

Pediatrics 1988; 82: 870-873.

Lawlor F, Peiris S. Harlequin fetus successfully treated with etretinate. *Br J Dermatol* 1985; 112: 585-590.

Natsuga K, Akiyama M, Shimizu H. Malignant skin tumours in inherited ichthyosis patients. *Br J Dermatol* (in press)

Oji V, Tadini G, Akiyama M, *et al.* Revised nomenclature and classification of inherited ichthyoses: Results of the First Ichthyosis Consensus Conference in Sorèze 2009. *J Am Acad Dermatol* 2010; 63: 607-641.

Oji V, Traupe H. Ichthyosis: clinical manifestations and practical treatment options. *Am J Clin Dermatol.* 10: 351-64, 2009.

Prasad RS, Pejaver RK, Hassan A, Al Dusari S, Wooldridge MA. Management and follow-up of harlequin siblings. *Br J Dermatol* 1994; 130: 650-653.

Roberts LJ. Long-term survival of a harlequin fetus. *J Am Acad Dermatol* 1989; 21: 335-339.

Rogers M, Scraf C. Harlequin baby treated with etretinate. *Pediatr Dermatol* 1989; 6: 216-221.

Shwayder T. Disorders of keratinization: diagnosis and management. *Am J Clin Dermatol.* 5: 17-29, 2004.

Shwayder T, Akland T. Neonatal skin barrier: structure, function, and disorders. *Dermatol Ther.* 18: 87-103, 2005.

Ward PS, Jones RD. Successful treatment of a harlequin fetus. *Arch Dis Child* 1989; 64: 1309-1311.

Yanagi T, Akiyama M, Nishihara H, Sakai K, Nishie W, Tanaka S, Shimizu H. Harlequin ichthyosis model mouse reveals alveolar collapse and fetal skin barrier defects. *Hum Mol Genet* 17: 3075-3083, 2008.

Yanagi T, Akiyama M, Sakai K, Nagasaki A, Ozawa N, Kosaki R, Sago H, Shimizu H. DNA-based prenatal exclusion of harlequin ichthyosis. *J Am Acad Dermatol* 58: 653-656, 2008.

Yanagi T, Akiyama M, Nishihara H, Ishikawa J, Sakai K, Miyamura Y, Naoe A, Kitahara T, Tanaka S, Shimizu H. Self-improvement of keratinocyte differentiation defects during skin maturation in ABCA12 deficient harlequin ichthyosis model mice. *Am J Pathol* 177: 106-118, 2010.

Yanagi T, Akiyama M, Nishihara H, Miyamura Y, Sakai K, Tanaka S, Shimizu H. AKT has an anti-apoptotic role in ABCA12-deficient keratinocytes. *J Invest Dermatol* 131: 1942-1945, 2011.

秋山真志. 道化師様魚鱗癬. 玉置邦彦, 他編. 最新皮膚科学大系 第7巻 角化異常性疾患. 中山書店、東京, pp 95-102, 2002.

秋山真志: 魚鱗癬. 今日の治療指針 2012 年版、医学書院、東京, pp 1017-1018, 2011.

秋山真志: 水疱症、角化症 魚鱗癬の病態-最近の知見. 玉置邦彦, 他編. 最新皮膚科学大系 2008-2009、中山書店、東京, pp 189-198, 2008.

