

# Medical articles

1. Is adermatoglyphia an additional feature of Kindler Syndrome?

Almeida HL Jr, Goetze FM, Fong K, Lai-Cheong J, McGrath J.

An Bras Dermatol. 2015 Jul-Aug;90(4):592-3. doi: 10.1590/abd1806-4841.20153501.

PMID: 26375235

2. Interstitial granulomatous drug reaction induced by quetiapine.

Tan ES, Robson A, Lai-Cheong JE, Wain EM.

Clin Exp Dermatol. 2016 Mar;41(2):210-1. doi: 10.1111/ced.12662.

PMID: 25960075

3. Somatic forward (nonrevertant) mosaicism in recessive dystrophic epidermolysis bullosa.

Shipman AR, Liu L, Lai-Cheong JE, McGrath JA, Heagerty A.

JAMA Dermatol. 2014 Sep;150(9):1025-7. doi: 10.1001/jamadermatol.2014.281.

PMID: 24989707

4. Kindlin-1 controls Wnt and TGF- $\beta$  availability to regulate cutaneous stem cell proliferation.

Rognoni E, Widmaier M, Jakobson M, Ruppert R, Ussar S, Katsougkri D, Böttcher RT, Lai-Cheong JE, Rifkin DB, McGrath JA, Fässler R.

Nat Med. 2014 Apr;20(4):350-9. doi: 10.1038/nm.3490.

PMID: 24681597

5. Sporadic Kindler syndrome with a novel mutation.

Almeida HL Jr, Heckler GT, Fong K, Lai-Cheong J, McGrath J.

An Bras Dermatol. 2013 Nov-Dec;88(6 Suppl 1):212-5. doi: 10.1590/abd1806-4841.20132173.

PMID: 24346923

6. Chemical atopy.

Puangpet P, Lai-Cheong J, McFadden JP.

Contact Dermatitis. 2013 Apr;68(4):208-13. doi: 10.1111/cod.12029.

PMID: 23510341

7. Revertant mosaicism in the skin.

Lai-Cheong JE, McGrath JA.

G Ital Dermatol Venereol. 2013 Feb;148(1):73-82.

PMID: 23407079

8. Pathogenesis-based therapies in ichthyoses.

Lai-Cheong JE, Elias PM, Paller AS.

Dermatol Ther. 2013 Jan-Feb;26(1):46-54. doi: 10.1111/j.1529-8019.2012.01528.x.

PMID: 23384020

9.Recurrent heterozygous missense mutation, p.Gly573Ser, in the TRPV3 gene in an Indian boy with sporadic Olmsted syndrome.

Lai-Cheong JE, Sethuraman G, Ramam M, Stone K, Simpson MA, McGrath JA.  
Br J Dermatol. 2012 Aug;167(2):440-2. doi: 10.1111/j.1365-2133.2012.11115.x.  
PMID: 22835024

10.MBTPS2 mutation in a British pedigree with keratosis follicularis spinulosa decalvans.

Fong K, Wedgeworth EK, Lai-Cheong JE, Tosi I, Mellerio JE, Powell AM, McGrath JA.  
Clin Exp Dermatol. 2012 Aug;37(6):631-4. doi: 10.1111/j.1365-2230.2011.04288.x.  
PMID: 22816986

11.Case of Kindler syndrome resulting from mutation in the FERMT1 gene.

Wada M, Masuda K, Tsuruta D, Tamai K, Lai-Cheong JE, McGrath JA, Katoh N.  
J Dermatol. 2012 Dec;39(12):1057-8. doi: 10.1111/j.1346-8138.2012.01598.x.  
PMID: 22672060

12.Germline mutation in ATR in autosomal- dominant oropharyngeal cancer syndrome.

Tanaka A, Weinel S, Nagy N, O'Driscoll M, Lai-Cheong JE, Kulp-Shorten CL, Knable A, Carpenter G, Fisher SA, Hiragun M, Yanase Y, Hide M, Callen J, McGrath JA.  
Am J Hum Genet. 2012 Mar 9;90(3):511-7. doi: 10.1016/j.ajhg.2012.01.007.  
PMID: 22341969

13.Mutations in AEC syndrome skin reveal a role for p63 in basement membrane adhesion, skin barrier integrity and hair follicle biology.

