人類遺伝学研究分野（原研遺伝）

論文

A　欧文

A-a

1．Itonaga M, Okanari K, Maeda T, Yoshiura KI, Ihara K: Simultaneous monitoring of oxygen and carbon dioxide for Pitt-Hopkins syndrome. Pediatr Int. 64(1): e15180,2022. doi: 10.1111/ped.15180. (IF: 1.4) ＊

2．Matsushima S, Kato K, Yoshimi A, Yoshiura KI, Tsuchida M: Pernicious anemia associated with Kabuki syndrome. Medicine (Baltimore) 64(1): e22816,2022. doi: 0.1111/ped.14960. (IF: 1.4) ＊

3．Tamura S, Kosako H, Furuya Y, Yamashita Y, Mushino T, Mishima H, Kinoshita A, Nishikawa A, Yoshiura KI, Sonoki T: A Patient with Kabuki Syndrome Mutation Presenting with Very Severe Aplastic Anemia. Acta Haematol. 145(1): 89-96,2022. doi: 10.1159/000518227. (IF: 2.4) ＊

4．Endo Y, Funakoshi Y, Koga T, Ohashi H, Takao M, Miura K, Yoshiura KI, Matsumoto T, Moriuchi H, Kawakami A: Large deletion in 6q containing the TNFAIP3 gene associated with autoimmune lymphoproliferative syndrome. Clin Immunol. 235: 108853,2022. doi: 10.1016/j.clim.2021.108853. (IF: 8.6) ＊

5．Kojima-Ishii K, Sakakibara N, Murayama K, Nagatani K, Murata S, Otake A, Koga Y, Suzuki H, Uehara T, Kosaki K, Yoshiura KI, Mishima H, Ichimiya Y, Mushimoto Y, Horinouchi T, Nagano C, Yamamura T, Iijima K, Nozu K: BCS1L mutations produce Fanconi syndrome with developmental disability. J Hum Genet. 67(3): 143-148,2022. doi: 10.1038/s10038-021-00984-0. (IF: 3.5) ＊

6．Motoyama Rie, Matsudaira Takashi, Terada Kiyoshito, Usui Naotaka, Yoshiura KI, Takahashi Yukitoshi: PRRT2 mutation in a Japanese woman: Adult-onset focal epilepsy coexisting with movement disorders and cerebellar atrophy. Epilepsy Behav Rep. 19: 100554,2022. doi: 10.1016/j.ebr.2022.100554. (IF: 1.5) ＊

7．Aoki S, Higashimoto K, Hidaka H, Ohtsuka Y, Aoki S, Mishima H, Yoshiura KI, Nakabayashi K, Hata K, Yatsuki H, Hara S, Ohba T, Katabuchi H, Soejima H: Aberrant hypomethylation at imprinted differentially methylated regions is involved in biparental placental mesenchymal dysplasia. Clin Epigenetics 14(1): 64,2022. doi: 10.1186/s13148-022-01280-0. (IF: 5.7) ＊

8．Hamaguchi Y, Kondoh T, Fukuda M, Yamasaki K, Yoshiura KI, Moriuchi H, Morii M, Muramatsu M, Minami T, Osato M: Leukopenia, macrocytosis, and thrombocytopenia occur in young adults with Down syndrome. Gene 835: 146663,2022. doi: 10.1016/j.gene.2022.146663. (IF: 3.5) ＊

9．Takahashi Y, Date H, Oi H, Adachi T, Imanishi N, Kimura E, Takizawa H, Kosugi S, Matsumoto N, Kosaki K, Matsubara Y.; IRUD Consortium, Mizusawa H: Six years' accomplishment of the Initiative on Rare and Undiagnosed Diseases: nationwide project in Japan to discover causes, mechanisms, and cures. J Hum Genet. 67(9): 505-513,2022. doi: 10.1038/s10038-022-01025-0. (IF: 3.5) ＊

10．Takase Y, Tanioka S, Ishimura M, Yoshiura KI, Mori Y, Sakaida E, Funakoshi Y, Moriuchi H: A familial case of B-cell expansion with NF-κB and T-cell anergy caused by a G123D heterozygous missense mutation in the CARD11 gene. Pediatr Blood Cancer 69(12): e29941,2022. doi: 10.1002/pbc.29941. (IF: 3.2) ＊

11．Sakamoto H, Ando K, Imaizumi Y, Mishima H, Kinoshita A, Kobayashi Y, Kitanosono H, Kato T, Sawayama Y, Sato S, Hata T, Nakashima M, Yoshiura KI, Miyazaki Y: Alvocidib inhibits IRF4 expression via super-enhancer suppression and adult T-cell leukemia/lymphoma cell growth. Cancer Sci. 113(12): 4049-4103,2022. doi: 10.1111/cas.15550. (IF: 5.7) ＊

12．Kosako H, Yamashita Y, Tanaka K, Mishima H, Iwamoto R, Kinoshita A, Murata SI, Ohshima K, Yoshiura KI, Sonoki T, Tamura S: Intestinal Mucosa-Associated Lymphoid Tissue Lymphoma Transforming into Diffuse Large B-Cell Lymphoma in a Young Adult Patient with Neurofibromatosis Type 1: A Case Report. Medicina (Kaunas-Lithuania) 58(12): 1830,2022. doi: 10.3390/medicina58121830. (IF: 2.6) ＊

13．Nagata Y, Watanabe R, Eichhorn C, Ohno S, Aiba T, Ishikawa T, Nakano Y, Aizawa Y, Hayashi K, Murakoshi N, Nakajima T, Yagihara N, Mishima H, Sudo T, Higuchi C, Takahashi A, Sekine A, Makiyama T, Tanaka Y, Watanabe A, Tachibana M, Morita H, Yoshiura KI, Tsunoda T, Watanabe H, Kurabayashi M, Nogami A, Kihara Y, Horie M, Shimizu W, Makita N, Tanaka T: Targeted deep sequencing analyses of long QT syndrome in a Japanese population. PLoS One 17(12): e0277242,2022. doi: 10.1371/journal.pone.0277242. (IF: 3.7) ＊

14．Hamaguchi Y, Mishima H, Kawai T, Saitoh S, Hata K, Kinoshita A, Yoshiura KI.: Identification of unique DNA methylation sites in Kabuki syndrome using whole genome bisulfite sequencing and targeted hybridization capture followed by enzymatic methylation sequencing. Hum Genet. 67(12): 711-720,2022. doi: 10.1038/s10038-022-01083-4. (IF: 3.5) ○＊

15．Kawakami A, Endo Y, Koga T, Yoshiura KI, Migita K: Autoinflammatory disease: clinical perspectives and therapeutic strategies. Inflamm Regen. 42(1): 37,2022. doi: 10.1186/s41232-022-00217-7. (IF: 8.1) ＊

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
2022	15	0	0	0	0	15	15	0	0	0	0	0	0	15

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
2022	0	0	1	1	0	2	6	8	9

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)
2022	1.000	5.000	1.000	5.000

Impact Factor 値一覧

	Impact Factor	教員当たりのImpact Factor	論文当たりのImpact Factor
2022	55.500	18.500	3.700