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（雑誌）

発表者氏名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Sugiura K, Sugiura N, Yagi T, Iguchi M, Ohno H, Miyazaki Y, Akiyama M	Cryptococcal cellulitis in a patient with bullous pemphigoid	Acta Dermato-Venereol			in press
Shinkuma S, Inoue A, Aoki J, Nishie W, Natsuga K, Ujiie H, Nomura T, Abe R, Akiyama M, Shimizu H	The $\beta 9$ loop domain of PA-PLA $_1\alpha$ has a crucial role in autosomal recessive woolly hair/hypotrichosis	J Invest Dermatol			in press
Sugiura K, Takeichi T, Kono M, Ogawa Y, Shimoyama Y, Muro Y, Akiyama M	A novel IL36RN/IL1F5 homozygous nonsense mutation, p.Arg10X, in a Japanese patient with adult-onset generalized pustular psoriasis	Br J Dermatol			in press
Tanahashi K, Sugiura K, Takeichi T, Takama H, Shinkuma S, Shimizu H, Akiyama M	Prevalent founder mutation c.736T>A of <i>LIPH</i> in autosomal recessive woolly hair of Japanese leads to variable severity of hypotrichosis in adulthood	J Eur Acad Dermatol Venereol			in press
Muro Y, Ishikawa A, Sugiura K, Akiyama M	Clinical features of anti-TIF1- α antibody-positive dermatomyositis patients are closely associated with coexistent dermatomyositis-specific autoantibodies and anti-TIF1- γ or anti-Mi-2 autoantibodies	Rheumatology			in press
Ishikawa A, Sugiura K, Sato A, Muro Y, Akiyama M	Drug eruption due to sodium picosulfate	Eur J Dermatol			in press
Hane H, Yokota K, Kono M, Muro Y, Akiyama M	Extraordinarily large, giant spider angioma in an alcoholic cirrhotic	Int J Dermatol			in press

	patient				
Ishikawa A, Muro Y, Sugiura K, Akiyama M	Development of an ELISA for detection of autoantibodies to nuclear matrix protein 2	Rheumatology			in press
Sugiura K, Muro Y, Akiyama M	Autoantibodies to nuclear matrix protein 2 /MJ in adult-onset dermatomyositis with severe calcinosis	J Am Acad Dermatol			in press
Yasuda K, Sugiura K, Ishikawa R, Kihira M, Negishi Y, Iwayama H, Ito K, Kimura H, Kosugi I, Akiyama M	Perinatal cytomegalovirus-associated bullae in an immunocompetent infant	Arch Dermatol			in press
Muro Y, Sugiura K, Hoshino K, Akiyama M	Disappearance of anti-MDA-5 autoantibodies in clinically amyopathic dermatomyositis/interstitial lung disease during disease remission	Rheumatology			in press
Kono M, Akiyama M, Sukanuma M, Sanchez-Valle A, Tomita Y	Dyschromatosis symmetrica hereditaria by ADAR1 mutations and viral encephalitis: a hidden link?	Int J Dermatol			in press
Mizuno O, Yanagi T, Baba K, Yamane N, Inokuma D, Ito K, Akiyama M, Shimizu H	Sweet's syndrome presenting with vegetative nodules on the hands: relationship to neutrophilic dermatosis of the dorsal hands	Int J Dermatol			in press
Kono M, Akiyama M, Kondo T, Suzuki T, Sukanuma M, Wataya-Kaneda M, Lam J, Shibaki A, Tomita Y	Four novel <i>ADAR1</i> gene mutations in patients with dyschromatosis symmetrica hereditaria	J Dermatol			in press
Karasawa T, Matsumoto T, Akiyama M	Metastatic skin lesions of multiple myeloma presenting as two extraordinarily large	Int J Dermatol			in press

