

CURRICULUM VITAE

Name: Keith A. Choate, M.D., Ph.D.

Professor, Department of Dermatology, with Secondary Appointments in the Department of Pathology and Department of Genetics

School: Yale University School of Medicine and the Graduate School

Education:

B.S. Stanford University (Biological Sciences) 1995
M.Phil. Yale University School of Medicine (Cell Biology) 2000
Ph.D. Yale University School of Medicine (Cell Biology) 2001
M.D. Yale University School of Medicine 2004

Career/Academic Appointments:

2004-2005 Intern, Internal Medicine, Yale-New Haven Hospital, New Haven, CT
2005-2008 Resident, Dermatology, Yale-New Haven Hospital, New Haven, CT
2006-2008 Post-Doctoral Fellow in Dermatology and Genetics, Yale University School of Medicine, New Haven, CT
2008-2009 Instructor, Dept. of Dermatology, Yale University School of Medicine, New Haven, CT
2009-2013 Assistant Professor, Dept. of Dermatology, Yale University School of Medicine, New Haven, CT
2014-2017 Associate Professor, Depts. of Dermatology, Pathology, and Genetics, Yale University School of Medicine, New Haven, CT
2015-present Co-chief of Dermatology, Yale-New Haven Hospital, Saint Raphael Campus
2015-present Member, Yale Cancer Center: Program in Cancer Genetics and Genomics, Yale University School of Medicine, New Haven, CT
2015-present Associate Director, Medical Scientist Training Program
2016-present Director of Research, Dept. of Dermatology, Yale University School of Medicine, New Haven, CT
2017-present Professor, Depts. of Dermatology, Pathology, and Genetics, Yale University School of Medicine, New Haven, CT

Board Certification:

American Board of Dermatology, 2008

Professional Honors & Recognition:

International/National/Regional

2016: Elected Member of The American Society for Clinical Investigation
2011: Young Investigator Award, American Academy of Dermatology
2004: Alpha Omega Alpha election
2004: M.D. awarded *cum laude*

Grants/Clinical Trials History:

Current Grants

Agency: NIH/NIAMS
I.D.# R01 AR071491
Title: Pathobiology of Cutaneous Mosaic Disorders
PI: Keith Choate
Percent effort: 45%
Direct costs per year: \$375,000
Total costs for project period: \$ 3,524,000
Project period: 03/01/2018– 02/28/2023

Agency: NIH/NIAMS
ID #: R01 AR068392
Title: Genetics and Pathobiology of Disorders of Keratinization
PI: Keith Choate
Percent effort: 30%
Direct costs per year: \$339,000
Total costs for project period: \$ 2,831,000
Project period: 07/01/2015– 06/30/2020

Agency: NIH/NIAMS
ID #: T32 AR068392
Title: Training in Investigative Dermatology
PI: Michael Girardi
Percent effort: N/A, Co-Director
Direct costs per year: \$237,984
Total costs for project period: \$ 1,255,115
Project period: 07/01/2016– 06/30/2021

Agency: Foundation for Ichthyosis and Related Skin Types
I.D.# Ichthyosis Registry Award
Title: N/A
PI: Keith Choate
Role on Project: PI
Percent effort: 1%
Direct costs per year: \$50,000
Total costs for project period: \$250,000
Project period: 08/01/2016 – 5/31/21

Invited Speaking Engagements, Presentations, Symposia & Workshops Not Affiliated With Yale:

International/National

- 2016: Grand Rounds Lecture, SUNY Downstate, “Genetic investigation of rare inherited and mosaic skin disorders reveals novel pathways for disease pathogenesis.”
- 2016: Grand Rounds Lecture, Brown University, Providence, RI, “Genetic investigation of rare inherited and mosaic skin disorders reveals novel pathways for disease pathogenesis.”
- 2015: Annual Meeting, Pediatric Dermatology Research Alliance, Dallas, TX, “Obtaining Funding for Your Research Program.”
- 2015: Reed Lecture in Dermatologic Genetics, University of California at San Francisco, San Francisco, CA, “Genetic Investigation of Rare Inherited and Mosaic Skin Disorders Reveals Novel Pathways for Disease Pathogenesis.”
- 2015: World Congress of Dermatology, Vancouver, CA, Genodermatoses - A Practical Approach to Molecular Diagnosis, “The Ichthyoses.”
- 2015: World Congress of Dermatology, Vancouver, CA, Pediatric Dermatology Basic and Advanced, “Update on Ichthyosis.”
- 2015: World Congress of Dermatology, Vancouver, CA, Genodermatoses - A Practical Approach to Molecular Diagnosis, “Update on Autosomal Dominant Congenital Ichthyosis.”
- 2015: World Congress of Dermatology, Vancouver, CA, Pediatric Dermatology Basic and Advanced, “Update on Ichthyosis.”
- 2015: Annual Meeting, American Academy of Dermatology, San Francisco, CA, Genetics and Genetic Testing Forum, “Exome Sequencing: New Mutations in Old Genes.”
- 2015: Annual Meeting, American Academy of Dermatology, San Francisco, CA, Syndrome Update Forum, “Genetic Investigation of Rare Skin Disorders Reveals Novel Pathways for Disease Pathogenesis.”
- 2014: Annual Meeting, Society for Pediatric Dermatology, Cour D’Alene ID, Plenary lecture, “Insights into Ichthyosis Pathogenesis Through Genetic Investigation.”
- 2014: Annual Meeting, Society for Investigative Dermatology, Albuquerque, NM, Genetics and Genomic Session, “Somatic activating mutations in HRAS and NRAS cause cutaneous skeletal hypophosphatemic syndrome.”
- 2014: Levin Lecture, Northwestern University, Chicago, IL, “Gene Discovery in Rare Inherited and Mosaic Disorders.”
- 2014: Annual Meeting, American Academy of Dermatology, Miami, FL, Genetics and Genetic Testing: Beyond Mendel Forum, “Exome Sequencing: New Mutations in Old Genes.”
- 2014: Pediatric Dermatology Research Alliance, Chicago, IL, “Lessons learned in the Yale Disorders of Keratinization Project.”
- 2013: Pediatric Dermatology Research Alliance Inaugural Meeting , Chicago, IL, “Gene discovery in rare mosaic and inherited skin disorders.”
- 2013: World Congress of Pediatric Dermatology, Madrid, Spain, Erythroderma in Children Session, “Advances in Ichthyosiform Erythroderma.”
- 2013: Annual Meeting, American Academy of Dermatology, Miami, FL, Careers in academic dermatology Symposium, “A Career in Academic Dermatology – Clinician Scientist Perspective.”
- 2013: Annual Meeting, American Academy of Dermatology, Miami, FL, Genetics and Genetic Testing forum, “Gene discovery in rare and inherited and mosaic skin disease.”
- 2013: Annual Meeting, American Academy of Dermatology, Miami, FL, Living with Skin Disease

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Symposium, “Next generation sequencing approaches to disorders of keratinization and mosaic disorders.”

- 2013: Annual Meeting, American Academy of Dermatology, Miami, FL, It’s all in the genes: Genetics and Genomics in Dermatology Symposium, “Gene discovery in rare and inherited and mosaic skin disorders.”
- 2012: Annual Meeting, Society for Investigative Dermatology, Raleigh, NC, State-of-the-Art Plenary Lecture “Opportunities for discovery utilizing next generation sequencing technology.”
- 2012: Annual Meeting, Society for Pediatric Dermatology, Monterrey, CA, Research forum, “Gene discovery in disorders of keratinization via whole exome sequencing.”
- 2012: Grand Rounds Lecture, Pediatric Dermatology Super Wednesday, Department of Dermatology, Columbia University, New York, NY, “Opportunities for gene discovery in rare inherited and mosaic genetic skin disorders.”
- 2012: Trainee Lecture, Pediatric Dermatology Super Wednesday, Department of Dermatology, Columbia University, New York, NY, “Disorders of Keratinization: Approach to Diagnosis and Management.”
- 2012: Grand Rounds Lecture, Department of Dermatology, Massachusetts General Hospital, Harvard University, Boston, MA, “Approaches to genetic diagnosis and gene discovery in rare disorders of keratinization and mosaic disorders.”
- 2012: Scientific Lecture, Genome Institute, Washington University, St. Louis, MO, “Gene discovery in rare disorders of keratinization and mosaic disorders.”
- 2012: Grand Rounds Lecture, Department of Dermatology, Washington University, St. Louis, MO, “Widespread somatic reversion of dominant mutations in *KRT10* in ichthyosis with confetti.”
- 2012: Grand Rounds Lecture, Department of Dermatology, Johns Hopkins University, Baltimore, MD, “Widespread somatic reversion of dominant mutations in *KRT10* in ichthyosis with confetti.”
- 2011: Annual Meeting, Society for Investigative Dermatology, Phoenix, AZ, Clinical Scholars Symposium, “Mitotic recombination in ichthyosis with confetti causes widespread reversion of dominant mutations in *KRT10*.”
- 2011: Duhring Lecture, Department of Dermatology, University of Pennsylvania, Philadelphia, PA, “Widespread somatic reversion of dominant mutations in *KRT10* in ichthyosis with confetti.”
- 2010: Frontiers in Ichthyosis Meeting, Foundation for Ichthyosis and Related Skin Types. Orlando, FL, “Somatic events facilitate identification of disease-causing genes.”

