

Alexandra Y. Kreins, Sophie Cypowyj, Laurent Abel, Capucine Picard, Stéphanie Boisson-Dupuis, Anne Puel, Jean-Laurent Casanova: 常染色体優性遺伝を呈する慢性皮膚粘膜カンジダ症の責任遺伝子の発見 -STAT1 機能獲得性変異による慢性皮膚粘膜カンジダ症- 第 39 回日本臨床免疫学会総会 2011 年 9 月 15-17 日

Tsumura M, Okada S, Sakai H, Nishikomori R, Yasunaga S, Ohtsubo M, Heike T, Nakahata T, Takihara Y, Kobayashi M: Identification of a novel type of AD-STAT1 deficiency with mutations in the SH2 domain 第 73 回日本血液学会学術集会 2011 年 10 月 14-16 日

2. 論文発表

Karakawa S, Okada S, Tsumura M, Mizoguchi Y, Ohno N, Yasunaga S, Ohtsubo M, Kawai T, Nishikomori R, Sakaguchi T, Takihara Y, Kobayashi M: Decreased Expression in Nuclear Factor- κ B Essential Modulator Due to a Novel Splice-Site Mutation Causes X-linked Ectodermal Dysplasia with Immunodeficiency. J Clin Immunol. 31:762-772,2011

Liù L, Okada S, Kong XF, Kreins AY, Cypowyj S, Abhyankar A, Toubiana J, Itan Y, Audry M, Nitschke P, Masson C, Toth B, Flatot J, Migaud M, Chrabieh M, Kochetkov T, Bolze A, Borghesi A, Toulon A, Hiller J, Eyerich S, Eyerich K, Gulácsy V, Chernyshova L, Chernyshov V, Bondarenko A, Grimaldo RM, Blancas-Galicia L, Beas IM, Roesler J, Magdorf K, Engelhard D, Thumerelle C,

Burgel PR, Hoernes M, Drexel B, Seger R, Kusuma T, Jansson AF, Sawalle-Belohradsky J, Belohradsky B, Jouanguy E, Bustamante J, Bué M, Karin N, Wildbaum G, Bodemer C, Lortholary O, Fischer A, Blanche S, Al-Muhsen S, Reichenbach J, Kobayashi M, Rosales FE, Lozano CT, Kilic SS, Oleastro M, Etzioni A, Traidl-Hoffmann C, Renner ED, Abel L, Picard C, Maródi L, Boisson-Dupuis S, Puel A, Casanova JL: Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. J Exp Med. 208:1635-48, 2011

Hoshina T, Takada H, Sasaki-Mihara Y, Kusuvara K, Ohshima K, Okada S, Kobayashi M, Ohara O, Hara T: Clinical and host genetic characteristics of Mendelian susceptibility to mycobacterial diseases in Japan. J Clin Immunol. 31:309-14,2011.

Nakagawa N, Imai K, Kanegae H, Sato H, Yamada M, Kondoh K, Okada S, Kobayashi M, Agematsu K, Takada H, Mitsuiki N, Oshima K, Ohara O, Suri D, Rawat A, Singh S, Pan-Hammarström Q, Hammarström L, Reichenbach J, Seger R, Ariga T, Hara T, Miyawaki T, Nonoyama S: Quantification of κ -deleting recombination excision circles in Guthrie cards for the identification of early B-cell maturation defects. J Allergy Clin Immunol. 128:223-225.e2,2011

H. 知的財産権の出願・登録状況 特になし

III. 研究成果の刊行に関する一覧表

研究成果の刊行に関する一覧表

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
永井功造、 <u>石井榮一</u>	血球貪食性リンパ組織球症		小児がんハンブック	医薬ジャーナル社		2011	404-413
永井功造、 <u>石井榮一</u>	ウイルス関連血球貪食症候群		小児救急治療ガイドライン 改訂第2版	診断と治療社		2011	356-362
<u>石井榮一</u>	小児の血球貪食性リンパ組織球症(HLH)と Langerhans 細胞組織球症 (LCH)		血液専門医テキスト	南江堂		2011	129-130
<u>安川正貴</u>	造血器腫瘍に対する細胞免疫療法	高久文磨他	Annual Review of Hematology 2012	中外医学社	東京	2012	174-182
<u>安川正貴</u>	免疫療法	金倉 譲	造血器腫瘍学	日本臨床社	東京	2012	230-235