Clements SE, Techanukul T, Lai-Cheong JE, Mee JB, South AP, Pourreyaon C, Burrows NP, Mellerio JE, McGrath JA.  
Br J Dermatol. 2012 Jul;167(1):134-44. doi: 10.1111/j.1365-2133.2012.10888.x.  
PMID: 22329826

14.Infantile systemic hyalinosis associated with a putative splice-site mutation in the ANTXR2 gene.

Fong K, Rama Devi AR, Lai-Cheong JE, Chirla D, Panda SK, Liu L, Tosi I, McGrath JA.  
Clin Exp Dermatol. 2012 Aug;37(6):635-8. doi: 10.1111/j.1365-2230.2011.04287.x.  
PMID: 22300424

15.Angioid streaks with severe macular dysfunction and generalised retinal involvement due to a homozygous duplication in the ABCC6 gene.

Tan MH, Vanakker OM, Tran HV, Robson AG, Lai-Cheong JE, Groves R, Holder GE, Moore AT.  
Eye (Lond). 2012 May;26(5):753-5. doi: 10.1038/eye.2011.344.  
PMID: 22261738

16.Autosomal recessive epidermolysis bullosa simplex due to loss of BPAG1-e expression.

Liu L, Dopping-Hepenstal PJ, Lovell PA, Michael M, Horn H, Fong K, Lai-Cheong JE, Mellerio JE, Parsons M, McGrath JA.  
J Invest Dermatol. 2012 Mar;132(3 Pt 1):742-4. doi: 10.1038/jid.2011.379.  
PMID: 22113475

17.Revertant mosaicism in Kindler syndrome.

Lai-Cheong JE, Moss C, Parsons M, Almaani N, McGrath JA.

J Invest Dermatol. 2012 Mar;132(3 Pt 1):730-2. doi: 10.1038/jid.2011.352.

PMID: 22089829

18.Next-generation diagnostics for inherited skin disorders.

Lai-Cheong JE, McGrath JA.

J Invest Dermatol. 2011 Oct;131(10):1971-3. doi: 10.1038/jid.2011.253.

PMID: 21918571

19.Schöpf-Schulz-Passarge syndrome resulting from a homozygous nonsense mutation, p.Cys107X, in WNT10A.

Petrof G, Fong K, Lai-Cheong JE, Cockayne SE, McGrath JA.

Australas J Dermatol. 2011 Aug;52(3):224-6. doi: 10.1111/j.1440-0960.2011.00788.x.

PMID: 21834823

20.What is Kindler syndrome?

Lai-Cheong JE, McGrath JA.

Skinmed. 2011 May-Jun;9(3):145-6.

PMID: 21675491

21. New homozygous SPINK5 mutation, p.Gln333X, in a Turkish pedigree with Netherton syndrome.

Fong K, Akdeniz S, Isi H, Taskesen M, McGrath JA, Lai-Cheong JE.

Clin Exp Dermatol. 2011 Jun;36(4):412-5. doi: 10.1111/j.1365-2230.2010.03976.x. Erratum in: Clin Exp Dermatol. 2011 Oct;36(7):831.

PMID: 21564178

22.HB-EGF induces COL7A1 expression in keratinocytes and fibroblasts: possible mechanism underlying allogeneic fibroblast therapy in recessive dystrophic epidermolysis Bullosa.

Nagy N, Almaani N, Tanaka A, Lai-Cheong JE, Techanukul T, Mellerio JE, McGrath JA.

J Invest Dermatol. 2011 Aug;131(8):1771-4. doi: 10.1038/jid.2011.85. No abstract available.

PMID: 21471992

23.Identical glycine substitution mutations in type VII collagen may underlie both dominant and recessive forms of dystrophic epidermolysis bullosa.

Almaani N, Liu L, Dopping-Hepenstal PJ, Lai-Cheong JE, Wong A, Nanda A, Moss C, Martínéz AE, Mellerio JE, McGrath JA.

Acta Derm Venereol. 2011 May;91(3):262-6. doi: 10.2340/00015555-1053.

PMID: 21448560

24.cis-Urocanic acid enhances prostaglandin E2 release and apoptotic cell death via reactive oxygen species in human keratinocytes.

Kaneko K, Walker SL, Lai-Cheong J, Matsui MS, Norval M, Young AR.

J Invest Dermatol. 2011 Jun;131(6):1262-71. doi: 10.1038/jid.2011.37.

