

研究成果の刊行に関する一覧表

書籍

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
松永達雄	遺伝性難聴の診断の進歩	山岨達也	医学のあゆみBOOKS 耳鼻咽喉科診療の進歩 40のエッセンス	医歯薬出版	東京	2018	7-10
角田和繁	遺伝性網膜疾患の臨床診断		「あたらしい眼科」『網膜変性診療の未来予想図』	医学書院		2018	427-436
仁科幸子	新生児・乳児の眼科的異常	五十嵐隆	小児科診療ガイドライン—最新の治療指針—第4版	総合医学社	東京	2019	741-744
仁科幸子	先天白内障	大橋裕一・村上晶	眼科疾患 最新の治療 2019-2021,	南江堂	東京	2019	195
仁科幸子	未熟児網膜症—眼底検査法と写真撮影法・リハビリテーション・ロービジョンケア・類縁疾患	東範行	未熟児網膜症	三輪書店	東京	2018	
Kubota M	Cockayne Syndrome: Clinical Aspects.	Nishigori C., Sugawawa K.	DNA Repair Disorders	Springer	Singapore	2019	pp115-132
後藤美和子	章 基本的診療眼科問診	松永 達雄	先天性および若年性の視覚聴覚二重障害の原因となる難病の診療マニュアル				42-45
後藤美和子	章 特記すべき診療・療育・支援成人への移行における課題と眼科的対応	松永 達雄	先天性および若年性の視覚聴覚二重障害の原因となる難病の診療マニュアル				161-163

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Kitao K, Mutai H, Namba K, Morimoto N, Nakanishi A, Arimoto Y, Sugiuchi T, Masuda S, Okamoto Y, Morita N, Sakamoto H, Shintani T, Fukuda S, Kaga K, <u>Matsunaga T*</u>	Deterioration in Distortion Product Otoacoustic Emissions in Auditory Neuropathy Patients with Distinct Clinical and Genetic Backgrounds.	Ear Hear.	40(1)	184-191	2019
DiStefano MT, Hemphill SE, Ozdamar AM., Siegert R K., Grant AR., Hughes MY., Cushman BJ., Azaiez H, Booth KT., Chahin A, Duzkale H, <u>Matsunaga T</u> , Shen J, Zhang W, Kenna M, Schimmenti LA., Tekin M, Rehm HL., Abou Tayoun AN., Amr SS* on behalf of the ClinGen Hearing Loss Clinical Domain Working Group.	ClinGen Expert Clinical Validity Curation of 164 Hearing Loss Gene-Disease Pairs. Genet Med.	doi: 10.1038/s41436-019-0487-0			2019
Minami S, Nara K, Mutai H, Morimoto N, Sakamoto H, Takiguchi T, Kaga K, <u>Matsunaga T*</u>	A clinical and genetic study of 16 Japanese families with Waardenburg syndrome.	Gene.	708(1)	86-90	2019
Matsushima K, Nakano A, Arimoto Y, Mutai H, Yamazawa K, Murayama K, <u>Matsunaga T*</u>	High-level heteroplasmism for the m.7445A>G mitochondrial DNA mutation can cause progressive sensorineural hearing loss in infancy.	Int J Pediatr Otorhinolaryngol	108	125-131	2018
Morimoto N, Mutai H, Namba K, Kaneko H, Kosaki R, <u>Matsunaga T*</u>	Homozygous EDNRB Mutation in a Patient with Waardenburg Syndrome Type 1.	Auris Nasus Larynx	45(2)	222-226	2018

Suzuki N, Mutai H, Miya F, Tsunoda T, Terashima H, Morimoto N, <u>Matsunaga T*</u>	A case report of reversible generalized seizure in a patient with Wardeburg syndrome associated with a novel nonsense mutation in the penultimate exon of SOX10.	BMC Pediatr.	18(1)	171	2018
Oza AM, DiStefano MT, Hemphill SE, Cushman B, Grant AR, Siebert RK, Shen J, Chapin A, Boczek NJ, Schimmenti LA, Murry JB, Hasadsri L, Narasimhan K, Kenna M, Booth KT, Azaiez H, Griffith A, Avraham KB, Kremer H, Rehm HL, Amr SS, Abou Tayoun AN*, <u>Clin Gen Hearing Loss Clinical Domain Working Group</u>	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss.	Hum Mutat.	39(11)	1593-1613	2018
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<u>松永達雄</u>	先天性難聴児のゲノム診療の意義と動向 .	公衆衛生	82(6)	468-473	2018
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<u>松永達雄</u>	臨床像起点の遺伝性難聴診療の確立	Otol Jpn.	28(2)	65-69	2018
<u>松永達雄</u>	ゲノム医療(遺伝子医療)の今 希少疾患・難病の遺伝カウンセリング	保健の科学	60(10)	677-681	2018

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Mahroo OA, <u>Fujinami K*</u> , Moore AT, Webster AR.	Retinal findings in a patient with mutations in ABCC6 and ABCA4.	Eye (Lond).	32	1542-1543.	2018
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Tanna P, Georgiou M, Strauss RW, Ali N, Kumaran N, Kalitzeos A, <u>Fujinami K</u> , Michaelides M.	Cross-Sectional and Longitudinal Assessment of the Ellipsoid Zone in Childhood-Onset Stargardt Disease.	Transl Vis Sci Technol.	8	1	2019
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<u>廣田栄子</u> ・ <u>齋藤佐和</u> 他	我が国における聴覚障害児の早期介入の現状	リハビリテーション連携科学	19(1)	inpress	2019
<u>守本倫子</u>	小児難聴	ファルマシア	54	1035-1039	2018
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