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
<u>Nagai K, Yamamoto K, Ishii E, Yasukawa M, et al.</u>	Subtypes of familial hemophagocytic lymphohistiocytosis in Japan based on genetic and functional analyses of cytotoxic T lymphocytes.	PLoS ONE	5	E14173	2011
<u>Matsuda K, Ishii E, et al.</u>	Detection of T-cell receptor gene rearrangement in children with Epstein-Barr virus-associated hemophagocytic lymphohistiocytosis using the IOMED-2 multiplex polymerase chain reaction combined with GeneScan analysis.	Clin Chim Acta	412	1554-1558	2011
<u>Murata Y, Yasumi T, Ishii E, et al.</u>	Rapid diagnosis of familial hemophagocytic lymphohistiocytosis type 3 (FHL3) by flow cytometric detection of intraplatelet Munc13-4 protein.	Blood	118	1225-1230	2011

Yanagimachi M, <u>Ishii E</u> , et al.	Association of <i>IRF5</i> polymorphisms with susceptibility to hemophagocytic lymphohistiocytosis in children.	J Clin Immunol	31	946-951	2011
Shikata H, <u>Yasukawa M</u> , et al.	The role of activation-induced cytidine deaminase (AID/AICDA) in the progression of follicular lymphoma.	Cancer Sci.	103	415-421	2012
Nagai K, <u>Ishii E</u> , <u>Yasukawa M</u> , et al.	Aurora kinase A-specific T-cell receptor gene transfer redirects T-lymphocytes to display effective anti-leukemia reactivity.	Blood	119	368-376	2012
Hasegawa H, <u>Yasukawa M</u> , et al.	Lysophosphatidylcholine enhances the suppressive function of human naturally occurring regulatory T cells through TGF- β production.	Biochem Biophys Res Commun.	415	526-531	2011
Ochi T, <u>Yasukawa M</u> , et al.	Novel adoptive T-cell immunotherapy using a WT1-specific TCR vector encoding silencers for endogenous TCRs shows marked anti-leukemia reactivity and safety.	Blood	118	1495-1503	2011
Takahara A, <u>Yasukawa M</u> , et al.	Gemcitabine enhances Wilms' tumor gene WT1 expression and sensitizes human pancreatic cancer cells with WT1-specific T-cell-mediated antitumor immune response.	Cancer Immunol. Immunother.	60	1289-1297	2011
Nagai K, <u>Ishii E</u> , <u>Yasukawa M</u> , et al.	Feasibility of gene-immunotherapy using WT1-specific T-cell receptor gene transfer for infant acute lymphoblastic leukemia with MLL gene rearrangement.	Blood Cancer J.	1	e10	2011
<u>Yasukawa M</u> , et al.	Adoptive T-cell immunotherapy using T-cell receptor gene transfer: aiming at a cure for cancer.	Immunotherapy	3	135-140	2011

An J, <u>Yasukawa M</u> , et al.	Activation of T-cell receptor signaling in peripheral T-cell lymphoma cells plays an important role in the development of lymphoma-associated hemophagocytosis.	Int. J. Hemattol.	93	176-185	2011
Ohta H, <u>Yasumi T</u> , et al.	Hematopoietic stem cell transplantation with reduced intensity conditioning from a family haploidentical donor in an infant with familial hemophagocytic lymphohistiocytosis.	Int J Hematol.	94	285-90	2011
Tanaka N, <u>Yasumi T</u> , et al.	High incidence of NLRP3 somatic mosaicism in patients with chronic infantile neurologic, cutaneous, articular syndrome: results of an International Multicenter Collaborative Study.	Arthritis Rheum.	63	3625-32	2011
Murata Y, <u>Yasumi T</u> , Ishii E, et al.	Rapid diagnosis of FHL3 by flow cytometric detection of intraplatelet Munc13-4 protein.	Blood.	118	1225-30	2011
Tahara M, <u>Yasumi T</u> , et al.	Patient with neonatal-onset chronic hepatitis presenting with mevalonate kinase deficiency with a novel MVK gene mutation.	Mod Rheumatol.	21	641-5	2011
Nakabayashi K, <u>Yamamoto K</u> , et al.	Identification of independent risk loci for Graves' disease within the MHC in the Japanese population.	J. Hum. Genet.	56	772-778	2011
Karakawa S, <u>Kobayashi M</u> , et al.	Decreased Expression in Nuclear Factor- κ B Essential Modulator Due to a Novel Splice-Site Mutation Causes X-linked Ectodermal Dysplasia with Immunodeficiency.	J Clin Immunol	31	762-772	2011
Liu L, <u>Kobayashi M</u> , et al.	Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis.	J Exp Med	208	1635-1648	2011
Hoshina T, <u>Kobayashi M</u> , et al.	Clinical and host genetic characteristics of Mendelian susceptibility to mycobacterial diseases in Japan.	J Clin Immunol	31	309-314	2011

Nakagawa N, <u>Kobayashi</u> <u>M</u> , et al.	Quantification of κ-deleting recombination excision circles in Guthrie cards for the identification of early B-cell maturation defects.	J Allergy Clin Immunol	128	223-225	2011
---	---	---------------------------	-----	---------	------