	subcutaneous tumors				
Izumi K, Yanagi T, Akiyama M, Moriuchi R, Arita K, Shimizu H	Intractable erythematous plaques on the hands: palmoplantar eosinophilic pustular folliculitis	Int J Dermatol			in press
Mori M, Sugiura M, Kono M, Matsumoto T, Sawada M, Yokota K, Yasue S, Shibata S, Sakakibara A, Nakamura S, Tomita Y, Akiyama M	Clinico-pathologic Analysis of 66 Japanese Thin Melanomas with Metastasis of Sentinel or Regional Lymph Node	J Cutan Pathol			in press
Saito N, Abe R, Yoshioka N, Murata J, Fujita Y, Shimizu H	Prolonged elevation of serum granulysin in drug-induced hypersensitivity syndrome	Br J Dermatol			in press
Hirata Y, Abe R, Kikuchi K, Hamasaka A, Shinkuma S, Nomura T, Nishie W, Arita K, Shimizu H	Intraepidermal neutrophilic IgA pemphigus successfully treated with dapsone	Eur J Dermatol			in press
Fujita Y, Inokuma D, Abe R, Sasaki M, Nakamura H, Shimizu T, Shimizu H	Conversion from human haematopoietic stem cells to keratinocytes requires keratinocyte secretory factors	Clin Exp Dermatol			in press
Yajima I, Kumasaka YM, Naito Y, Yoshikawa T, Takahashi H, Funasaka Y, Suzuki T, Kato M	Reduced GNG2 expression levels in mouse malignant melanomas and human melanoma cell lines	Am J Can Res			in press
Ezzedine K, Lim HW, Suzuki T, Katayama I, Hamzavi I, Lan CCE, Goh BK, Anbar T, Silva de Castro C, Lee AY, Parsad D, Geel van	Revised classification/nomenclature of vitiligo and related issues: The Vitiligo Global Issues Consensus Conference	Pigment Cell Melanoma Res			in press

N, Poole le IC, Oiso N, Benzekri L, Spritz R, Hann SK, Picardo M, Taieb A					
Bilen N, Aktürk AS, Kawaguchi M, Salman S, Erçin C, Hozumi Y, Suzuki T	Dyschromatosis symmetrica hereditaria: a case report from Turkey, a new association and a novel gene mutation	J Dermatol			in press
Kawaguchi M, Hayashi M, Murata I, Hozumi Y, Suzuki N, Ishii Y, Wataya-Kaneda M, Funasaka Y, Kawakami T, Fukai K, Ochiai T, Nishigori C, Mitsuhashi Y, Suzuki T	Eleven novel mutations of the <i>ADARI</i> gene in dyschromatosis symmetrica hereditaria	J Dermatol Sci			in press
Oiso N, Kimura M, Tanemura A, Tsuruta D, Ito T, Suzuki T, Katayama I, Kawada A	Blaschkitis-like eruptions with hypodontia and low IκB kinase gamma expression	J Dermatol			in press
Shimanuki M, Takeda K, Kawaguchi M, Suzuki T, Shibahara S	Lipocalin-type prostaglandin D synthase as a marker for the growth potential of melanocyte-lineage cells in the human skin	J Dermatol			in press
Kono M, Kondo T, Ito S, Suzuki T, Wakamatsu K, Ito S, Tomita Y	Oculocutaneous albinism 1 minimal pigment type: A case report on the analysis of genotype of an OCA1MP patient	Br J Dermatol			in press
Hayashi M, Yamada M, Hiroshima Y, Suzuki T	A case of multiple myeloma presenting as a subcutaneous nodule on the abdomen	J Dermatol			in press
Matsumoto Y, Mitsuhashi Y, Monma F, Suzuki T, Kawaguchi M, Igarashi A, Miyabe	Nephrogenic Systemic Fibrosis; A case report and review on Japanese patients	J Dermatol			in press

C, Tsuboi R					
Natsuga K, Shinkuma S, Kanda M, Suzuki Y, Chosa N, Narita Y, Setoyama M, Nishie W, Akiyama M, Shimizu H	Possible modifier effects of keratin 17 gene mutation on keratitis-ichthyosis-deafness syndrome	Br J Dermatol	166 (4)	903-905	2012
Sasaki K, Akiyama M, Yanagi T, Sakai K, Miyamura Y, Sato M, Shimizu H	CYP4F22 is highly expressed at the site and onset of keratinization during human skin development	J Dermatol Sci	65 (2)	156-158	2012
Fukuda S, Hamada T, Ishii N, Sakaguchi S, Sakai K, Akiyama M, Shimizu H, Masuda K, Izu K, Teye K, Tsuruta D, Karashima T, Nakama T, Yasumoto S, Hashimoto T	Novel adenosine triphosphate (ATP)-binding cassette, subfamily A, member 12 (ABCA12) mutations associated with congenital ichthyosiform erythroderma	Br J Dermatol	166	218-221	2012
Hasegawa M, Asano Y, Endo H, Fujimoto M, Goto D, Ihn H, Inoue K, Ishikawa O, Kawaguchi Y, Kuwana M, Muro Y, Ogawa F, Sasaki T, Takahashi H, Tanaka S, Takehara K, Sato S	Investigation of prognostic factors for skin sclerosis and lung function in Japanese patients with early systemic sclerosis: a multicentre prospective observational study	Rheumatology (Oxford)	51	129-133	2012
Hayashi I, Abe R, Yanagi T, Abe Y, Shimizu H	Yellow nail syndrome: Nail change reflects disease severity	J Dermatol	39	415-416	2012
Hayashi M, Nakano H, Sawamura D, Suzuki T	Case of epidermolytic palmoplantar keratoderma with knuckle pads	J Dermatol	39	84-87	2012
Muro Y, Sugiura K, Hoshino K, Akiyama M, Tamakoshi K	Epidemiologic study of clinically amyopathic dermatomyositis and anti-melanoma differentiation-associate	Arthritis Res Ther	13	R214	2011

