人類遺伝学研究分野（原研遺伝）

論文

A　欧文

A-a

1．Inoue T, Takase R, Uchida K, Kodo K, Suda K, Watanabe Y, Yoshiura KI, Kunimatsu M, Ishizaki R, Azuma K, Inai K, Muneuchi J, Furutani Y, Akagawa H, Yamagishi H: The c.1617del variant of TMEM260 is identified as the most frequent single gene determinant for Japanese patients with a specific type of congenital heart disease. J Hum Genet 69(5): 215-222,2024. doi: 10.1038/s10038-024-01225-w. ＊

2．Matsumoto M, Oyake M, Itonaga T, Maeda M, Suenobu S, Sato D, Sasahara Y, Mishima H, Yoshiura KI, Ihara K: Characteristic phenotypes of ADH5/ALDH2 deficiency during childhood. Eur J Med Genet 69: 104939,. doi: 10.1016/j.ejmg.2024.104939. ＊

3．Ibe M, Tamura S, Kosako H, Yamashita Y, Ishii M, Tanaka M, Mishima H, Kinoshita A, Iwabuchi S, Morita S, Yoshiura KI, Hashimoto S, Nakao N, Inoue S: Familial schwannomatosis carrying LZTR1 variant p.R340X with brain tumor: A case report. Mol Genet Metab Rep 40: 101107,2024. doi: 10.1016/j.ymgmr.2024.101107. ＊

4．Hatta D, Makiya S, Kanamoto K, Watanabe K, Fuchigami Y, Kawakami S, Kinoshita A, Yoshiura KI, Kurotaki N, Shirotani K, Iwata N: Proline-rich transmembrane protein 2 regulates the magnitude and frequency of dopamine release by repetitive neuronal stimuli in the striatum of L-dopa-treated mice. Neuropsychopharmacol Rep 44(4): 829-834,2024. doi: 10.1002/npr2.12478. ＊

5．Wang Z, Kometani M, Zeitlin L, Wilnai Y, Kinoshita A, Yoshiura KI, Ninomiya H, Imamura T, Guo L, Xue J, Yan L, Ohashi H, Pretemer Y, Kawai S, Shiina M, Ogata K, Cohn DH, Matsumoto N, Nishimura G, Toguchida J, Miyake N, Ikegawa S: Heterozygous mutations in the straitjacket region of the latency-associated peptide domain of TGFB2 cause Camurati-Engelmann disease type II. J Hum Genet 69(11): 599-605,2024. doi: 10.1038/s10038-024-01274-1. ＊

6．Kobayashi Y, Ando K, Imaizumi Y, Sakamoto H, Kitanosono H, Taguchi M, Mishima H, Kinoshita A, Bekytbek S, Baba M, Kato T, Horai M, Itonaga H, Sato S, Yoshiura KI, Miyazaki Y: RUNX1 expression is regulated by a super-enhancer and is a therapeutic target in adult T-cell leukemia/lymphoma. Leuk Lymphoma 65(14): 2116-2128,2024. doi: 10.1080/10428194.2024.2393258. ＊

7．Koga T, Kita K, Okumura J, Yoshiura KI, Kawakami A: A novel frameshift mutation in ADCK1 identified in a case of chronic fatigue syndrome successfully treated with oral 5-ALA/SFC. Immunol Med 26: 1-5,2024. doi: 10.1080/25785826.2024.2445399. ＊

B　邦文

B-e-2

1．三嶋博之：最先端医療の今　顔貌解析による遺伝性希少疾患診断支援. Medical Science Digest 50(11): 624-625.

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
2024	7	0	0	0	0	7	7	0	0	0	0	1	1	8

学会発表数一覧

	A-a	A-b ｼﾝﾎﾟｼﾞｳﾑ	A-b 学会	合計	B-a	B-b ｼﾝﾎﾟｼﾞｳﾑ	B-b 学会	合計	総計
2024	0	0	1	1	0	1	6	7	8

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）
2024	0.875	2.333	1.000	2.333