放射線生命科学部門　人類遺伝学研究分野（原研遺伝）

A 欧文

A-a

1. Sasaki K, Mishima H, Miura K, Yoshiura KI : Uniparental disomy analysis in trios using genome-wide SNP array and whole-genome sequencing data imply segmental uniparental isodisomy in general populations. Gene 512(2): 267-274, 2013(IF:2.196) ＊ ◇
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3. Higashijima A, Miura K, Mishima H, Kinoshita A, Jo O, Abe S, Hasegawa Y, Miura S, Yamasaki K, Yoshida A, Yoshiura K, Masuzaki H: Characterization of placenta-specific microRNAs in fetal growth restriction pregnancy. Prenat Diagn 33(3): 214-222, 2013(IF:2.683) ＊ ○ ◇
4. Abe S, Miura K, Kinoshita A, Mishima H, Miura S, Yamasaki K, Hasegawa Y, Higashijima A, Jo O, Sasa K, Yoshida A, Yoshiura K, Masuzaki H: Copy number variation of the antimicrobial-gene, defensin beta 4, is associated with susceptibility to cervical cancer. J Hum Genet 58(5): 250-253, 2013(IF:2.365) ＊ ○ ◇
5. Kashiyama K, Nakazawa Y, Pilz D, Guo C, Shimada M, Sasaki K, Fawcett H, Wing J, Lewin S, Carr L, Yoshiura K, Utani A, Hirano A, Yamashita S, Greenblatt D, Nardo T, Stefanini M, McGibbon D, Sarkany R, Fassihi H, Takahashi Y, Nagayama Y, Mitsutake N, Lehmann AR, and Ogi T: Malfunction of the ERCC1/XPF endonuclease results in diverse clinical manifestations and causes three nucleotide excision-repair-deficient disorders, Cockayne Syndrome, xeroderma pigmentosum and Fanconi Anemia. Am J Hum Genet 92(5): 807-819, 2013(IF:11.202) ＊
6. Miyake N, Koshimizu E, Okamoto N, Mizuno S, Ogata T, Nagai T, Kosho T, Ohashi H, Kato M, Sasaki G, Mabe H, Watanabe Y, Yoshino M, Matsuishi T, Takanashi J, Shotelersuk V, Tekin M, Ochi N, Kubota M, Ito N, Ihara K, Hara T, Tonoki H, Ohta T, Saito K, Matsuo M, Urano M, Enokizono T, Sato A, Tanaka H, Ogawa A, Fujita T, Hiraki Y, Kitanaka S, Matsubara Y, Makita T, Taguri M, Nakashima M, Tsurusaki Y, Saitsu H, Yoshiura K, Matsumoto N, Niikawa N: MLL2 and KDM6A mutations in patients with Kabuki syndrome. Am J Med Gene A. 161(9): 2234-2243, 2013(IF:2.304) ＊
7. Nakao K, Oikawa M, Arai J, Mussazhanova Z, Kondo H, Shichijo K, Nakashima M, Hayashi T, Yoshiura K, Hatachi T, Nagayasu T: A Predictive Factor of the Quality of Microarray Comparative Genomic Hybridization Analysis for Formalin-fixed Paraffin-embedded Archival Tissue. Diagn Mol Pathol 22(3): 174-180, 2013(IF:1.861) ＊
8. Hasegawa Y, Miura K, Furuya K, Yoshiura K, Masuzaki H: Identification of Complete Hydatidiform Mole Pregnancy-Associated MicroRNAs in Plasma. Clin Chem 59(9): 1410-1412, 2013(IF:7.149) ＊ ◇
9. Higashimoto K, Maeda T, Okada J, Ohtsuka Y, Sasaki K, Hirose A, Nomiyama M, Takayanagi T, Fukuzawa R, Yatsuki H, Koide K, Nishioka K, Joh K, Watanabe Y, Yoshiura KI, Soejima H: Homozygous deletion of DIS3L2 exon 9 due to non-allelic homologous recombination between LINE-1s in a Japanese patient with Perlman syndrome. Eur J Hum Genet 21(11):1316-1319, 2013(IF:4.319) ＊
10. Hamaguchi D, Miura K, Abe S, Kinoshita A, Miura S, Yamasaki K, Yoshiura KI, Masuzaki H: Initial viral load in cases of single human papillomavirus 16 or 52 persistent infection is associated with progression of later cytopathological findings in the uterine cervix. J Med Virol 85(12): 2093-2100, 2013(IF:2.373) ＊ ◇

A-b

1. Ishikawa T, Toyoda Y, Yoshiura K, Niikawa N: Pharmacogenetics of human ABC transporter ABCC11: new insights into apocrine gland growth and metabolite secretion. Front Genet 3:306., 2013

B 邦文

B-b

1. Ogi T, Nakazawa Y, Sasaki K, Guo C, Yoshiura K, Utani A, Nagayama Y: [Molecular cloning and characterisation of UVSSA, the responsible gene for UV-sensitive syndrome]. Seikagaku. 85(3): 133-144, 2013. Review. Japanese. No abstract available.

B-c

1. 吉浦孝一郎：遺伝性疾患におけるエクソーム解析の有用性と近将来．医学のあゆみ 245(5):363-368, 2013.
2. 三嶋博之：全エクソーム解析における情報処理．医学のあゆみ 245(5):345-351, 2013.
3. 黒滝直弘，小野慎二，小澤寛樹，吉浦孝一郎：発作性運動誘発性舞踏アテトーゼの分子メカニズム．神経内科 79(6):718-725, 2013.

B-d

1. 吉浦孝一郎（研究代表）：地域集積・収集した稀少疾患の系統的原因究明（難病・がん等の疾患分野の医療の実用化研究事業（難病関係研究分野））総括・分担研究報告 pp: 1-27, 2012.

論文数一覧

	A-a	A-b	A-c	A-d	A-e	合計	SCI	B-a	B-b	B-c	B-d	B-e	合計	総計
2013	10	1	0	0	0	11	10	0	1	3	1	0	5	16

学会発表数一覧

	A-a	A-b		合計		B-a	B-b		合計	総計
		ｼﾝﾎﾟｼﾞｳﾑ	学会				ｼﾝﾎﾟｼﾞｳﾑ	学会		
2013	0	0	1	1		2	2	7	11	12

論文総数に係る教員生産係数一覧

	欧文論文総数 / 論文総数	教員生産係数（欧文論文）		SCI 掲載論文数 / 欧文論文総数	教員生産係数（SCI 掲載論文）
2013	0.688	3.667		0.909	3.333

Impact factor 値一覧

	Impact factor	教員当たり Impact factor	論文当たり Impact factor
2013	40.921	13.64	4.092