	d gene 5 antibodies in central Japan				
Natsuga K, Akiyama M, Shimizu H	Malignant skin tumours in patients with inherited ichthyosis	Br J Dermatol	165	263-268	2011
Akiyama M	Updated molecular genetics and pathogenesis of ichthyoses	Nagoya J Med Sci	73	79-90	2011
Akiyama M	The roles of ABCA12 in keratinocyte differentiation and lipid barrier formation in the epidermis	Dermato-Endocrinology	3	107-112	2011
Osawa R, Akiyama M, Shimizu H	Filaggrin gene defects and the risk of developing allergic disorders	Allergol Int	60	1-9	2011
Kaibuchi-Noda K, Yokota K, Matsumoto T, Sawada M, Sakakibara A, Kono M, Tomita Y, Watanabe D, Fukumoto H, Katano H, Akiyama M	Detection of Merkel cell polyomavirus in cutaneous squamous cell carcinoma prior to occurrence of Merkel cell carcinoma	J Am Acad Dermatol	65	e152-4	2011
Li Q, Frank M, Akiyama M, Shimizu H, Ho S-Y, Thisse C, Thisse B, Sprecher E, Uitto J	Abca12-mediated lipid transport and snap29-dependent trafficking of lamellar granules are critical for epidermal morphogenesis in Zebrafish Disease Model of Ichthyosis	Dis Model Mech	4	777-785	2011
Takeichi T, Sugiura K, Muro Y, Ogawa Y, Akiyama M	LEDGF/DFS70 activates the MK2/IL6/STAT3 pathway in HaCaT	J Dermatol Sci	63	203-205	2011
Suga H, Tsunemi Y, Sugaya M, Shinkuma S, Akiyama M, Shimizu H, Sato S	Hair shaft abnormalities in localized autosomal recessive hypotrichosis 2 and a review of other non-syndromic human alopecias	Acta Dermato-Venereol	91	486-488	2011

