genet A* 136(1): 49-51, 2005 \*
11. Nagai T, Obata K, Tonoki H, Temma S, Murakami N, Katada Y, Yoshino A, Sakazume S, Takahashi E, Sakuta R, Niikawa N: Cause of sudden, unexpected death of Prader-Willi syndrome patients with or without growth hormone treatment. *Am J Med Genet A* 136(1): 45-48, 2005 \*
12. Masuzaki H, Miura K, Yamasaki K, Miura S, Yoshiura K, Yoshimura S, Nakayama D, Mapendano CK, Niikawa N, Ishimaru T: Clinical Applications of plasma circulating mRNA analysis in cases of gestational trophoblastic disease. *Clin Chem* 51(7): 1261-1263, 2005 \*○◇
13. Jin Z-B, Liu X-Q, Uchida A, Vervoot R, Morishita K, Hayakawa M, Murakami A, Matsumoto N, Niikawa N, Nao-I N: Novel

- deletion spanning RCC1-like domain of RPGR in Japanese X-linked retinitis pigmentosa family. Mol Vis 11: 535-541, 2005 \*
14. Iwanaga H, Tsujino A, Shirabe S, Eguchi H, Fukushima N, Niikawa N, Yoshiura K, Eguchi K: A large deletion involving the 5'-UTR in the spastin gene caused mild phenotype of autosomal dominant hereditary spastic paraparesis. Am J Med Genet A 133 (1): 13-17, 2005 \* ○○▽◇
  15. Masuzaki H, Miura K, Yoshiura K, Yamasaki K, Yoshimura S, Miura S, Nakayama D, Mapendano CK, Niikawa N, Ishimaru T: Placental mRNA in maternal plasma and its clinical application to the evaluation of the placental status in a pregnant woman with placenta previa-percreta. Clin Chem 51 (5): 923-925, 2005 \* ○◇
  16. Miyake N, Visser R, Kinoshita A, Yoshiura K, Harada N, Okamoto N, Sonoda T, Kaname T, Chinen Y, Naritomi K, Tonoki H, Kondoh T, Kurosawa K, Niikawa N, Matsumoto N: Four novel *NIPBL* mutations in Japanese patients with Cornelia de Lange syndrome. Am J Med Genet A 135 (1): 103-105, 2005 \* ○◇
  17. Kurotaki N, Stankiewicz P, Wakui K, Niikawa N, Lupski JR: Sotos syndrome common deletion is mediated by directly oriented subunits within inverted Sos-REP low-copy repeats. Hum Mol Genet 14 (4): 535-542, 2005 \*
  18. Visser R, Shimokawa O, Harada N, Kinoshita A, Ohta T, Niikawa N, Matsumoto N: Identification of a 3.0-kb major recombination hotspot in patients with Sotos syndrome who carry a common 1.9-Mb microdeletion. Am J Hum Genet 76 (1): 52-67, 2005 \* ○◇

#### A-b

1. Miura K, Niikawa N: Do monochorionic dizygotic twins increase after pregnancy by assisted reproductive technology? J Hum Genet 50 (1): 1-6, 2005 \*

#### B 邦 文

##### B-c

1. 吉浦孝一郎, 新川詔夫: 疾患遺伝子のポジショナルクローニング. 日本臨床 63 (Suppl 12): 421-426, 2005
2. 新川詔夫: Protective management for genetic information. 日本臨床 63 (3): 383-388, 2005

##### B-d

1. 吉浦孝一郎: 眼瞼下垂原因ホメオボックス遺伝子の機能とOARドメイン相互作用蛋白質の単離. 平成15-16年度科学研究費補助金(基盤研究(C) (2))研究成果報告書, 2005年3月

#### 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
2005	18	1	0	0	19	17	0	0	2	1	3	22

#### 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			
2005	0	0	4	4	0	0	18	18	22

#### 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)
2005	0.864	4.75	0.895	4.25

#### Impact factor値一覧

	Impact factor	教員当たり Impact factor	論文当たり Impact factor
2005	96.646	24.162	5.685