

## 原著 (英文)

●平成27年度 (2015.4~2016.3) ●

● 1) Chemical chaperone treatment for galactosialidosis : effect of NOEV on  $\beta$  galactosidase activities in fibroblasts.

Hossain MA, Higaki K※, Shinpo M, Nanba E※, Suzuki Y※, Ozono K, Sakai N.  
Brain Dev, 38 (2) : 175-180, 2016.

● 2) Pathogenesis and diagnostic criteria for rickets and osteomalacia-proposal by an expert panel supported by the ministry of health, labour and welfare, Japan, the Japanese Society for Bone and Mineral Research, and the Japan Endocrine Society.

Fukumoto S※, Ozono K, Michigami T※, Minagawa M※, Okazaki R※, Sugimoto T※, Takeuchi Y※, Matsumoto T※.  
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● 3) Detection of hereditary 1, 25-hydroxyvitamin D-resistant rickets caused by uniparental disomy of Chromosome 12 using Genome-Wide Single Nucleotide Polymorphism Array.

Tamura M※, Isojima T※, Kawashima M※, Yoshida H※, Yamamoto K, Kitaoka T, Namba N, Oka A※, Ozono K, Tokunaga K※, Kitanaka S※.  
PLoS One, 10 (7) : e0131157, 2015.

● 4) Nationwide survey of fibroblast growth factor 23 (FGF23) -related hypophosphatemic diseases in Japan : prevalence, biochemical data and treatment.

Endo I※, Fukumoto※, Ozono K, Namba N, Inoue D※, Okazaki R※, Yamauchi M※, Sugimoto T※, Minagawa M※, Michigami T※, Nagai M※, Matsumoto T※.  
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● 5) Chaperone therapy for Krabbe disease : potential for late-onset GALC mutations.

Hossain MA, Higaki K※, Saito S※, Ohno K※, Sakuraba H※, Nanba E※, Suzuki Y※, Ozono K, Sakai N.  
J Hum Genet, 60 (9) : 539-545, 2015.

● 6) Rare pseudoautosomal copy-number variations involving SHOX and/or its flanking regions in individuals with and without short stature.

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.  
J Hum Genet, 60 (9) : 553-556, 2015.

● 7) Treatment situation of male hypogonadotropic hypogonadism in pediatrics and proposal of testosterone and gonadotropins replacement therapy protocols.

Sato N※, Hasegawa T※, Hasegawa Y※, Arisaka O※, Ozono K, Amemiya S※, Kikuchi T※, Tanaka H※, Harada S※, Miyata I※, Tanaka T※.

Clin Pediatr Endocrinol, 24 (2) : 37-49, 2015.

● 8) Prognostic factors for acute encephalopathy with bright tree appearance.

Azuma J※, Nabatame S, Nakano S※, Iwatani Y※, Kitai Y※, Tominaga K, Kagitani-Shimono K, Okinaga T, Yamamoto T※, Nagai T※, Ozono K.

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Yamazaki M※, Kawai M※, Miyagawa K※, Ohata Y, Tachikawa K※, Kinoshita S※, Nishino J※, Ozono K, Michigami T※.

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● 11) Negative feedback loop of cholesterol regulation is impaired in the livers of patients with Alagille syndrome.

Miyahara Y, Bessho K, Kondou H, Hasegawa Y, Yasuda K, Ida S※, Ihara Y※, Mizuta K※, Miyoshi Y, Ozono K.

Clin Chim Acta, 440 : 49-54, 2015.

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Kuroyanagi Y※, Kawasaki H※, Noda Y※, Ohmachi T※, Sekiya S※, Yoshimura K※, Ohe C※, Michigami T※, Ozono K, Kaneko K※.

Tohoku J Exp Med, 234 (4) : 309-312, 2014.

● 13) Skeletal overgrowth syndrome caused by overexpression of C-type natriuretic peptide in a girl with balanced chromosomal translocation, t(1;2)(q41;q37.1) .

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※.

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J Matern Fetal Neonatal Med, 29 (6) : 982-986, 2016.

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