Regional:

- 2014: New England Dermatological Society Meeting. New Haven, CT. “Gene Discovery in Rare Inherited and Mosaic Disorders”
- 2014: Quinsigamond Dermatological Society Meeting. New Haven, CT. “Genetic Insights into Rare Inherited and Mosaic Disorders”
- 2013: Advances in Genome Science, Connecticut Illumina User’s Group Meeting, New Haven, CT, “Gene discovery in rare inherited and mosaic skin Disorders.”
- 2009: New England Dermatological Society Meeting. New Haven, CT. “Clinical update: The genetic basis of epidermolysis bullosa pruriginosa”

Peer-Reviewed Presentations & Symposia Given at Meetings Not Affiliated With Yale:

- 2012: Annual Meeting, Society for Investigative Dermatology, Raleigh, NC, Genomics Workshop,

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“A sequence of events: Approaches to gene discovery using exome sequencing.”

- 2012: Pre-AAD Meeting, Society for Pediatric Dermatology, New Orleans, LA, Cases of the Year, “Mutations in *Keratin-10* cause widespread revertant mosaicism in ichthyosis with confetti”
- 2011: Annual Meeting, Society for Pediatric Dermatology, Monterey, CA, Research Forum, “Gene Discovery in Disorders of Keratinization via Whole Exome Sequencing”
- 2011: Annual Meeting, European Society for Dermatology Research, Barcelona, Spain, Natural Gene Therapy of the Skin Symposium, “Mitotic recombination in ichthyosis with confetti causes widespread reversion of dominant mutations in *KRT10*”
- 1999: 30th Annual Meeting, Congress of Nephrology, Freiburg, Germany, Understanding Tubular Transport: Genes Lead the Way Symposium, “Positional cloning and characterization of *Paracellin-1*, a novel tight junction protein required for paracellular Mg⁺⁺ reabsorption”
- 1999: Annual Meeting of the American Society of Nephrology. Miami, FL, “Paracellin-1 localizes to tight junctions in the thick ascending limb, revealing its role in paracellular Mg²⁺ reabsorption”
- 1997: Annual Meeting, Society for Investigative Dermatology, Washington, D.C., Plenary Session in Gene Therapy, “Corrective impact of direct plasmid delivery versus viral gene transfer in lamellar ichthyosis *in vivo*”
- 1996: Annual Meeting, Society for Investigative Dermatology, Washington, D.C., Plenary Session, “Corrective gene delivery in lamellar ichthyosis”

Professional Service

Peer Review Groups/Grant Study Sections:

- 2015- NIH/ACTS study section member
- 2014- Member, AAD Young Investigator Award Grant Review Committee
- 2012-2015 Member, Dermatology Foundation Grant Review Committee
- 2010 Member, Foundation for Ichthyosis and Related Skin Types Grant Review Committee

Journal Service:

Associate Editor:

The British Journal of Dermatology
JCI Insight

Section Editor:

The Journal of Investigative Dermatology

Reviewer:

2009-present Reviewer for *Nature Genetics*, *PLOS Genetics*, *The British Journal of Dermatology*, *JAMA Dermatology*, *eLife*, *Pediatric Dermatology*, *The Journal of Investigative Dermatology*

Professional Service for Professional Organizations:

American Board of Dermatology

2013-present Member, Basic Science Test Question Writing Committee

Society for Investigative Dermatology

Keith A. Choate, M.D., Ph.D.

