

## 平成27年度 抄読会 (毎週水曜日 17:30~)

○April 15, 2015

### T. Ito (伊藤智城)

A Narita, et al.

Abnormal pupillary light reflex with chromatic pupillometry in Gauche disease.  
Ann Clin Transl Neurol. 1(2):135-140, 2014

○April 22, 2015

### M. Narugami (鳴神雅史)

H Sugano, et al.

Posterior quadrant disconnection surgery for Sturge-Weber syndrome.  
Epilepsia 55(5): 683-9, 2014

### K. Terada (寺田健作)

D.W. Kimberlin, et al.

Valganciclovir for Symptomatic Congenital Cytomegalovirus Disease.  
N Engl J Med 372:933-943, 2015

○May 13, 2015

### M. Yamada (山田雅文)

T Suzuki, et al.

Pulmonary macrophage transplantation therapy. Nature 514(7523):450-4, 2014

CM Doerschuk.

Pulmonary alveolar proteinosis and macrophage transplantation.

N Engl J Med. 372(18):1762-4, 2015

### S. Morikawa (森川俊太郎)

K Ikegami, et al.

Tissue-specific posttranslational modification allows functional targeting of thyrotropin.  
Cell Rep 9(3):801-10, 2014

○June3, 2015

A. Iguchi (井口晶裕)

Shannon L. Maude, et al.

Chimeric Antigen Receptor T Cells for Sustained Remissions in Leukemia.

N Engl J Med 371:1507-1517, 2014

G. Izumi (泉 岳)

MC. Escobar-Diaz, et al.

Perinatal Outcome in Fetuses with Heterotaxy Syndrome and Atrioventricular Block or Bradycardia. Pediatr Cardiol 35(6):906-913, 2014

○June 10, 2015

K. Cho (長 和俊)

L. Enaud, et al.

Pulmonary alveolar proteinosis in children on La R .A Niunion Island: a new inherited disorder? Orphanet J Rare Dis. 9:85, 2014

○June 17, 2015

K. Morioka (森岡圭太)

N.M. van der Lugt, et al.

Outcome and management in neonatal thrombocytopenia due to maternal idiopathic thrombocytopenic purpura. Vox Sanq. 105(3): 236-243, 2013

○June 24, 2015

I. Hayasaka (早坂 格)

K. Ishituka, et al.

Medical procedures and outcomes of Japanese patients with trisomy 18 or trisomy 13: Analysis of a nationwide administrative database of hospitalized patients.  
Am J Med. 2015

○July 1, 2015

H. Yamazawa (山澤弘州)

Lacro RV, et al.

Atenolol versus losartan in children and young adults with Marfan's syndrome.

N Engl J Med. 371(22): 2061-2071, 2014

Y. Uzuki (卯月ゆたか)

H Adle-Biassette, et al.

Neuropathological review of 138 cases genetically tested for X-linked hydrocephalus.

Acta Neuropathol. 126(3): 427-442, 2013

○July 15, 2015

D. Sato (佐藤大介)

Santos-Cortez RL, et al.

Rare A2ML1 variants confer susceptibility to otitis media. Nat Genet. 2015

T. Tajima (田島敏広)

A Yamashita, et al.

Statin treatment rescues FGFR3 skeletal dysplasia phenotypes.

Nature. 513(7519): 507-511, 2014

○July 29, 2015

T. Yamazaki (山崎健史)

Tomas NM, et al.

Thrombospondin type-1 domain-containing 7A in idiopathic membranous nephropathy.

N Engl J Med. 371(24): 2277-2287, 2014

A. Shima Said (シェイマーさん)

Ruby F. Fernandez, et al.

Pioglitazone restores phagocyte mitochondrial oxidants and bactericidal capacity in chronic granulomatous disease. J Allergy Clin Immunol. 135(2): 517-527, 2015

○August 26, 2015

T. Akimoto (秋元琢真)

T Ikeda, et al.

Changes in the perfusion waveform of the internal cerebral vein and intraventricular hemorrhage in the acute management of extremely low-birth-weight infants.

Eur J Pediatr. 174(3): 331-338, 2015

M. Ueki (植木将弘)

Basu R, et al.

IL-1 signaling modulates activation of STAT transcription factors to antagonize retinoic acid signaling and control the TH17 cell-iTreg cell balance.

Nature immunology 16(3): 286-295, 2015

○September 2, 2015

T. Ariga (有賀 正)

Lim JS, et al.