Furue M, Yamazaki S, Jimbow K, Tsuchida T, Amagai M, Tanaka T, Matsunaga K, Muto M, Morita E, Akiyama M, Soma Y, Terui T, Manabe M	Prevalence of dermatological disorders in Japan: A nationwide, cross-sectional, seasonal, multicenter, hospital-based study	J Dermatol	38	353-363	2011
Nakajima K, Uchida Y, Akiyama M, Morita Y, Shimizu H, Sano S	Altered lipid profiles in the stratum corneum of Sjögren-Larsson syndrome	J Dermatol Sci	63	64-66	2011
Natsuga K, Nishie W, Smith BJ, Shinkuma S, Smith TA, Parry DAD, Oiso N, Kawada A, Yoneda K, Akiyama M, Shimizu H	Consequences of two different amino acid substitutions at the same codon in <i>KRT14</i> indicate definitive roles of structural distortion in epidermolysis bullosa simplex pathogenesis	J Invest Dermatol	131	1869-1876	2011
Yanagi T, Akiyama M, Nishihara H, Miyamura Y, Sakai K, Tanaka S, Shimizu H	AKT has an anti-apoptotic role in ABCA12-deficient keratinocytes	J Invest Dermatol	131	1942-1945	2011
Kikuchi K, Arita K, Tateishi Y, Onozawa M, Akiyama M, Shimizu H	Recurrence of hydroxyurea-induced leg ulcer after discontinuation of treatment	Acta Dermato-Venereol	91	373-374	2011
Nakamura H, Natsuga K, Nishie W, McMillan JR, Nakamura H, Sawamura D, Akiyama M, Shimizu H	DNA-based prenatal diagnosis of plectin-deficient epidermolysis bullosa simplex associated with pyloric atresia	Int J Dermatol	50	439-442	2011
Umemoto H, Akiyama M, Yanagi T, Sakai K, Aoyama Y, Oizumi A, Suga Y, Kitagawa Y, Shimizu H	New insight into genotype/phenotype correlations in ABCA12 mutations in harlequin ichthyosis	J Dermatol Sci	61	136-138	2011
Natsuga K, Nishie W, Shinkuma S, Nakamura H, Arita	A founder effect of c.1938delC in <i>ITGB4</i> underlies junctional	Exp Dermatol	20	74-76	2011

K, Yoneda K, Kusaka T, Yanagihara T, Kosaki R, Sago H, Akiyama M, Shimizu H	epidermolysis bullosa and its application for prenatal testing				
Kusajima E, Akiyama M, Sato M, Natsuga K, Shimizu H	Type XVII collagen ELISA indices significantly decreased after bullous pemphigoid remission	Int J Dermatol	50	238-240	2011
Osawa R, Akiyama M, Izumi K, Ujiie H, Sakai K, Nemoto-Hasebe I, Yanagi T, Koizumi H, Shimizu H	Extremely severe palmoplantar hyperkeratosis in a generalized epidermolytic hyperkeratosis patient with a keratin 1 gene mutation	J Am Acad Dermatol	64	991-993	2011
Lin H-Y, Yanagi T, Akiyama M, Iitani MM, Moriuchi R, Natsuga K, Shinkuma S, Yamane N, Inokuma D, Arita K, Shimizu H	Childhood subepidermal blistering disease with autoantibodies against type VII collagen and laminin-332	Br J Dermatol	164	452-454	2011
Hamaguchi Y, Kuwana M, Hoshino K, Hasegawa M, Kaji K, Matsushita T, Komura K, Nakamura M, Kodera M, Suga N, Higashi A, Ogusu K, Tsutsui K, Furusaki A, Tanabe H, Sasaoka S, Muro Y, Yoshikawa M, Ishiguro N, Ayano M, Muroi E, Fujikawa K, Umeda Y, Kawase M, Mabuchi E, Asano Y, Sodemoto K, Seishima M,	Clinical correlations with dermatomyositis-specifi c autoantibodies in adult Japanese patients with dermatomyositis: a multicenter cross-sectional study	Arch Dermatol	147	391-398	2011

