研究成果の刊行に関する一覧表

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著者氏名	論文タイトル名	書籍全体の編集者名	書　籍　名	出版社名	出版地	出版年	ページ
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仁科 幸子	レーバー先天盲. ～知っておきたい稀な網膜・硝子体ジストロフィー	オクリスタ	75 (6)	31-37	2019
仁科 幸子	手持ちフォトスクリーナー装置、乳幼児期の眼鏡・コンタクトレンズ	チャイルドヘルス	22 (6)	21-23, 47-49	2019
N. Hatabu, N. Katori, T. Sato, N. Maeda, E. Suzuki, O. Komiyama, H. Tsutsui, T. Nagao, H. Nakauchi-Takahashi, T. Matsunaga, T. Ishii, T. Hasegawa and K. Yamazawa.	A Familial Case of a Whole Germline CDC73 Deletion Discordant for Primary Hyperparathyroidism.	Horm Res Paediatr.	92(1)	56-63.	2019
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M. T. Distefano, S. E. Hemphill, A. M. Oza, R. K. Siegert, A. R. Grant, M. Y. Hughes, B. J. Cushman, H. Azaiez, K. T. Booth, A. Chapin, H. Duzkale, T. Matsunaga, J. Shen, W. Zhang, M. Kenna, L. A. Schimmenti, M. Tekin, H. L. Rehm, A. N. A. Tayoun, S. S. Amr and G. Clingen Hearing Loss Clinical Domain Working.	Correction: ClinGen expert clinical validity curation of 164 hearing loss gene-disease pairs.	Genet Med.	21(10)	2409	2019

S. B. Minami, K. Nara, H. Mutai, N. Morimoto, H. Sakamoto, T. Takiguchi, K. Kaga and T. Matsunaga.	A clinical and genetic study of 16 Japanese families with Waardenburg syndrome.	Gene.	704	86-90.	2019
J. Shen, A. M. Oza, I. Del Castillo, H. Duzkale, T. Matsunaga, A. Pandya, H. P. Kang, R. Mar-Heyming, S. Guha, K. Moyer, C. Lo, M. Kenna, J. J. Alexander, Y. Zhang, Y. Hirsch, M. Luo, Y. Cao, K. Wai Choy, Y. F. Cheng, K. B. Avraham, X. Hu, G. Garrido, M. A. Moreno-Pelayo, J. Greinwald, K. Zhang, Y. Zeng, Z. Brownstein, L. Basel-Salmon, B. Davidov, M. Frydman, T. Weiden, N. Nagan, A. Willis, S. E. Hemphill, A. R. Grant, R. K. Siegert, M. T. Distefano, S. S. Amr, H. L. Rehm, A. N. Abou Tayoun and G. Clingen Hearing Loss Working.	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel.	Genet Med.	21(11)	2442-2452.	2019
K. Wasano, S. Takahashi, S. K. Rosenberg, T. Kojima, H. Mutai, T. Matsunaga, K. Ogawa and K. Homma.	Systematic quantification of the anion transport function of pendrin (SLC26A4) and its disease-associated variants.	Hum Mutat.	41(1)	316-331.	2020
A. Maeda-Katahira, N. Nakamura, T. Hayashi, S. Katagiri, S. Shimizu, H. Ohde, T. Matsunaga, K. Kaga, T. Nakano, S. Kameya, T. Matsuura, K. Fujinami, T. Iwata and K. Tsunoda.	Autosomal dominant optic atrophy with OPA1 gene mutations accompanied by auditory neuropathy and other systemic complications in a Japanese cohort.	Mol Vis.	25	559-573.	2019
山本修子、南修司郎、榎本千江子、加藤秀敏、松永達雄、伊藤文展、遠藤理奈子、橋本陽介、石川直明、加我君孝.	東京医療センターにおける成人人工内耳症例の適応と有用性の検討.	日本耳鼻咽喉科学会会報	122(8)	1118-1126	2019
松永達雄*.	「第 119 回日本耳鼻咽喉科学会総会シンポジウム」難聴のゲノム医療	日本耳鼻咽喉科学会会報	122	16-21	2019
松永達雄*.	視覚聴覚二重障害の診療.	JOHNS	35(9)	1377-1378	2019
松永達雄*.	遺伝情報をどう管理し、活用するか―耳科領域―.	JOHNS	Vol. 35 No.10	1452-1454	2019
R. Kosaki, M. Kubota, T. Uehara, H. Suzuki, T. Takenouchi and K. Kosaki.	Consecutive medical exome analysis at a tertiary center: Diagnostic and health-economic outcomes.	Am J Med Genet A.			2020
S. Suzuki-Muromoto, R. Kosaki, K. Kosaki and M. Kubota.	Familial hemiplegic migraine with a PRRT2 mutation: Phenotypic	Brain Dev.	42(3)	293-297.	2020

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T. Niihori, K. Nagai, A. Fujita, H. Ohashi, N. Okamoto, S. Okada, A. Harada, H. Kihara, T. Arbogast, R. Funayama, M. Shirota, K. Nakayama, T. Abe, S. I. Inoue, I. C. Tsai, N. Matsumoto, E. E. Davis, N. Katsanis and Y. Aoki.	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome.	Am J Hum Genet.	104(6)	1233-1240.	2019
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M. Tominaga, T. Saito, M. Masuno, Y. Umeda and K. Kurosawa.	Developmental delay and dysmorphic features in a girl with a de novo 5.4 Mb deletion of 13q12.11-q12.13.	Congenit Anom (Kyoto).			2019

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T. Kosho, S. Mizumoto, T. Watanabe, T. Yoshizawa, N. Miyake and S. Yamada.	Recent Advances in the Pathophysiology of Musculocontractural Ehlers-Danlos Syndrome.	Genes (Basel).	11(1).		2019
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Y. Yokota, H. Moteki, S. Y. Nishio, T. Yamaguchi, K. Wakui, Y. Kobayashi, K. Ohyama, H. Miyazaki, R. Matsuoka, S. Abe, K. Kumakawa, M. Takahashi, H. Sakaguchi, N. Uehara, T. Ishino, T. Kosho, Y. Fukushima and S. I. Usami.	Frequency and clinical features of hearing loss caused by STRC deletions.	Sci Rep.	9(1)	4408	2019
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