

研究成果の刊行に関する一覧表レイアウト（参考）

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 |
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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 |
|--|--|-----------------------|--------|-----------|------|
| Ohtaka K, Fujisawa Y, Takada F, Hasegawa Y, Miyoshi T, Hasegawa T, Miyoshi H, Kameda H, Kurokawa-Seo M, Fukami M, Ogata T* | <i>FGFR1</i> Analyses in Four Patients with Hypogonadotropic Hypogonadism with Split-Hand/Foot Malformation: Implications for the Promoter Region. | <i>Hum Mutat</i> | 38(5) | 503-506 | 2017 |
| Fukami M, Suzuki E, Izumi Y, Torii T, Narumi S, Igarashi M, Miyado M, Katsumi M, Fujisawa Y, Nakabayashi K, Hata K, Umezawa A, Matsubara Y, Yamauchi J, Ogata T | Paradoxical gain-of-function mutant of the G-protein coupled receptor PROKR2 promotes early puberty. | <i>J Cell Mol Med</i> | 21(10) | 2623-2626 | 2017 |
| Nakamura S, Miyado M, Saito K, Katsumi M, Nakamura A, Kobori Y, Tanaka Y, Ishikawa H, Yoshida A, Okada H, Hata K, Nakabayashi K, Okamura K, Ogata H, Matsubara Y, <u>Ogata T</u> , Nakai H, Fukami M | Next-generation sequencing for patients with non-obstructive azoospermia: implications for significant roles of monogenic/oligogenic mutations. | <i>Andrology</i> | 5(4) | 824-831 | 2017 |

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|--|---|--|---------|------------|------|
| Shozu M*, Ishikawa H, Horikawa R, Sakakibara H, Izumi SI, Ohba T, Hirota Y, <u>Ogata T</u> , Osuga Y, Kugu K | Nomenclature of primary amenorrhea: a proposal document of the Japan Society of Obstetrics, and Gynecology committee for the redefinition of primary amenorrhea. | <i>J Obstet Gynaecol Res</i> | 43(11) | 1738-1742 | 2017 |
| Yamamoto K, Saitsu H, Nakagawa N, Nakajima H, Hasegawa T, Fujisawa Y, Kagami M, Fukami M, <u>Ogata T*</u> | <i>De Novo IGF2</i> Mutation on the Paternal Allele in a Patient with Silver-Russell Syndrome and Ectrodactyly. | <i>Hum Mutat</i> | 38(8) | 953-958 | 2017 |
| <u>Miyado M</u> , <u>Yoshida K</u> , <u>Miyado K</u> , <u>Katsumi M</u> , <u>Saito K</u> , Nakamura S, <u>Ogata T</u> , <u>Fukami M*</u> | Knockout of murine Mamld1 impairs testicular growth and daily sperm production but permits normal postnatal androgen production and fertility. | <i>Int J Mol Sci</i> | 18(6) | pii: E1300 | 2017 |
| Suzuki E, Bo R, Sue K, Awano H, <u>Ogata T</u> , Narumi S, Kagami M, Sano S, Fukami M* | <i>De Novo</i> 50-bp <i>GNAS</i> (<i>Gs-alpha</i>) Intragenic Duplication in a Patient with Pseudohypoparathyroidism Type 1a. | <i>Cytogenet Genome Res</i> | 153(3) | 125-130 | 2017 |
| Sakata S, Okada S*, Aoyama K, Hara K, Tani C, Kagawa R, Utsunomiya-Nakamura A, Miyagawa S, <u>Ogata T</u> , Mizuno H, Kobayashi M | Individual Clinically Diagnosed with CHARGE syndrome but with a Mutation in <i>KMT2D</i> , a Gene Associated with Kabuki syndrome: A Case Report. | <i>Front Genet</i> (Genetic Disorders Section) | 11 | 210 | 2017 |
| Takasawa K, Gau M, Sutani A, Igarashi M, Ono M, Takemoto A, Takada S, Yamataka A, <u>Ogata T</u> , Morio T, Fukami M, Kashimada K* | Phenotypic variation in 46,XX disorders of sex development due to the <i>NR5A1</i> p.R92W variant: a sibling case report and literature review. | <i>Sex Dev</i> | 11(5-6) | 284-288 | 2017 |
| Ohsako S*, Aiba T, Miyado M, Fukami M, <u>Ogata T</u> , Hayashi Y, Mizuno K, Kojima Y | <u>Expression of Xenobiotic Biomarkers CYP1 Family in Preputial Tissue of Patients with Hypospadias and Phimosi and Its Association with DNA Methylation Level of <i>SRD5A2</i> Minimal Promoter.</u> | <i>Arch Environ Contam Toxicol</i> | 74(2) | 240-247 | 2018 |

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|---|---|-----------------------------------|--------|---------|------|
| Yamamoto K, Okamoto S, Fujisawa Y, Fukami M, Saitsu H, <u>Ogata T*</u> | <i>FGFR1</i> Disruption Identified by Whole Genome Sequencing in a Male With a Complex Chromosomal Rearrangement and Hypogonadotropic Hypogonadism. | <i>Am J Med Genet A</i> | 176(1) | 139-143 | 2018 |
| Shima H, MD*, Koehler K*, Nomura Y, Sugimoto K, Satoh A, <u>Ogata T</u> , Fukami M, Schuelke M, Huebner A, Narum S | Two patients with MIRAGE syndrome lacking haematological features: role of somatic second-site reversion <i>SAMD9</i> mutations. | <i>J Med Genet</i> | 55(2) | 81-85 | 2018 |
| Ono H, Numakura C, Homma K, Hasegwa T, Tsutsumi S, Kato F, Fujisawa Y, Fukami M, <u>Ogata T*</u> | Longitudinal Serum and Urine Steroid Metabolite Profiling in a 46,XY Infant with Prenatally Identified <i>POR</i> Deficiency. | <i>J Steroid Biochem Mol Biol</i> | 178 | 177-184 | 2018 |
| <u>Nakamura S</u> , <u>Kobori Y</u> , <u>Ueda Y</u> , <u>Tanaka Y</u> , <u>Ishikawa H</u> , <u>Yoshida A</u> , <u>Katsumi M</u> , <u>Saito K</u> , <u>Nakamura A</u> , <u>Ogata T</u> , <u>Okada H</u> , <u>Nakai H</u> , <u>Miyado M</u> , <u>Fukami M*</u> | <i>STX2</i> is a causative gene for non-obstructive azoospermia. | <i>Hum Mutat</i> | | | 2018 |
| Ono H, Saitsu , <u>Horikawa R</u> , <u>Nakashima S</u> , <u>Ohkubo Y</u> , <u>Yanagi K</u> , <u>Nakabayashi K</u> , <u>Fukami M</u> , <u>Fujisawa Y</u> , <u>Ogata T*</u> | Partial androgen insensitivity syndrome caused by a deep intronic mutation creating an alternative splice acceptor site of the <i>AR</i> gene. | <i>Sci Rep</i> | 8(1) | 2287 | 2018 |
| Fukami M*, Shima H, Suzuki E, <u>Ogata T</u> , Matsubara K, Kamimaki T | Catastrophic Cellular Events Leading to Complex Chromosomal Rearrangements in the Germline. | <i>Clin Genet</i> | 91(5) | 653-660 | 2017 |
| Fukami M*, Suzuki E, Igarashi M, Miyado M, <u>Ogata T</u> | Gain-of-Function Mutations in G-protein Coupled Receptor Genes Associated with Human Endocrine Disorders. | <i>Clin Endocrinol</i> | 88(3) | 351-359 | 2018 |