Yamada H, Sato S, Takehara K, Fujimoto M					
Watanabe K, Muro Y, Sugiura K, Tomita Y	IgE and IgG ₄ autoantibodies against DFS70/LEDGF in atopic dermatitis	Autoimmunity	44	511-519	2011
Hibino M, Sugiura K, Muro Y, Shimoyama Y, Tomita Y	Cyclosporin A induces the unfolded protein response in keratinocytes	Arch Dermatol Res	303	481-489	2011
Kikuchi K, Abe R, Shinkuma S, Hamasaka E, Natsuga K, Hata H, Tateishi Y, Shibata M, Tomita Y, Abe Y, Aoyagi S, Mukai M, Shimizu H	Spontaneous remission of solitary-type infantile myofibromatosis	Case Rep Dermatol	3	181-185	2011
Fujita Y, Yoshioka N, Abe R, Murata J, Hoshina D, Mae H, Shimizu H	Rapid immunochromatographi c test for serum granulysin is useful for the prediction of Stevens-Johnson syndrome and toxic epidermal necrolysis	J Am Acad Dermatol	65	65-68	2011
Yoshihisa Y, Makino T, Matsunaga K, Honda A, Norisugi O, Abe R, Shimizu H, Shimizu T	Macrophage migration inhibitory factor is essential for eosinophil recruitment in allergen-induced skin inflammation	J Invest Dermatol	131	925-931	2011
Narita T, Oiso N, Fukai K, Kabashima K, Kawada A, Suzuki T	Generalized vitiligo and associated autoimmune diseases in Japanese patients and their families	Allergol Int	60	505-508	2011
Arase A, Wataya-Kaneda M, Oiso N, Tanemura A, Kawada A, Suzuki T, Katayama I	Repigmentation of leukoderma in a piebald patient associated with a novel c-KIT gene mutation, G592E, of the tyrosine kinase domain	J Dermatol Sci	58	147-149	2011
Murata T, Yagi Y, Tanioka M, Suzuki T, Miyachi Y,	Dyschromatosis symmetrica hereditaria with acral hypertrophy	Euro J Dermatol	21	649-650	2011

Morita K, Utani A					
Yamada M, Hayashi M, Sakai K, Hozumi Y, Abe Y, Kawaguchi M, Ihn H, Suzuki T	Oculocutaneous Albinism Type 3: a Japanese Girl With Novel Mutations in <i>TYRP1</i> gene	J Dermatol Sci	64	217-222	2011
Hayashi M, Suzuki T	A case of subcutaneous lobular capillary hemangioma	J Dermatol	38	1003-1006	2011
Narita T, Oiso N, Fukai K, Motokawa T, Hayashi M, Yokoyama K, Hozumi Y, Kawada A, Suzuki T	Two children with a mild or moderate piebaldism phenotype and a father with no leukoderma in a family with the same recurrent missense mutation in the kinase domain of <i>KIT</i>	Euro J Dermatol	21	446-447	2011
Hayashi M, Kawaguchi M, Hozumi Y, Nakano H, Sawamura D, Suzuki T	Dystrophic epidermolysis bullosa pruriginosa of elderly onset	J Dermatol	38	173-178	2011
Oiso N, Murata I, Hayashi M, Amatsu A, Yoshida M, Suzuki T, Kawada A	Dermoscopic features of dyschromatosis symmetrica hereditaria	J Dermatol	38	91-93	2011
Oiso N, Suzuki T, Fukai K, Katayama I, Kawada A	Nonsegmental Vitiligo and Autoimmune Mechanism.	Dermatology Research and Practice	—	Article ID 518090, doi:10.1155/2011/518090	2011
Akiyama M	<i>ABCA12</i> mutations and autosomal recessive congenital ichthyosis: A review of genotype/phenotype correlations and of pathogenetic concepts	Hum Mutation	31	1090-1096	2010
Akiyama M, Sakai K, Yanagi T, Fukushima S, Ihn H, Hitomi K, Shimizu H	Transglutaminase 1 preferred substrate peptide K5 is an efficient tool in diagnosis of lamellar ichthyosis	Am J Pathol	176	1592-1599	2010

