別紙４

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
Yamamoto M, Takahashi Y	Pituitary-Immune interface	S Meled	***The Pituitary*** 5th edition	Elsevier	USA	2022	375-384
Takahashi Y	Autoimmune paraneoplastic hypophysitis	S Meled	***Williams text book of Endocrinology*** 15th edition	Elsevier	USA	2022	In press

雑誌

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Hiraide T, Masunaga Y, Honda A, Kato F, Fukuda T, Fukami M, Nakashima M, Saitsu H, Ogata T*	Retrotransposition disrupting EBP in a girl and her mother with X-linked dominant chondrodysplasia punctata.	J Hum Genet	67(5)	303-306	2022
Yamoto K, Okada S, Kato F, Fujisawa Y, Fukami M, Saitsu H, Ogata T*	A novel intronic PORCN variant creating an alternative splice acceptor site in a mother and her daughter with focal dermal hypoplasia.	Am J Med Genet A	188(5)	1612-1617	2022
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Fujisawa Y, Ono H, Konno A, Yao I, Itoh H, Baba T, Morohashi K, Katoh-Fukui Y, Miyado M, Fukami M, Ogata T*	Intrauterine hyponutrition reduces fetal testosterone production and postnatal sperm count in the mouse.	J Endocr Soc	6(4)	bvac022	2022
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Fukami M, Shindo J, Ogata T, Kageyama I, Kamimaki T	SHOX far-downstream deletion in a patient with non-syndromic short stature.	Am J Med Genet A	188(7)	2173-2177	2022
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Sato T, Ishii T, Fukami M, Ogata T, Hasegawa T*	The first adult case of cytochrome P450 oxidoreductase deficiency with sufficient semen volume and sperm concentration.	Congenit Anom (Kyoto)	62(3)	136-137	2022
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Hara-Isono K, Nakamura A, Fuke T, Inoue T, Kawashima S, Matsubara K, Sano S, Yamazawa K, Fukami M, Ogata T, Kagami M*	Pathogenic copy number and sequence variants in children born SGA with short stature without imprinting disorders.	J Clin Endocrinol Metab	107(8)	e3121-e3133	2022

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Mackay D, Bliek J, Kagami M, Tenorio-Castano J, Pereda A, Brioude F, Netchine I, Papingi D, de Franco E, Lever M, Sillibourne J, Lombardi P, Gaston V, Tauber M, Diene G, Bieth E, Fernandez L, Nevado J, Tümer Z, Riccio A, Maher ER, Beygo J, Tannorella P, Russo S, de Nanclares GP, Temple IK, Ogata T, Lapunzina P, Eggermann T*	First step towards a consensus strategy for multi-locus diagnostic testing of imprinting disorders.	Clin Epigenetics	14(1)	143	2022
Fukahori K, Yamoto K, Saitsu H, Ogata T, Nagasaki K*	PORCN-related microphthalmia with limb anomalies: case report and literature review.	Am J Med Genet A	191(2)	636-639	2022
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Kawai M*, Muroya K, Murakami N, Ihara H, Takahashi Y, Horikawa R, Ogata T	A questionnaire-based survey of medical conditions in adults with Prader-Willi syndrome in Japan: Implications for transitional care.	Endocr J	doi: 10.1507/endocrj.EJ22-0561		2023
Hiraide T, Shimizu K, Okumura Y, Miyamoto S, Nakashima M, Ogata T, Saistu H*	A deep intronic TCTN2 variant activating a cryptic exon predicted by SpliceRover in a patient with Joubert syndrome.	J Hum Genet	doi: 10.1038/s10038-023-01143-3		2023
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Kawai M, Etani Y, Ida S.	Subcutaneous adipose tissue is a positive predictor for bone mineral density in prepubertal children with Prader-Willi syndrome independent of lean mass.	J Pediatr Endocrinol Metab	35	603-609	2022
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