Brain somatic mutations in MTOR cause focal cortical dysplasia type II leading to intractable epilepsy. Nat Med. 21(4):395-400, 2015

K. Terada (寺田健作)

Matthias Begemann, et al.

Paternally Inherited IGF2 Mutation and Growth Restriction.

N Engl Med. 373: 349-356, 2015

○September 9, 2015

H. Shiraishi (白石秀明)

Lewis EC, et al.

MR-guided laser interstitial thermal therapy for pediatric drug-resistant lesional epilepsy. Epilepsia. 2015

○September 16, 2015

A. Takeda (武田充人)

Orenstein JM, et al.

Three linked vasculopathic processes characterize Kawasaki disease: a light and transmission electron microscopic study. PLoS One. 7(6), 2012

M. Sugiyama (杉山未奈子)

von Bahr L, et al.

Increased incidence of chronic GvHD and CMV disease in patients with vitamin D deficiency before allogeneic stem cell transplantation.

Bone Marrow Transplant. 50(9):1217-23, 2015

○September 30, 2015

J. Abe (阿部二郎)

Fang EF, et al.

Defective Mitophagy in XPA via PARP-1 Hyperactivation and NAD+/SIRT1 Reduction.

Cell. 157(4):882-896, 2014

○October 7, 2015

Y. Tozawa (戸澤雄介)

Botta Gordon-Smith S, et al.

Correlation of low CD73 expression on synovial lymphocytes with reduced adenosine generation and higher disease severity in juvenile idiopathic arthritis.

Arthritis Rheumatol. 67(2):545-554, 2015

○October 21, 2015

K. Egawa (江川 潔)

Higurashi N, et al.

A human Dravet syndrome model from patient induced pluripotent stem cells.

Mol Brain. 6:19, 2013

Liu Y, et al.

Dravet syndrome patient-derived neurons suggest a novel epilepsy mechanism.

Ann Neurol. 74(1):128-139, 2013

Chamberlain SJ, et al.

Induced pluripotent stem cell models of the genomic imprinting disorders Angelman and Prader-Willi syndromes. Proc Natl Acad Sci U S A. 107(41): 17668-17673, 2010

○October 28, 2015

T. Ito (伊藤智城)

Sasaki M, et al.

Genotype-phenotype correlations in alternating hemiplegia of childhood.

Neurology. 82(6):482-490, 2014

W. Furusawa (古澤 弥)

M Claussnitzer, et al.

FTO Obesity Variant Circuitry and Adipocyte Browning in Humans.

N Engl J Med. 373:895-907, 2015

○November 4, 2015

M. Yamada (山田雅文)

Chen K, et al.

Germline Mutations in NFKB2 Implicate the Noncanonical NF- $\kappa$ B pathway in the Pathogenesis of Common Variable Immunodeficiency.

Am J Hum Genet. 93(5):812-824, 2013

Lee CE, et al.

Autosomal-dominant B-cell deficiency with alopecia due to a mutation in NFKB2 that results in nonprocessable p100. Blood. 124(19):2964-2972, 2014

Vassilios Lougaris, et al.

Defective natural killer-cell cytotoxic activity in NFKB2-mutated CVID-like disease.

J ALLERGY CLIN IMMUNOL. 135:1641-1643, 2015

Y. Sato (佐藤泰征)

Vernon KA, et al.

Partial Complement Factor H Deficiency Associates with C3 Glomerulopathy and Thrombotic Microangiopathy. J Am Soc Nephrol. 2015

○November 11, 2015

S. Morikawa (森川俊太郎)

De Franco E, et al.

The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. Lancet. 386(9997):957-963, 2015

○November 18, 2015

K. Cho (長 和俊)

O'Reilly R, et al.

Diffuse lung disease in infants less than 1 year of age: Histopathological diagnoses and clinical outcome. Pediatr Pulmonol. 50(10):1000-8, 2015

○November 25, 2015

K. Morioka (森岡圭太)

Paul A Fowler, et al.

Maternal Cigarette Smoking and Effects on Androgen Action in Male Offspring:

Unexpected Effects on Second-Trimester.

J Clin Endocrinol Metab. 96(9):1502-1506, 2011

○December 2, 2015

J. Ohshima (大島淳二郎)

Churchman ML, et al.

Efficacy of Retinoids in IKZF1-Mutated BCR-ABL1 Acute Lymphoblastic Leukemia.

Cancer Cell. 28(3):343-56, 2015

I. Hayasaka (早坂 格)

Rauch D, et al.