Saito N, Yanagi T, Akiyama M, Lin K, Kasai S, Fujita Y, Yamane N, Inokuma D, Kase S, Ono K, Minakawa H, Shimizu H	Pyoderma gangrenosum of the eyelid: report of two cases and review of the literature	Dermatol	221	211-215	2010
Natsuga K, Nishie W, Shinkuma S, Nakamura H, Arita K, Yoneda K, Kusaka T, Yanagihara T, Kosaki R, Sago H, Akiyama M, Shimizu H	A founder effect of c.1938delC in <i>ITGB4</i> underlies junctional epidermolysis bullosa and its application for prenatal testing	Exp Dermatol	20	74-76	2010
Sakai K, Akiyama M, Yanagi T, Nampoothiri S, Mampilly T, V S, Shimizu H	An Indian family with Sjögren-Larsson syndrome caused by a novel <i>ALDH3A2</i> mutation	Int J Dermatol	49	1031-1033	2010
Oji V, Tadani G, Akiyama M, Blanchet-Bardon C, Bodemer C, Bourrat E, Coudiere P, DiGiovanna JJ, Elias P, Fischer J, Fleckmann P, Gina M, Harper J, Hashimoto T, Hausser I, Hennies HC, Hohl D, Hovnanian A, Ishida-Yamamoto A, Jacyk WK, Leachman S, Leigh I, Mazereeuw-Hautier J, Milstone L, Morice-Picard F, Paller AS, Richard G, Schmuth M, Shimizu H, Sprecher E, van Steensel M, Taieb A, Toro JR, Vabres	Revised nomenclature and classification of inherited ichthyoses: Results of the First Ichthyosis Consensus Conference in Sorèze 2009	J Am Acad Dermatol	63	607-641	2010

P, Vahlquist A, Williams M, Traupe H					
Akiyama M, Sakai K, Yanagi T, Tabata N, Yamada M, Shimizu H	Partially disturbed lamellar granule secretion in mild congenital ichthyosiform erythroderma with <i>ALOX12B</i> mutations	Br J Dermatol	163	201-204	2010
Shinkuma S, Akiyama M, Inoue A, Aoki J, Natsuga K, Nomura T, Arita K, Abe R, Ito K, Nakamura H, Ujiie H, Shibaki A, Suga H, Tsunemi Y, Nishie W, Shimizu H	Prevalent <i>LIPH</i> founder mutations lead to loss of P2Y5 activation ability of PA-PLA ₁ α in autosomal recessive hypotrichosis	Hum Mutation	31	602-610	2010
Yanagi T, Akiyama M, Nishihara H, Ishikawa J, Sakai K, Miyamura Y, Naoe A, Kitahara T, Tanaka S, Shimizu H	Self-improvement of keratinocyte differentiation defects during skin maturation in ABCA12 deficient harlequin ichthyosis model mice	Am J Pathol	177	106-118	2010
Uchida Y, Cho Y, Moradian S, Kim J, Nakajima K, Crumrine D, Park K, Ujihara M, Akiyama M, Shimizu H, Holleran WM, Sano S, Elias PM	Neutral lipid storage leads to acylceramide deficiency, likely contributing to the pathogenesis of Dorfman-Chanarin syndrome	J Invest Dermatol	130	2497-2499	2010
Natsuga K, Nishie W, Arita K, Shinkuma S, Nakamura H, Kubota S, Imakado S, Akiyama M, Shimizu H	Complete paternal isodisomy of chromosome 17 in junctional epidermolysis bullosa with pyloric atresia	J Invest Dermatol	130	2671-2674	2010
Osawa R, Konno S, Akiyama M, Nemoto-Hasebe I, Nomura T, Nomura	Japanese-specific filaggrin gene mutations in Japanese patients suffering from atopic	J Invest Dermatol	130	2834-2836	2010

Y, Abe R, Sandilands A, McLean WHI, Hizawa N, Nishimura M, Shimizu H	eczema and asthma				
Nomura Y, Akiyama M, Nomura T, Nemoto-Hasebe I, Abe R, McLean WHI, Shimizu H	Chromosome 11q13.5 variant: No association with atopic eczema in the Japanese population	J Dermatol Sci	59	210-212	2010
Fujita Y, Abe R, Inokuma D, Sasaki M, Hoshina D, Natsuga K, Nishie W, McMillan JA, Nakamura H, Shimizu T, Akiyama M, Sawamura D, Shimizu H	Bone marrow transplantation restores epidermal basement membrane protein expression and rescues epidermolysis bullosa model mice	Proc Natl Acad Sci USA	107	14345- 14350	2010
Yamane A, Fukui M, Sugimura Y, Itoh M, Perez Alea M, Thomas V, El Alaoui S, Akiyama M, Hitomi K	Identification of a preferred substrate peptide for transglutaminase 3 and detection of in situ activity in skin and hair follicles	FEBS J	277	3564-3574	2010
Mitsutake S, Suzuki C, Akiyama M, Tsuji K, Yanagi T, Shimizu H, Igarashi Y	ABCA12 dysfunction causes a disorder in glucosylceramide accumulation during keratinocyte differentiation	J Dermatol Sci	60	128-129	2010
Hayashi M, Saito H, Monma F, Katagiri Y, Kawaguchi M, Mitsubishi Y, Abe Y, Suzuki T	Linear IgA bullous dermatosis associated with herpes simplex virus infection and Kawasaki disease	J Dermatol	37	276-279	2010
Hayashi M, Suzuki T	A missense mutation c.G2747A (p.R916Q) of ADAR1 gene in dyschromatosis symmetrica hereditaria is not a novel mutation	Arch Dermatol Res	302	481-482	2010