PMID: 21412256

25. Successful treatment of microstomia with UVA1 phototherapy in systemic sclerosis.  
Tewari A, Garibaldinos T, Lai-Cheong J, Groves R, Sarkany R, Branislav Novakovic L.  
*Photodermatol Photoimmunol Photomed*. 2011 Apr;27(2):113-4. doi: 10.1111/j.1600-0781.2011.00570.x.  
PMID: 21392116
26. Novel and recurrent FERMT1 gene mutations in Kindler syndrome.  
Techanukul T, Sethuraman G, Zlotogorski A, Horev L, Macarov M, Trainer A, Fong K, Lens M, Medenica L, Ramesh V, McGrath JA, Lai-Cheong JE.  
*Acta Derm Venereol*. 2011 May;91(3):267-70. doi: 10.2340/00015555-1063.  
PMID: 21336475
27. Revertant mosaicism in skin: natural gene therapy.  
Lai-Cheong JE, McGrath JA, Uitto J.  
*Trends Mol Med*. 2011 Mar;17(3):140-8. doi: 10.1016/j.molmed.2010.11.003.  
PMID: 21195026
28. Images in clinical medicine. Tinea.  
Lai-Cheong J, McGrath J.  
*N Engl J Med*. 2010 Dec 23;363(26):e39. doi: 10.1056/NEJMicm1003685.  
PMID: 21175309
29. The molecular skin pathology of familial primary localized cutaneous amyloidosis.  
Tanaka A, Lai-Cheong JE, van den Akker PC, Nagy N, Millington G, Diercks GF, van Voorst Vader PC, Clements SE, Almaani N, Techanukul T, Hide M, South AP, McGrath JA.  
*Exp Dermatol*. 2010 May;19(5):416-23. doi: 10.1111/j.1600-0625.2010.01083.x.  
PMID: 20507362
30. Revertant mosaicism in recessive dystrophic epidermolysis bullosa.  
Almaani N, Nagy N, Liu L, Dopping-Hepenstal PJ, Lai-Cheong JE, Clements SE, Techanukul T, Tanaka A, Mellerio JE, McGrath JA.  
*J Invest Dermatol*. 2010 Jul;130(7):1937-40. doi: 10.1038/jid.2010.64.  
PMID: 20357813
31. Spectrum of mutations in the ANTXR2 (CMG2) gene in infantile systemic hyalinosi and juvenile hyaline fibromatosis.  
El-Kamah GY, Fong K, El-Ruby M, Afifi HH, Clements SE, Lai-Cheong JE, Amr K, El-Darouti M, McGrath JA.  
*Br J Dermatol*. 2010 Jul;163(1):213-5. doi: 10.1111/j.1365-2133.2010.09769.x.  
PMID: 20331448
32. A homozygous nonsense mutation within the dystonin gene coding for the coiled-coil domain of the epithelial isoform of BPAG1 underlies a new subtype of autosomal recessive epidermolysis bullosa simplex.  
Groves RW, Liu L, Dopping-Hepenstal PJ, Markus HS, Lovell PA, Ozoemena L, Lai-Cheong JE, Gawler J, Owaribe K, Hashimoto T, Mellerio JE, Mee JB, McGrath JA.  
*J Invest Dermatol*. 2010 Jun;130(6):1551-7. doi: 10.1038/jid.2010.19.  
PMID: 20164846

33.Somatic mitochondrial DNA deletions accumulate to high levels in aging human extraocular muscles.

Yu-Wai-Man P, Lai-Cheong J, Borthwick GM, He L, Taylor GA, Greaves LC, Taylor RW, Griffiths PG, Turnbull DM.

Invest Ophthalmol Vis Sci. 2010 Jul;51(7):3347-53. doi: 10.1167/iovs.09-4660.

PMID: 20164450

34.Kindler syndrome.

Lai-Cheong JE, McGrath JA.

Dermatol Clin. 2010 Jan;28(1):119-24. doi: 10.1016/j.det.2009.10.013.

PMID: 19945624

35.The role of kindlins in cell biology and relevance to human disease.

Lai-Cheong JE, Parsons M, McGrath JA.

Int J Biochem Cell Biol. 2010 May;42(5):595-603. doi: 10.1016/j.biocel.2009.10.015.

