雑誌

著者氏名	論文タイトル	発表誌名	巻・頁・出版年
Abe S, Miura K, Kinoshita A, Mishima H, Miura S, Yamasaki K, Hasegawa Y, Higashijima A, Jo O, Yoshida A, Kaneuchi M, Yoshiura KI, Masuzaki H.	Single human papillomavirus 16 or 52 infection and later cytological findings in Japanese women with NILM or ASC-US.	J. Hum. Genet	Epub ahead of print
Tsukamoto O, Miura K, Mishima H, Abe S, Kaneuchi M, Higashijima A, Miura S, Kinoshita A, Yoshiura K, Masuzaki H.	Identification of endometrioid endometrial carcinoma-associated microRNAs in tissue and plasma.	Gynecol Oncol.	PubMed - in process
Miura K, Higashijima A, Miura S, Mishima H, Yamasaki K, Abe S, Hasegawa Y, Kaneuchi M, Yoshida A, Kinoshita A, Yoshiura KI, Masuzaki H.	Predominantly placenta-expressed mRNAs in maternal plasma as predictive markers for twin-twin transfusion syndrome.	Prenat Diagn.	Epub ahead of print
Hamaguchi D, Miura K, Abe S, Kinoshita A, Miura S, Yamasaki K, Yoshiura K, Masuzaki H.	Initial viral load in cases of single human papillomavirus 16 or 52 persistent infection is associated with progression of later cytopathological findings in the uterine cervix.	J Med Virol.	85: 2093-2100, 2013

著者氏名	論文タイトル	発表誌名	巻・頁・出版年
Abe S, Miura K, Kinoshita A, Mishima H, Miura S, Yamasaki K, Hasegawa Y, Higashijima A, Jo O, Sasaki K, Yoshida A, Yoshiura K, Masuzaki H.	Copy number variation of the antimicrobial-gene, defensin beta 4, is associated with susceptibility to cervical cancer.	J Hum Genet.	58: 250-253, 2013
Higashijima A, Miura K, Mishima H, Kinoshita A, Jo O, Abe S, Hasegawa Y, Miura S, Yamasaki K, Yoshida A, Yoshiura K, Masuzaki H.	Characterization of placenta-specific microRNAs in fetal growth restriction pregnancy.	Prenat Diagn.	33: 214-222, 2013
Shimizu K, Wakui K, **Kosho T (corresponding author)**, Okamoto N, Mizuno S, Itomi K, Hattori S, Nishio K, Samura O, Kobayashi Y, Kako Y, Arai T, Oishi T, Kawame H, Narumi Y, Ohashi H, Fukushima Y.	Microarray and FISH-based genotype-phenotype analysis of 22 Japanese patients with Wolf-Hirschhorn syndrome.	Am J Med Genet Part A	in press

著者氏名	論文タイトル	発表誌名	巻・頁・出版年
Nishi E, Takamizawa S, Iio K, Yamada Y, Yoshizawa K, Hatata T, Hiroma T, Mizuno S, Kawame H, Fukushima Y, Nakamura T, Kosho T (corresponding author).	Surgical intervention for esophageal atresia in patients with trisomy 18.	Am J Med Genet Part A	Epub ahead of print
Kosho T (corresponding author), Kuniba H, Tanikawa Y, Hashimoto Y, Sakurai H.	Natural history and parental experience of children with trisomy 18 based on a questionnaire given to a Japanese trisomy 18 parental support group.	Am J Med Genet Part A	161A(7): 1531-1542, 2013.
Kosho T (corresponding author), Okamoto N, Ohashi H, Tsurusaki Y, Imai Y, Hibi-Ko Y, Kawame H, Homma T, Tanabe S, Kato M, Hiraki Y, Yamagata T, Yano S, Sakazume S, Ishii T, Nagai T, Ohta T, Niikawa N, Mizuno S, Kaname T, Naritomi K, Narumi Y, Wakui K, Fukushima Y, Miyatake S, Mizuguchi T, Saitsu H, Miyake N, Matsumoto N.	Clinical correlations of mutations affecting six components of the SWI/SNF complex: detailed description of 21 patients and a review of the literature.	Am J Med Genet Part A	161A(6): 1221-1237, 2013.

著者氏名	論文タイトル	発表誌名	巻・頁・出版年
Tsurusaki Y, Kosho T (equal contribution, corresponding author), Hatasaki K, Narumi Y, Wakui K, Fukushima Y, Doi H, Saitsu H, Miyake N, Matsumoto N.	Exome sequencing in a family with an X-linked lethal malformation syndrome: clinical consequences of hemizygous truncating OFD1 mutations in male patients.	Clin Genet.	83(2): 135-144, 2013.
Higashimoto K, Jozaki K, Kosho T, Matsubara K, Fuke T, Yamada D, Yatsuki H, Maeda T, Ohtsuka Y, Nishioka K, Joh K, Koseki H, Ogata T, Soejima H.	A novel de novo point mutation of the OCT-binding site in the IGF2/H19-imprinting control region in a Beckwith-Wiedemann syndrome patient.	Clin Genet	Epub ahead of print
Sugiura K, Takeichi T, Tanahashi K, Ito Y, Kosho T, Saida K, Uhara H, Okuyama R, Akiyama M.	Lamellar ichthyosis in a collodion baby caused by CYP4F22 mutations in a non-consanguineous family outside the Mediterranean.	J Dermatol Sci.	72(2): 193-195, 2013.
Nitta H, Unoki M, Ichiyanagi K, Kosho T, Shigemura T, Takahashi H, Velasco	hree novel ZBTB24 mutations identified in Japanese and Cape Verdean type 2	J Hum Genet.	58(7): 455-460, 2013.

著者氏名	論文タイトル	発表誌名	巻・頁・出版年
G, Francastel C, Picard C, Kubota T, Sasaki H.	ICF syndrome patients.		
Miyake N, Koshimizu E, Okamoto N, Mizuno S, Ogata T, Nagai T, Kosho T, Ohashi H, Kato M, Sasaki G, Mabe H, Watanabe Y, Yoshino M, Matsuishi T, Takanashi J, Shotelersuk V, Tekin M, Ochi N, Kubota M, Ito N, Ihara K, Hara T, Tonoki H, Ohta T, Saito K, Matsuo M, Urano M, Enokizono T, Sato A, Tanaka H, Ogawa A, Fujita T, Hiraki Y, Kitanaka S, Matsubara Y, Makita T, Taguri M, Nakashima M, Tsurusaki Y, Saitsu H, Yoshiura K, Matsumoto N, Niikawa N.	MLL2 and KDM6A mutations in patients with Kabuki syndrome.	Am J Med Genet Part A	61(9): 2234-2243, 2013.
Tanaka K, Sekijima Y, Yoshida K, Tamai M, Kosho T, Sakurai A, Wakui K, Ikeda S, Fukushima Y.	Follow-up nationwide survey on predictive genetic testing for late-onset hereditary neurological diseases in Japan.	J Hum Genet.	58(8): 560-563, 2013.

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
Kosho T (corresponding author), Mizumoto S, Sugahara K.	Carbohydrate (N-acetylgalactosamine 4-O) sulfotransferase 14 (CHST14).	Taniguchi N, Honke K, Fukuda M, Narimatsu H, Yamaguchi Y, Angata T	Handbook of glycosyltransferases and related genes	Springer	Germany	In press	
Kosho T (corresponding author).	Discovery and delineation of dermatan 4-*O*-sulfotransferase-1 (D4ST1)-deficient Ehlers-Danlos syndrome.	Oiso N, Kawada A	Current Genetics in Dermatology	InTech	Croatia	2013	73-86