

人類遺伝学研究分野 (原研遺伝)

論文

A 欧文

A-a

- 1 . 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.* 57(1): 80-87, 2020 doi: 10.1177/1055665619857650. (IF: 1.433) ○◇*
- 2 . Taguchi M, Mishima H, Shiozawa Y, Hayashida C, Kinoshita A, Nannya Y, Makishima H, Horai M, Matsuo M, Sato S, Itonaga H, Kato T, Taniguchi H, Imanishi D, Imaizumi Y, Hata T, Takenaka M, Moriuchi Y, Shiraishi Y, Miyano S, Ogawa S, Yoshiura KI, Miyazaki Y: Genome analysis of myelodysplastic syndromes among atomic bomb survivors in Nagasaki. *Haematologica* 105(2): 358-365, 2020 10.3324/haematol.2019.219386. (IF: 9.941) *
- 3 . Shimizu K, Oba D, Nambu R, Tanaka M, Oguma E, Murayama K, Ohtake A, Yoshiura KI, Ohashi H: Possible mitochondrial dysfunction in a patient with deafness, dystonia, and cerebral hypomyelination (DDCH) due to BCAP31 Mutation. *Mol Genet Genomic Med* 8(3): e1129 2020 doi: 10.1002/mgg3.1129. (IF: 2.183) *
- 4 . Morita S, Takeshima K, Ariyasu H, Furukawa Y, Kishimoto S, Tsuji T, Uraki S, Mishima H, Kinoshita A, Takahashi Y, Inaba H, Iwakura H, Furuta H, Nishi M, Doi A, Murata SI, Yoshiura KI, Akamizu T: Expression of unfolded protein response markers in the pheochromocytoma with Waardenburg syndrome: a case report. *BMC Endocr Disord* 20(1): 90 2020 doi: 10.1186/s12902-020-00574-9. (IF: 2.763) *
- 5 . Nakano Y, Komiya C, Shimizu H, Mishima H, Shiba K, Tsujimoto K, Ikeda K, Kashimada K, Dateki S, Yoshiura KI, Ogawa Y, Yamada T: A case of ezetimibe-effective hypercholesterolemia with a novel heterozygous variant in ABCG5. *Endocr J.* 67(11): 1099-1105, 2020 doi: 10.1507/endocrj.EJ20-0044. (IF: 2.349) *
- 6 . Ishikawa T, Mishima H, Barc J, Takahashi MP, Hirono K, Terada S, Kowase S, Sato T, Mukai Y, Yui Y, Ohkubo K, Kimoto H, Watanabe H, Hata Y, Aiba T, Ohno S, Chishaki A, Shimizu W, Horie M, Ichida F, Nogami A, Yoshiura KI, Schott JJ, Makita N: Cardiac Emerinopathy: A Non-syndromic Nuclear Envelopathy with Increased Risk of Thromboembolic Stroke due to Progressive Atrial Standstill and Left Ventricular Non-compaction. *Circ Arrhythm Electrophysiol* 13(10): e008712 2020 doi: 10.1161/CIRCEP.120.008712. (IF: 6.568) *
- 7 . Yamashita Y, Morita S, Hosoi H, Kobata H, Kishimoto S, Ishibashi T, Mishima H, Kinoshita A, Backes BJ, Yoshiura KI, Papa FR, Sonoki T, Tamura S: Targeting Adaptive IRE1 α Signaling and PLK2 in Multiple Myeloma: Possible Anti-Tumor Mechanisms of KIRA8 and Nilotinib. *Int J Mol Sci* 21(17): 6314 2020 doi: 10.3390/ijms21176314. (IF: 5.923) *
- 8 . Kaneko N, Kurata M, Yamamoto T, Shigemura T, Agematsu K, Yamazaki T, Takeda H, Sawasaki T, Koga T, Kawakami A, Yachie A, Migita K, Yoshiura K-i, Urano T, Masumoto J: KN3014, a piperidine-containing small compound, inhibits auto-secretion of IL-1 β from PBMCs in a patient with Muckle-Wells syndrome. *Scientific Reports* 10(1): 13562 2020 doi: 10.1038/s41598-020-70513-0. (IF: 4.379) *