PMID: 19854292

36.Loss-of-function FERMT1 mutations in kindler syndrome implicate a role for fermitin family homolog-1 in integrin activation.

Lai-Cheong JE, Parsons M, Tanaka A, Ussar S, South AP, Gomathy S, Mee JB, Barbaroux JB, Techanukul T, Almaani N, Clements SE, Hart IR, McGrath JA.

Am J Pathol. 2009 Oct;175(4):1431-41. doi: 10.2353/ajpath.2009.081154.

PMID: 19762710

37.Novel IL31RA gene mutation and ancestral OSMR mutant allele in familial primary cutaneous amyloidosis.

Lin MW, Lee DD, Liu TT, Lin YF, Chen SY, Huang CC, Weng HY, Liu YF, Tanaka A, Arita K, Lai-Cheong J, Palisson F, Chang YT, Wong CK, Matsuura I, McGrath JA, Tsai SF.

Eur J Hum Genet. 2010 Jan;18(1):26-32. doi: 10.1038/ejhg.2009.135.

PMID: 19690585

38.New insight into mechanisms of pruritus from molecular studies on familial primary localized cutaneous amyloidosis.

Tanaka A, Arita K, Lai-Cheong JE, Palisson F, Hide M, McGrath JA.

Br J Dermatol. 2009 Dec;161(6):1217-24. doi: 10.1111/j.1365-2133.2009.09311.x.

PMID: 19663869

39.New glycine substitution mutations in type VII collagen underlying epidermolysis bullosa pruriginosa but the phenotype is not explained by a common polymorphism in the matrix metalloproteinase-1 gene promoter.

Almaani N, Liu L, Harrison N, Tanaka A, Lai-Cheong J, Mellerio JE, McGrath JA.

Acta Derm Venereol. 2009;89(1):6-11. doi: 10.2340/00015555-0605.

PMID: 19197535

40. Kindler syndrome: a focal adhesion genodermatosis.

Lai-Cheong JE, Tanaka A, Hawche G, Emanuel P, Maari C, Taskesen M, Akdeniz S, Liu L, McGrath JA.

Br J Dermatol. 2009 Feb;160(2):233-42. doi: 10.1111/j.1365-2133.2008.08976.x.

PMID: 19120339

41. Autosomal dominant junctional epidermolysis bullosa.

Almaani N, Liu L, Dopping-Hepenstal PJ, Lovell PA, Lai-Cheong JE, Graham RM, Mellerio JE, McGrath JA.

Br J Dermatol. 2009 May;160(5):1094-7. doi: 10.1111/j.1365-2133.2008.08977.x.

PMID: 19120338

42. Novel truncating mutations in PKP1 and DSP cause similar skin phenotypes in two Brazilian families.

Tanaka A, Lai-Cheong JE, Café ME, Gontijo B, Salomão PR, Pereira L, McGrath JA.

Br J Dermatol. 2009 Mar;160(3):692-7. doi: 10.1111/j.1365-2133.2008.08900.x.

PMID: 19016709

43. Colocalization of kindlin-1, kindlin-2, and migfilin at keratinocyte focal adhesion and relevance to the pathophysiology of Kindler syndrome.

Lai-Cheong JE, Ussar S, Arita K, Hart IR, McGrath JA.

J Invest Dermatol. 2008 Sep;128(9):2156-65. doi: 10.1038/jid.2008.58.

PMID: 18528435

44. Indeterminate cell histiocytosis responding to total skin electron beam therapy.

Malhomme de la Roche H, Lai-Cheong JE, Calonje E, Davies M, Morris S, Whittaker SJ.

Br J Dermatol. 2008 Apr;158(4):838-40. doi: 10.1111/j.1365-2133.2007.08409.x.

PMID: 18241280

45. Oncostatin M receptor-beta mutations underlie familial primary localized cutaneous amyloidosis.

Arita K, South AP, Hans-Filho G, Sakuma TH, Lai-Cheong J, Clements S, Odashiro M, Odashiro DN, Hans-Neto G, Hans NR, Holder MV, Bhogal BS, Hartshorne ST, Akiyama M, Shimizu H, McGrath JA.

Am J Hum Genet. 2008 Jan;82(1):73-80. doi: 10.1016/j.ajhg.2007.09.002.

PMID: 18179886

46. Genetic diseases of junctions.

Lai-Cheong JE, Arita K, McGrath JA.

