

# 人類遺伝学研究分野

## 論文

### A 欧文

#### A-a

1. Nikitski A, Rogounovitch T, Bychkov A, Takahashi M, Yoshiura KI, Mitsutake N, Kawaguchi T, Matsuse M, Drozd VM, Demidchik YE, Nishihara E, Hirokawa M, Miyauchi A, Rubanovich AV, Matsuda F, Yamashita S, Saenko VA. Genotype analyses in the Japanese and Belarusian populations reveal independent effects of rs965513 and rs1867277 but do not support the role of FOXE1 polyalanine tract length in conferring risk for papillary thyroid carcinoma. *Thyroid* 27(2): 224-235, 2016. doi:10.1089/thy.2015.0541. (IF: 3.784) \*
2. Uchiyama Y, Nakashima M, Watanabe S, Miyajima M, Taguri M, Miyatake S, Miyake N, Saito H, Mishima H, Kinoshita A, Arai H, Yoshiura K, Matsumoto N. Ultra-sensitive droplet digital PCR for detecting a low-prevalence somatic GNAQ mutation in Sturge-Weber syndrome. *Sci Rep* 6:22985, 2016. doi:10.1038/srep22985. (IF: 5.228) \*◇
3. Koga T, Migita K, Sato S, Umeda M, Nonaka F, Kawashiri SY, Iwamoto N, Ichinose K, Tamai M, Nakamura H, Origuchi T, Ueki Y, Masumoto J, Agematsu K, Yachie A, Yoshiura K, Eguchi K, Kawakami A. Multiple Serum Cytokine Profiling to Identify Combinational Diagnostic Biomarkers in Attacks of Familial Mediterranean Fever. *Medicine (Baltimore)* 95(16): e3449, 2016. doi:10.1097/MD.0000000000003449. (IF: 2.133) \*
4. Watanabe S, Shimizu K, Ohashi H, Kosaki R, Okamoto N, Shimojima K, Yamamoto T, Chinen Y, Mizuno S, Dowa Y, Shiomi N, Toda Y, Tashiro K, Shichijo K, Minatozaki K, Aso S, Minagawa K, Hiraki Y, Shimokawa O, Matsumoto T, Fukuda M, Moriuchi H, Yoshiura K, Kondoh T. Detailed analysis of 26 cases of 1q partial duplication/triplication syndrome. *Am J Med Genet A* 170(4): 908-917, 2016. doi:10.1002/ajmg.a.37496. (IF: 2.082) \*◇
5. Dateki S, Watanabe S, Nakatomi A, Kinoshita E, Matsumoto T, Yoshiura K, Moriuchi H. Genetic background of hyperphenylalaninemia in Nagasaki, Japan. *Pediatr Int* 2016 May;58(5): 431-433. doi:10.1111/ped.12924. (IF: 0.868) \*
6. Mussazhanova Z, Akazawa Y, Matsuda K, Shichijo K, Miura S, Otsubo R, Oikawa M, Yoshiura KI, Mitsutake N, Rogounovitch T, Saenko V, Kozykenova Z, Zhetpisbaev B, Shabdarbaeva D, Sayakenov N, Amantayev B, Kondo H, Ito M, Nakashima M. Association between p53-binding protein 1 expression and genomic instability in oncocytic follicular adenoma of the thyroid. *Endocr J* 63(5): 457-467, 2016. doi:10.1507/endocrj.EJ15-0629. (IF: 1.895) \*
7. Migita K, Izumi Y, Jiuchi Y, Iwanaga N, Kawahara C, Agematsu K, Yachie A, Masumoto J, Fujikawa K, Yamasaki S, Nakamura T, Ubara Y, Koga T, Nakashima Y, Shimizu T, Umeda M, Nonaka F, Yasunami M, Eguchi K, Yoshiura K, Kawakami A. Familial Mediterranean fever is no longer a rare disease in Japan. *Arthritis Res Ther* 18: 175, 2016. doi:10.1186/s13075-016-1071-5. (IF: 3.979) \*
8. Wada H, Matsuda K, Akazawa Y, Yamaguchi Y, Miura S, Ueki N, Kinoshita A, Yoshiura K, Kondo H, Ito M, Nagayasu T, Nakashima M. Expression of Somatostatin Receptor Type 2A and PTEN in Neuroendocrine Neoplasms Is Associated with Tumor Grade but Not with Site of Origin. *Endocr Pathol* 27(3): 179-187, 2016. doi:10.1007/s12022-016-9436-5. (IF: 1.817) \*◇
9. Konomoto T, Imamura H, Orita M, Tanaka E, Moritake H, Sato Y, Fujimoto S, Harita Y, Hisano S, Yoshiura KI, Nunoi H. Clinical and histological findings of autosomal dominant renal-limited disease with LMX1B mutation. *Nephrology (Carlton)*. 21(9): 765-773, 2016. doi:10.1111/nep.12666. (IF: 1.796) \*
10. Miura K, Mishima H, Yasunami M, Kaneuchi M, Kitajima M, Abe S, Higashijima A, Fuchi N, Miura S, Yoshiura KI, Masuzaki H. A significant association between rs8067378 at 17q12 and invasive cervical cancer originally identified by a genome-wide association study in Han Chinese is replicated in a Japanese population. *J Hum Genet* 61(9): 793-796, 2016. doi:10.1038/jhg.2016.50. (IF: 2.487) \*◇
11. Matsuda K, Tateishi S, Akazawa Y, Kinoshita A, Yoshida S, Morisaki S, Fukushima A, Matsuwaki T, Yoshiura KI, Nakashima M. Rapid growth of mitotically active cellular fibroma of the ovary: a case report and review of the literature. *Diagn Pathol* 11(1): 101, 2016. doi:10.1186/s13000-016-0554-7 (IF: 1.895) \*

### B 邦文

#### B-c

1. ゲノム医学 -ゲノム情報を活かす医療のために- 監訳：菅野純夫，福嶋義光．第8章訳担当（株）メディカルサイエンスインターナショナル．

**研究業績集計表**

教室等名：505 ゲノム機能解析部門 人類遺伝学研究分野（原研遺伝）

**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 |
|------|-----|-----|-----|-----|-----|----|-----|-----|-----|-----|-----|-----|----|----|
| 2016 | 11  | 0   | 0   | 0   | 0   | 11 | 11  | 0   | 0   | 1   | 0   | 0   | 1  | 12 |

**学会発表数一覧**

|      | A-a | A-b    |    | 合計 |  | B-a | B-b    |    | 合計 | 総計 |
|------|-----|--------|----|----|--|-----|--------|----|----|----|
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| 2016 | 0   | 0      | 6  | 6  |  | 0   | 0      | 2  | 2  | 8  |

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|------|-----------------------|------------------|--|---------------------------|---------------------|
| 2016 | 0.917                 | 3.667            |  | 1                         | 3.667               |

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| 2016 | 27.964        | 9.321               | 2.542               |