2005-10 Resident/ Fellow Member, Board of Directors
2009 Representative, Skin Disease Research Day at the Capitol

Foundation for Ichthyosis and Related Skin Types

2010-Present Member, Medical and Scientific Advisory Board
2007-Present Member, Clinical Screening Panel, Biennial Family Conference

NIH/NIAMS

2016 Co-Director, NIAMS Roundtable on Ichthyosis
2016 Member, ACTS study section
2013 Panelist, NIAMS Skin Biology and Diseases Listening Session
2012 Panelist, NIAMS Forum for Clinical Mentored K Awardees
2012 Panelist, NIAMS Roundtable on Pediatric Dermatology
2012 Panelist, NIAMS Listening Session with Dermatology Research Community

Pediatric Dermatology Research Alliance

2012-present Member, Scientific Advisory Committee and Meeting Planning Committee
2012 Panelist, PeDRA planning workshop
2012 Member, R13 Grant Writing Committee

Meeting Planning/Participation

- 2015-present Society for Investigative Dermatology Genetic Disease Forum Moderator
2013-present Pediatric Dermatology Research Alliance Meeting Planning Committee
2013 Pediatric Dermatology Research Alliance Inaugural Meeting, Scientific Co-director and Member of Planning Committee
2013 Society for Investigative Dermatology, Faculty Mentor for Retreat for Future Academicians, Annual Meeting
2011-present Society for Pediatric Dermatology, Abstract Selection Committee: Basic and Translational Research session
2011-present Society for Investigative Dermatology, Abstract Selection Committee: Pediatric Dermatology Research Symposium
2011 Society for Investigative Dermatology, Blank Forum Organizing Committee, 2012 Annual Meeting

Yale University Service:

Medical School Committees

- 2016 Pediatrics Search Committee for Chairman
2015-present Associate Director, Medical Scientist Training Program
2011-present Member, Medical Scientist Training Program Committee

Departmental Committees

- 2004-present Director, Yale Visiting Professor Lecture Series Coordinating Committee

Hospital Boards & Committees

- 2012-present Member, Pediatric Dermatology Fellowship Clinical Competency Committee

Public Service:

- 2012 Featured Expert, WCBS News, *Your Health: Shingles*, WCBS Channel 3 News

Bibliography

Peer-Reviewed Original Research:

1. Freiberg RA, Spencer DM, **Choate KA**, Peng PD, Schreiber SL, Crabtree GR and Khavari PA. Specific triggering of the Fas signal transduction pathway in normal human keratinocytes. *Journal of Biological Chemistry* 1996; 271: 31666-9.
2. **Choate KA**, Medalie DA, Morgan JR, and Khavari PA. Corrective gene delivery in the human skin disease lamellar ichthyosis. *Nature Medicine* 1996; 2: 1263-1267.
3. **Choate KA**, Kinsella T, Williams M, Nolan G, and Khavari PA. Transglutaminase 1 delivery to lamellar ichthyosis keratinocytes. *Human Gene Therapy* 1996; 7:2247-2253.
4. Freiberg RA, Spencer DM, **Choate KA**, Duh HJ, Schreiber SL, Crabtree GR and Khavari PA. Fas signal transduction triggers either proliferation or apoptosis in human fibroblasts. *Journal of Investigative Dermatology* 1997; 108:215-219.
5. **Choate KA** and Khavari PA. Sustainability of keratinocyte gene transfer and cell survival in vivo. *Human Gene Therapy* 1997; 8: 895-901.

