

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
倉橋浩樹	網羅的手法による次世代型着床前診断	荒木康久	着床前診断検査 (PGT-A) の基礎知識と細胞分離手技	医歯薬出版	東京	In press	
倉橋浩樹	小児科診療における遺伝カウンセリング		今日の小児治療指針第17版	医学書院	東京	In press	
山本俊至	11p13欠失症候群 (WAGR症候群)		内分泌症候群 (3版) IV-その他の内分泌疾患を含めて	(株)日本臨床社	大阪	In press	

雑誌

研究代表者・倉橋浩樹

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Katagiri S, Iwasa M, Hayashi T, Hosono K, Yamashita T, Kuniyoshi K, Ueno S, Kondo M, Ueyama H, Ogita H, Shichida Y, Inagaki H, Kurahashi H, Kondo H, Ohji M, Hotta Y, Nakano T.	Genotype determination of the OPN1LW/OPN1MW genes: novel disease-causing mechanisms in Japanese patients with blue cone monochromacy.	Sci Rep	8(1)	11507	2018

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Tsutsumi M, Fujita N, Suzuki F, Mishima T, Fujieda S, Watarai M, Takahashi N, Tonoki H, Moriwaka O, Endo T, <u>Kurahashi H</u> .	A constitutional jumping translocation involving the Y and acrocentric chromosomes.	Asian J Androl	In press.	In press.	2019
Yokoi K, Nakajima Y, Ohye T, Inagaki H, Wada Y, Fukuda T, Sugie H, Yuasa I, Ito T, <u>Kurahashi H</u> .	Disruption of the responsible gene in a phosphoglucomutase 1 deficiency patient	JIMD Rep	In press.	In press.	2019
Hayano S, Okuno Y, Tsutsumi M, Inagaki H, Fukasawa Y, <u>Kurahashi H</u> , Kojima S, Takahashi Y, Kato T.	Frequent intragenic microdeletions of elastin in familial supravalvular aortic stenosis.	Int J Cardiol	In press.	In press.	2019
Miura H, Kawamura Y, Hattori F, Kozawa K, Ihira M, Ohye T, <u>Kurahashi H</u> , Yoshikawa T.	Chromosomally integrated human herpesvirus 6 in the Japanese population.	J Med Virol.	90(10)	1636-1642	2018
Kibe M, Ibara S, Inagaki H, Kato T, <u>Kurahashi H</u> , Ikeda T.	Lethal persistent pulmonary hypertension of the newborn in Bohring-Opitz syndrome	Am J Med Genet A	176(5)	1245-1248	2018
Kawai M, Tsutsumi M, Suzuki F, Sameshima K, Dowa Y, Kyoya T, Inagaki H, <u>Kurahashi H</u> .	Two siblings with 11qter deletion syndrome that had been rescued in their mother by uniparental disomy.	Eur J Med Genet	S1769-7212(18)	30164-2	2018
Boda H, Miyata M, Inagaki H, Shinkai Y, Kato T, Yoshikawa T, <u>Kurahashi H</u> .	FOXA2 gene mutation in a patient with congenital complex pituitary hormone deficiency.	Eur J Med Genet	S1769-7212(18)	30408-7	2018
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Ohwaki A, Nishizawa H, Aida N, Kato T, Kambayashi A, Miyazaki J, Ito M, Urano M, Kiriya Y, Kuroda M, Nakayama M, Sonta SI, Suzumori K, Sekiya T, <u>Kurahashi H.</u> , Fujii T.	Twin pregnancy with chromosomal abnormalities mimicking a gestational trophoblastic disorder and coexistent foetus on ultrasound.	J Obstet Gynaecol	38(7)	1023-1025	2018
Yamaguchi T, Yamaguchi M, Akeno K, Fujisaki M, Sumiyoshi K, Ohashi M, Sameshima H, Ozaki M, Kato M, Kato T, Hosoba E, <u>Kurahashi H.</u>	Prenatal diagnosis of premature chromatid separation/mosaic variegated aneuploidy (PSCS/MVA) syndrome.	J Obstet Gynaecol Res.	44(7)	1313-1317	2018
Terasawa S, Kato A, Nishizawa H, Kato T, Yoshizawa H, Noda Y, Miyazaki J, Ito M, Sekiya T, Fujii T, <u>Kurahashi H.</u>	Multiplex PCR in non-invasive prenatal diagnosis for FGFR3-related disorders.	Congenit Anom (Kyoto).	In press.	In press.	2019
Noda Y, Kato T, Kato A, Nishizawa H, Miyazaki J, Ito M, Terasawa S, Sekiya T, Fujii T, <u>Kurahashi H.</u>	Potentially effective method for fetal gender determination by non-invasive prenatal testing for X-linked disease.	Congenit Anom (Kyoto)	In press.	In press.	2019
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Hitachi K, Nakatani M, Takasaki A, Ouchi Y, Uezumi A, Ageta H, Inagaki H, <u>Kurahashi H.</u> , Tsuchida K.	Myogenin promoter-associated lncRNA Myoparr is essential for myogenic differentiation.	EMBO Rep	In press.	In press.	2019
Inagaki H, Ota S, Nishizawa H, Miyamura H, Nakahira K, Suzuki M, Nishiyama S, Kato T, Yanagihara I, <u>Kurahashi H.</u>	Obstetric complication-associated ANXA5 promoter polymorphisms may affect gene expression via DNA secondary structures.	J Hum Genet	In press.	In press.	2019

研究分担者・大橋博文

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研究分担者・涌井敬子

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