II. 研究成果の刊行に関する一覧表

書籍

なし

診断基準作成

大薗恵一　指定難病の「軟骨無形成症の診断基準」2015.7.
澤井英明　指定難病の「タナトフォリック骨異形成症の診断基準」2015.7

雑誌

Nobuzane T, Yamada T, Miura K, Sawai H, Masuzaki H, Kudo Y. Survey of prenatal testing for genetic disorders in Japan: Recent report. J Obstet Gynaecol Res. 2016 Feb 18.

Saito T, Nagasaki K, Nishimura G, Wada M, Nyuzuki H, Takagi M, Hasegawa T, Amano N, Murotsuki J, Sawai H, Yamada T, Sato S, Saitoh A. Criteria for radiologic diagnosis of hypochondroplasia in neonates. Pediatr Radiol. 2016 Feb 11. [Epub ahead of print]

Takagi M, Kouwaki M, Kawase K, Shinohara H, Hasegawa Y, Yamada T, Fujiwara I, Sawai H, Nishimura G, Hasegawa T. A novel mutation Ser344Cys in FGFR3 causes achondroplasia with severe platyspondyly. Am J Med Genet A. 2015 Nov;167A(11):2851-4.

Sago H, Sekizawa A; Japan NIPT consortium. Nationwide demonstration project of next-generation sequencing of cell-free DNA in maternal plasma in Japan: 1-year experience. Prenat Diagn. 2015 Apr;35(4):331-6.

Okada M, Ikegawa S, Morioka M, Yamashita A, Saito A, Sawai H, Murotsuki J, Ohashi H, Okamoto T, Nishimura G, Imaizumi K, Tsumaki N. Modeling type II collagenopathy skeletal dysplasia by directed conversion and induced pluripotent stem cells. Hum Mol Genet. 2015 Jan 15;24(2):299-313.

Inokuchi H, Tojima M, Mano H, Ishikawa Y, Ogata N, Haga N: Neck range of motion measurements using a new three-dimensional motion analysis system:
validity and repeatability. Eur Spine J, 2015 Apr 7. [Epub ahead of print]

Ko JM, Bae JS, Choi JS, Miura K, Lee HR, Kim OH, Kim NK, Oh SK, Ozono K, Lee CK, Choi IH, Park WY, Cho TJ. Skeletal overgrowth syndrome caused by overexpression of C-type natriuretic peptide in a girl with balanced chromosomal translocation, t(1;2)(q41;q37.1). Am J Med Genet A. 2015 May;167A(5):1033-8.

Fukami M, Naiki Y, Muroya K, Hamajima T, Soneda S, Horikawa R, Jinno T, Katsumi M, Nakamura A, Asakura Y, Adachi M, Ogata T, Kanzaki S; Japanese SHOX study group. Rare pseudoautosomal copy-number variations involving SHOX and/or its flanking regions in individuals with and without short stature. J Hum Genet. 2015

Tamura M, Isojima T, Kawashima M, Yoshida H, Yamamoto K, Kitaoka T, Namba N, Oka A, Detection of Hereditary 1,25-Hydroxyvitamin D-Resistant Rickets Caused by Uniparental Disomy of Chromosome 12 Using Genome-Wide Single Nucleotide Polymorphism Array. Ozono K, Tokunaga K, Kitanaka S.
PLoS One. 2015 Jul 8;10(7):e0131157.

Imai R, Miyazaki O, Horiuchi T, Kurosawa H, Nosaka S: Local diagnostic reference level (DRL) based on size-specific dose estimates (SSDE): Assessment of pediatric abdominal/pelvic computed tomography (CT) at a Japanese national children's hospital. Pediatr Radiol 2015; 45: 345-353 Akaishi R, Yamada T, Kawaguchi S, Kojima T, Koyama T, Umazume T, Morikawa M, Cho K, Minakami H. Uptake of non-invasive prenatal testing by Japanese women. Ultrasound Obstet Gynecol. 2015; 45:113-4.

Yamada T, Cho K, Morikawa M, Yamada T, Minakami H. Intrapartum risk factors for neonat

al encephalopathy leading to cerebral palsy in women without apparent sentinel events. J Obstet Gynaecol Res. 2015 Jul 14. doi: 10.1111/jog.12772. [Epub ahead of print]

Yamada T, Abe K, Baba Y, Inubashiri E, Kawabata K, Kubo T, Maegawa Y, Fuchi N, Nomizo M, Shimada M, Shiozaki A, Hamada H, Matsubara S, Akutagawa N, Kataoka S, Maeda M, Masuzaki H, Sagawa N, Nakai A, Saito S, Minakami H. Vaccination during the 2013-2014 influenza season in pregnant Japanese women. Eur J Clin Microbiol Infect Dis. 2015 Mar;34(3):543-8.

Tsuda H, Kotani T, Sumigama S, Mano Y, Kawabata I, Takahashi Y, Iwagaki S, Hirakawa A, Kikkawa F. Amniotic lamellar body count: predicting and distinguishing neonatal respiratory complications in twin pregnancies. Clin Chim 2015 20;75-8.