- 9 . Otsuki Y, Ueda K, Nuri T, Satoh C, Maekawa R, Yoshiura KI: EEC-LM-ADULT syndrome caused by R319H mutation in TP63 with ectrodactyly, syndactyly, and teeth anomaly: A case report. *Medicine (Baltimore)* 99(44): e22816 2020 doi: 10.1097/MD.00000000000022816. (IF: 1.889) *
- 10 . Morishima S, Maeda M, Itonaga T, Sato-Kawano N, Yoshiura KI, Ihara K: Sphenoethmoidal meningoencephalocele with variable hypopituitarism: A case report and review of literature. *Clinical Pediatric Endocrinology* 29(4): 183-187, 2020 doi: 10.1297/cpe.29.183. (IF: 0.26) *
- 11 . Koga T, Sato S, Mishima H, Migita K, Endo Y, Umeda M, Sumiyoshi R, Nonaka F, Fukui S, Kawashiri SY, Iwamoto N, Ichinose K, Tamai M, Nakamura H, Origuchi T, Ueki Y, Masumoto J, Agematsu K, Yachie A, Yoshiura KI, Eguchi K, Kawakami A: Next-generation sequencing of the whole MEFV gene in Japanese patients with familial Mediterranean fever: a case-control association study. *Clin Exp Rheumatol Suppl* 127(5): 35-41, 2020 . (IF: 4.473) *
- 12 . Satoh C, Kondoh T, Shimizu H, Kinoshita A, Mishima H, Nishimura G, Miyazaki M, Okano K, Kumai Y, Yoshiura KI: Brothers with novel compound heterozygous mutations in COL27A1 causing dental and genital abnormalities. *Eur J Med Genet* 64(2): 104125 2020 doi: 10.1016/j.ejmg.2020.104125. (IF: 2.708) ◇*
- 13 . Vos RA, Katayama T, Mishima H, Kawano S, Kawashima S, Kim J-D, Moriya Y, Tokimatsu T, Yamaguchi A, Yamamoto Y, Wu H, Amstutz P, Antezana E, Aoki NP, Arakawa K, Bolleman JT, Bolton E, Bonnal RJP, Bono H, Burger K, Chiba H, Cohen KB, Deutsch EW, Fernández-Breis JT, Fu G, Fujisawa T, Fukushima A, García A, Goto N, Groza T, Hercus C, Hoehndorf R, Itaya K, Juty N, Kawashima T, Kim J-H, Kinjo AR, Kotera M, Kozaki K, Kumagai S, Kushida T, Lütteke T, Matsubara M, Miyamoto J, Mohsen A, Mori H, Naito Y, Nakazato T, Nguyen-Xuan J, Nishida K, Nishida N, Nishide H, Ogishima S, Ohta T, Okuda S, Paten B, Perret J-L, Prathipati P, Prins P, Queralt-Rosinach N, Shinmachi D, Suzuki S, Tabata T, Takatsuki T, Taylor K, Thompson M, Uchiyama I, Vieira B, Wei C-H, Wilkinson M, Yamada I, Yamanaka R, Yoshitake K, Yoshizawa AC, Dumontier M, Kosaki K, Takagi T: BioHackathon 2015: Semantics of data for life sciences and reproducible research. *F1000Res* 9: 136 2020 doi: 10.12688/f1000research.18236.1. *

A-c

1. Masuzaki H. ed: General Remarks About Autosomal Diseases. Fetal Morph Functional Diagnosis (Comprehensive Gynecology and Obstetrics) 1 st ed.: 191-195, 2020 .

B 邦文

B-c

1. 監修 新川詔夫, 共著 太田亨, 吉浦孝一郎, 三宅紀子: 遺伝医学への招待. 遺伝医学への招待 (第6版) : 2020年1月10日

論文研究業績集計表

論文数一覧

	A-a	A-b	A-c	A-d	A-e	SCI	合計	B-a	B-b	B-c	B-d	B-e	合計	総計
2020	13	0	1	0	0	13	14	0	0	1	0	0	1	15

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
2020	0	0	1	1	0	2	1	3	4

論文総数に係る教員生産係数

	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)
2020	0.933	4.667	0.929	4.333

Impact Factor 値一覧

	Impact Factor	教員当たりのImpact Factor	論文当たりのImpact Factor
2020	44.869	14.956	3.451