人類遺伝学研究分野 (原研遺伝)

論文

A 欧文

A-a

1. Dateki S, Watanabe S, Mishima H, Shirakawa T, Morikawa M, Kinoshita E, Yoshiura KI, Moriuchi H. A homozygous splice site ROBO1 mutation in a patient with a novel syndrome with combined pituitary hormone deficiency. J Hum Genet. 2019 Apr;64(4):341-346. doi: 10.1038/s10038-019-0566-8. (IF: 3.545) ＊◇

2. Kousa YA, Zhu H, Fakhouri WD, Lei Y, Kinoshita A, Roushangar RR, Patel NK, Agopian AJ, Yang W, Leslie EJ, Busch TD, Mansour TA, Li X, Smith AL, Li EB, Sharma DB, Williams TJ, Chai Y, Amendt BA, Liao EC, Mitchell LE, Bassuk AG, Gregory S, Ashley-Koch A, Shaw GM, Finnell RH, Schutte BC. The TFAP2A-IRF6-GRHL3 genetic pathway is conserved in neurulation. Hum Mol Genet. 2019 May 15;28(10):1726-1737.Jan 25. doi: 10.1093/hmg/ddz010. (IF: 4.544) ＊☆

3. Shimizu H, Watanabe S, Kinoshita A, Mishima H, Nishimura G, Moriuchi H, Yoshiura KI, Dateki S. Identification of a homozygous frameshift variant in RFLNA in a patient with a typical phenotype of spondylocarpotarsal synostosis syndrome. J Hum Genet. 2019 May;64(5):467-471. doi: 10.1038/s10038-019-0581-9. (IF: 3.545) ＊○◇

4. Morimoto Y, Yoshida S, Kinoshita A, Satoh C, Mishima H, Yamaguchi N, Matsuda K, Sakaguchi M, Tanaka T, Komohara Y, Imamura A, Ozawa H, Nakashima M, Kurotaki N, Kishino T, Yoshiura KI, Ono S. Nonsense mutation in CFAP43 causes normal-pressure hydrocephalus with ciliary abnormalities. Neurology. 2019 May 14;92(20):e2364-e2374. doi: 10.1212/WNL.0000000000007505. (IF: 8.689) ＊★◇

5. Yamashita Y, Nishikawa A, Iwahashi Y, Fujimoto M, Sasaki I, Mishima H, Kinoshita A, Hemmi H, Kanazawa N, Ohshima K, Imadome KI, Murata SI, Yoshiura KI, Kaisho T, Sonoki T, Tamura S. Identification of a novel CCDC22 mutation in a patient with severe Epstein-Barr virus-associated hemophagocytic lymphohistiocytosis and aggressive natural killer cell leukemia. Int J Hematol. 2019 Jun;109(6):744-750. doi: 10.1007/s12185-019-02595-0. (IF: 2.251) ＊

6. Matsuno S, Furuta H, Kosaka K, Doi A, Yorifuji T, Fukuda T, Senmaru T, Uraki S, Matutani N, Furuta M, Mishima H, Iwakura H, Nishi M, Yoshiura K, Fukui M, Akamizu T. Identification of a variant-associated with early-onset diabetes in the intron of INS gene with exome sequencing. J Diabetes Investig. 2019 Jul; 10(4): 947-950. doi: 10.1111/jdi.12974. (IF: 3.902) ＊

7. Mishima H, Suzuki H, Doi M, Miyazaki M, Watanabe S, Matsumoto T, Morifuji K, Moriuchi H, Yoshiura KI, Kondoh T, Kosaki K. Evaluation of Face2Gene using facial images of patients with congenital dysmorphic syndromes recruited in Japan. J Hum Genet. Aug; 64(8) 789-794. doi: 10.1038/s10038-019-0619-z. (IF: 3.545) ＊

8. Masui D, Fukahori S, Mizuochi T, Watanabe Y, Fukui K, Ishii S, Saikusa N, Hashizume N, Higashidate N, Sakamoto S, Takato A, Yoshiura KI, Tanaka Y, Yagi M. Cystic biliary atresia with paucity of bile ducts and gene mutation in KDM6A: a case report. Surgical Case Reports 2019 Aug 14;5(1):132. doi: 10.1186/s40792-019-0688-4.◇

9. Shibano M, Watanabe A, Takano N, Mishima H, Kinoshita A, Yoshiura KI, Shibahara T. Target Capture/Next-Generation Sequencing for Nonsyndromic Cleft Lip and Palate in the Japanese Population. Cleft Palate Craniofac J. 2020 Jan;57(1): 80-87. doi: 10.1177/1055665619857650. (IF: 1.471) ＊◇

10. Tanaka A, Matsuse M, Saenko V, Nakao T, Yamanouchi K, Sakimura C, Yano H, Nishihara E, Hirokawa M, Suzuki K, Miyauchi A, Eguchi S, Yoshiura KI, Yamashita S, Nagayasu T, Mitsutake N. TERT mRNA Expression as a Novel Prognostic Marker in Papillary Thyroid Carcinomas. Thyroid. 2019 Aug;29(8):1105-1114. doi: 10.1089/thy.2018.0695. (IF: 7.786) ＊◇

11. Endo Y, Koga T, Nakashima M, Mishima H, Yoshiura KI, Kawakami A. Atypical phenotype without fever in a Japanese family with an autosomal dominant transmission of familial Mediterranean fever due to heterozygous MEFV Thr577Asn mutations. Clin Exp Rheumatol. 2019 Nov-Dec; 37 Suppl 121(6):161-162. ◇

12. Hamaguchi Y, Aoki M, Watanabe S, Mishima H, Yoshiura KI, Moriuchi H, Dateki S. KAT6B-related disorder in a patient with a novel frameshift variant (c.3925dup). Hum Genome Var. 2019 Dec 13;6:54. doi: 10.1038/s41439-019-0085-3. eCollection 2019. ◇

B 邦文

B-c

1. 吉浦孝一郎．全ゲノム関連解析研究（GWAS）の原理と考え方．遺伝子医学 27, Vol9 (1): 114-121. 2019 年 1 月発行．メディカルドゥ

2. 吉浦孝一郎．連鎖解析．遺伝子医学 27, Vol9 (4): 109-115. 2019 年 10 月発行．メディカルドゥ

3. 三嶋博之：0 から始める疾患ゲノム解析 ver2，学研メディカル秀潤社，2019，細胞工学別冊・次世代シークエンサー DRY 解析教本改訂第二版（清水厚志，坊農秀雅 編），64-80.

4. 三嶋博之・清水厚志：コマンドラインの使い方，学研メディカル秀潤社，2019，細胞工学別冊・次世代シークエンサ

ーDRY 解析教本改訂第二版（清水厚志，坊農秀雅 編），20-39.

論文研究業績集計表

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
2019	12	0	0	0	0	12	10	0	0	4	0	0	4	16

学会発表数一覧

	A-a	A-b シンポジウム	A-b 学会	合計	B-a	B-b シンポジウム	B-b 学会	合計	総計
2019	0	0	3	3	2	0	8	10	13

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)
2019	0.750	4.000	0.833	3.333

Impact factor 値一覧

	Impact factor	教員当たり Impact factor	論文当たり Impact factor
2019	39.278	13.093	3.928