J Invest Dermatol. 2007 Dec;127(12):2713-25.

PMID: 18007692

47. Focal dermal hypoplasia resulting from a new nonsense mutation, p.E300X, in the PORCN gene.

Clements SE, Wessagowit V, Lai-Cheong JE, Arita K, McGrath JA.

J Dermatol Sci. 2008 Jan;49(1):39-42.

PMID: 17951029

48.Recurrent KIND1 (C20orf42) gene mutation, c.676insC, in a Brazilian pedigree with Kindler syndrome.

Martignago BC, Lai-Cheong JE, Liu L, McGrath JA, Cestari TF.

Br J Dermatol. 2007 Dec;157(6):1281-4.

PMID: 17916195

49.Unusual molecular findings in Kindler syndrome.

Arita K, Wessagowit V, Inamadar AC, Palit A, Fassihi H, Lai-Cheong JE, Pourreyron C, South AP, McGrath JA.

Br J Dermatol. 2007 Dec;157(6):1252-6.

PMID: 17854379

50.Highly active antiretroviral therapy: a treatment for cutaneous polyarteritis nodosa-like syndrome in a HIV positive patient?

Lai-Cheong JE, Tang V, Mazhude C, Baker L, Menagé Hdu P.

J Eur Acad Dermatol Venereol. 2007 Sep;21(8):1138-40.

PMID: 17714158

51.Linear IgA bullous dermatosis associated with adenocarcinoma of the ascending colon.

Lai-Cheong JE, Groves RW, Banerjee P.

J Eur Acad Dermatol Venereol. 2007 Aug;21(7):978-9.

PMID: 17659011

52.Five new homozygous mutations in the KIND1 gene in Kindler syndrome.

Lai-Cheong JE, Liu L, Sethuraman G, Kumar R, Sharma VK, Reddy SR, Vahlquist A, Pather S, Arita K, Wessagowit V, McGrath JA.

J Invest Dermatol. 2007 Sep;127(9):2268-70.

PMID: 17460733

53.Cutaneous manifestations of tuberculosis.

Lai-Cheong JE, Perez A, Tang V, Martinez A, Hill V, Menagé Hdu P.

Clin Exp Dermatol. 2007 Jul;32(4):461-6.

PMID: 17376216

54.Bullous acute haemorrhagic oedema of skin in infancy.

Lai-Cheong JE, Banerjee P, Hill V, Kenny P, Ross J.

Clin Exp Dermatol. 2007 Jul;32(4):467-8.

PMID: 17376214

55.A rash on the back. Diagnosis: confluent and reticulated papillomatosis (CRP) of Gougerot and Carteaud.

Chaudhry SI, Lai Cheong JE, O'Donoghue NB.

Clin Exp Dermatol. 2006 Sep;31(5):727-8.

PMID: 16901326

56. Etanercept-induced dermatitis in a patient with rheumatoid arthritis.

Lai-Cheong J, Warren R, Bucknall R, Parslew R.

J Eur Acad Dermatol Venereol. 2006 May;20(5):614-5.

PMID: 16684300

57. Cutaneous manifestations of internal malignancy: diagnosis and management.

Kleyn CE, Lai-Cheong JE, Bell HK.

Am J Clin Dermatol. 2006;7(2):71-84.

PMID: 16605288

58. Blistering plaques.

Lai Cheong JE, Chaudhry SI, Black MM.

Clin Exp Dermatol. 2006 Jan;31(1):163-4.

PMID: 16309532

59. Unusual scars in a young female patient.

Lai Cheong JE, Rajpara SM, Azurdia RM.

Postgrad Med J. 2005 Jul;81(957):e4-5.

PMID: 15998813

60. Retinal vein thrombosis associated with a herbal phytoestrogen preparation in a susceptible patient.

Lai Cheong JE, Bucknall R.

Postgrad Med J. 2005 Apr;81(954):266-7. Erratum in: Postgrad Med J. 2006 Feb;82(964):135.

Cheong, JL [corrected to Lai Cheong, JE].

PMID: 15811894

61. Molecular abnormalities of the desmosomal protein desmoplakin in human disease.

Lai Cheong JE, Wessagowit V, McGrath JA.

Clin Exp Dermatol. 2005 May;30(3):261-6.

PMID: 15807686