Persistent Tachypnea of Infancy-Usual and Aberrant. Am J Respir Crit Care Med.2015

○December 9, 2015

H. Yamazawa (山澤弘州)

Opdahl A, et al.

Resting heart rate as predictor for left ventricular dysfunction and heart failure: MESA

(Multi-Ethnic Study of Atherosclerosis). J Am Coll Cardiol. 63(12):1182-9, 2014

Y. Uzuki (卯月ゆたか)

Nestor E Vain, et al.

Effect of gravity on volume of placental transfusion: a multicentre, randomised, non-inferiority trial. Lancet. 384:235-40, 2014

○December 16, 2015

D. Sato (佐藤大介)

Peterson DC, et al.

Outcomes of Medical Emergencies on Commercial Airline Flights.

N Engl J Med. 368(22):2075-83, 2013

○January 20, 2016

Y. Cho (長 祐子)

JN. Stinson, et al.

Construct validity and reliability of a real-time multidimensional smartphone app to assess pain in children and adolescents with cancer. Pain. 156(12):2607-15, 2015

H. Yamamoto (山本啓之)

Videman M, et al.

Effects of prenatal antiepileptic drug exposure on newborn brain activity.

Epilepsia. \*\*(\*)1-11, 2015

○January 27, 2016

K. Ishizu (石津 桂)

Moore WV, et al.

A Randomized Safety and Efficacy Study of Somavaratan (VRS-317), a Long-Acting rhGH, in Pediatric Growth Hormone Deficiency. J Clin Endocrinol Metab . 2015

A. Shima Said (シェイマーさん)

Castiello MC, et al.

B-cell reconstitution after lentiviral vector-mediated gene therapy in patients with Wiskott-Aldrich syndrome. J Allergy Clin Immunol. 136(3):692-702, 2015

○February 3, 2016

T. Akimoto (秋元琢真)

LC Weeke, et al

Lidocaine response rate in aEEG-confirmed neonatal seizures: Retrospective study of 413 full-term and preterm infants. Epilepsia. \*\*(\*)1-10, 2015

M. Ueki (植木将弘)

Castiello MC, et al.

Dominant Mutations in the Autoimmune Regulator AIRE are Associated with Common Organ-specific autoimmune Diseases.

Immunity. 42(6):1185-96, 2015

○February 10, 2016

T. Ariga (有賀 正)

Egan CE, et al

Toll-like receptor 4-mediated lymphocyte influx induces neonatal necrotizing enterocolitis. J Clin Invest. 2015

T. Yamazaki (山崎健史)

Salim S. Hayek, et al.

Soluble Urokinase Receptor and Chronic Kidney Disease.

N Engl J Med. 373:1916-1925, 2015

W. Furusawa (古澤 弥)

Armstrong GT, et al.

Reduction in Late Mortality among 5-Year Survivors of Childhood Cancer. N Engl J Med. 2016

○February 17, 2016

H. Shiraishi (白石秀明)

Kang JY, et al

Laser interstitial thermal therapy for medically intractable mesial temporal lobe epilepsy. Epilepsia. 57(2):325-34, 2016

○February 24, 2016

A. Takeda (武田充人)

Weirather J, et al

Foxp3+ CD4+ T cells improve healing after myocardial infarction by modulating monocyte/macrophage differentiation. Circ Res. 115(1):55-67, 2014

M. Sugiyama (杉山未奈子)

Peyvand F, et al

Caplacizumab for Acquired Thrombotic Thrombocytopenic Purpura.

N Eng J Med. 374(6):511-22, 2016

○March 2, 2016

J. Abe (阿部 二郎)

Philippe Menasche, et al.

Human embryonic stem cell-derived cardiac progenitors for severe heart failure treatment: first clinical case report. Eur Heart J. 36(30):2011-7, 2015

○March 9, 2016

M. Narugami (鳴神雅史)

Lehn A, et al.

Functional neurological disorders: mechanisms and treatment.

J Neurol. 2015 [Epub ahead of print]

○March 16, 2016

K. Egawa (江川 潔)

Whissell PD, et al.

$\gamma$ -aminobutyric acid type A receptors that contain the  $\delta$  subunit promote memory and neurogenesis in the dentate gyrus. Ann Neurol. 74(4):611-21, 2013

Y. Tozawa (戸澤雄介)

Ayse Kilic, et al.

Relationship between clinical findings and genetic mutations in patients with familial Mediterranean fever. Pediatr Rheumatol Online J. 13:59, 2015