Murata I, Hayashi M, Hozumi Y, Fujii K, Mitsuhashi Yi, Oiso N, Fukai K, Kuroki N, Mori Y, Utani A, Tomita Y, Fujita Y, Suzuki T	Mutation analysis of patients with dyschromatosis symmetrica hereditaria: five novel mutations of the <i>ADAR1</i> gene	J Dermatol Sci	58	218-220	2010
Matsui Y, Sugiyama K, Kamei M, Takahashi T, Suzuki T, Katagata Y, Ito T	Extract of passion fruit (<i>Passiflora edulis</i>) seed containing high amounts of piceatannol inhibits melanogenesis and promotes collagen synthesis	J Agric Food Chem	58	11112-11118	2010
Yoshihisa Y, Honda A, Zhao QL, Makino T, Abe R, Matsui K, Shimizu H, Miyamoto Y, Kondo T, Shimizu T	Protective effects of platinum nanoparticles against UV-light-induced epidermal inflammation	Exp Dermatol	19 (11)	1000-1006	2010
Iitani MM, Abe R, Yanagi T, Hamasaka A, Tateishi Y, Abe Y, Ito M, Kondo T, Kubota K, Shimizu H	Aleukemic leukemia cutis with extensive bone involvement	J Am Acad Dermatol	63(3)	539-541	2010
Hamasaka A, Yoshioka N, Abe R, Kishino S, Umezawa K, Ozaki M, Todo S, Shimizu H	Topical application of dehydroxymethylepoxy quinomicin improves allergic inflammation via NF-kappaB inhibition	J Allergy Clin Immunol	126 (2)	400-403	2010
Natsuga K, Nishie W, Shinkuma S, Arita K, Nakamura H, Ohyama M, Osaka H, Kambara T, Hirako Y, Shimizu H	Plectin deficiency leads to both muscular dystrophy and pyloric atresia in epidermolysis bullosa simplex	Hum Mutat	31 (10)	E1687-1698	2010
Li Q, Ujiie H, Shibaki A, Wang G, Moriuchi R, Qiao HJ, Morioka H, Shinkuma S, Natsuga K, Long	Human IgG1 monoclonal antibody against human collagen 17 noncollagenous 16A domain induces blisters via complement	J Immunol	185 (12)	7746-7755	2010

HA, Nishie W, Shimizu H	activation in experimental bullous pemphigoid model				
Inokuma D, Shibaki A, Shimizu H	Two cases of cutaneous sporotrichosis in continental/microtherm al climate zone: global warming alert?	Clin Exp Dermatol	35(6)	668-669	2010

IV. 研究成果の刊行物・別刷

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A novel *IL36RN/IL1F5* homozygous nonsense mutation, p.Arg10X, in a Japanese patient with adult-onset generalized pustular psoriasis

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Abbreviations: C-reactive protein (CRP), generalized pustular psoriasis (GPP), interleukin 36 receptor antagonist (*IL36RN*), palmoplantar pustulosis (PPP), psoriasis vulgaris (PV)

Keywords: adult onset, generalized pustular psoriasis, *IL36RN*, immunohistochemistry, nonsense mutation

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