6. Freiberg RA, **Choate KA**, Deng H, Alperin E, Shapiro LJ, and Khavari PA. A model of gene transfer in X-linked ichthyosis. *Human Molecular Genetics* 1997; 6:927-33.
7. **Choate KA** and Khavari PA. Direct cutaneous gene delivery in a human genetic skin disease. *Human Gene Therapy* 1997; 8:1659-65.
8. **Choate KA**, Williams ML, and Khavari PA. Abnormal epidermal gene expression in a subset of patients with autosomal recessive ichthyosis and erythroderma. *Journal of Investigative Dermatology* 1998; 110:8-12.
9. **Choate KA**, Williams ML, Elias PM, and Khavari PA. Keratinocyte transglutaminase in harlequin ichthyosis. *Journal of the American Academy of Dermatology* 1998; 38:325-9.
10. Deng H, **Choate KA**, Lin Q, Khavari PA. High efficiency gene transfer and pharmacologic selection of genetically engineered human keratinocytes. *BioTechniques* 1998; 25:274-280.
11. Simon DB & Lu Y, **Choate KA**, Velazquez H, Al-Sabban E, Praga M, Casari G, Bettinelli A, Colussi G, Rodriguez-Soriano J, McCredie D, Milford D, Sanjad S, Lifton RP. Paracellin-1, a renal tight junction protein required for paracellular Mg²⁺ resorption. *Science* 1999; 285:103-6.
12. Smith AN, Skaug J, **Choate KA**, Nayir A, Bakkaloglu A, Ozen S, Hulton SA, Sanjad SA, Al-Sabban EA, Lifton RP, Scherer SW, Karet FE. Mutations in *ATP6N1B*, encoding a new kidney vacuolar proton pump 116-kD subunit, cause recessive distal renal tubular acidosis with preserved hearing. *Nature Genetics* 2000; 26:71-5.
13. Wilson FH, Disse-Nicodeme S & **Choate KA**, Ishikawa K, Nelson-Williams C, Desitter I, Gunel M, Milford DV, Lipkin GW, Achard JM, Feely MP, Dussol B, Berland Y, Unwin RJ, Mayan H, Simon DB, Farfel Z, Jeunemaitre X, Lifton RP. Human hypertension caused by mutations in WNK kinases. *Science* 2001; 293:1107-12.
14. Gunel M, Laurans MS, Shin D, DiLuna ML, Voorhees J, **Choate K**, Nelson-Williams C, Lifton RP. *KRT11*, a gene mutated in cerebral cavernous malformation, encodes a microtubule-associated protein. *Proceedings of the National Academy of Sciences USA* 2002; 99:10677-82.
15. **Choate KA**, Kahle KT, Wilson FH, Nelson-Williams C, Lifton RP. *WNK1*, a kinase mutated in inherited hypertension with hyperkalemia, localizes to diverse Cl⁻-transporting epithelia. *Proceedings of the National Academy of Sciences USA* 2003; 100:663-8.
16. **Choate KA**, Lu Y, Zhou J, Choi M, Elias P, Farhi A, Nelson-Williams C, Crumrine D, Williams ML, Nopper AJ, Bree AF, Milstone LM, Lifton RP. Mitotic recombination in patients with ichthyosis causes reversion of dominant mutations in *KRT10*. *Science* 2010; 330:94-7.
17. Yang CS, Lu Y, Farhi A, Nelson-Williams C, Kashgarian M, Glusac EJ, Lifton RP, Antaya RJ, **Choate KA**. An incompletely penetrant novel mutation in *COL7A1* causes epidermolysis bullosa pruriginosa and dominant dystrophic epidermolysis bullosa phenotypes in an extended kindred. *Pediatric Dermatology* 2012; 10: 1525-1470.
18. Ko CJ, McNiff JM, Bosenberg M, **Choate K**. Keratoacanthoma: clinical and histopathologic features of regression. *Journal of the American Academy of Dermatology* 2012; 67:1008-12.
19. Boyden LM, Orme CM, Antaya RJ, **Choate KA**, King BA. Capillary malformation-arteriovenous malformation syndrome: Identification of a family with a novel mutation. *Journal of the American Academy of Dermatology* 2012; 67:e287-9.
20. Boyden LM, Choi M, **Choate KA**, Nelson Williams CJ, Farhi A, Toka HR, Tikhonova IR, Bjornson R, Mane SM, Colussi G, Lebel M, Gordon RD, Semmekrot BA, Poujol A, Vlimki J, De Ferrari ME, Sanjad SA, Gutkin M, Karet FE, Tucci JR, Stockigt JR, Keppler Noreuil KM, Porter C, Anand K, Whiteford ML, Davis ID, Dewar SB, Bettinelli A, Fadrowski J, Belsha CW, Hunley TE, Nelson RD,

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21. Levinsohn JL, Tian LC, Boyden LM, McNiff JM, Narayan D, Loring ES, Yun D, Sugarman JL, Overton JD, Mane SM, Lifton RP, Paller AS, Wagner AM, Antaya RJ, **Choate KA**. Whole-Exome Sequencing Reveals Somatic Mutations in *HRAS* and *KRAS*, which Cause Nevus Sebaceus. *Journal of Investigative Dermatology* 2013; 133: 827-830.
 22. Lim YH, Ovejero D, Sugarman JS, Deklotz CM, Maruri A, Eichenfield LF, Kelley PK, Jüppner H, Gottschalk M, Tiffit CJ, Gafni RI, Boyce AM, Cowen EW, Bhattacharyya N, Guthrie LC, Gahl WA, Golas G, Loring EC, Overton JD, Mane SM, Lifton RP, Levy ML, Collins MT, **Choate KA**. Multi-lineage somatic activating mutations in *HRAS* and *NRAS* cause mosaic cutaneous and skeletal lesions, elevated FGF23, and hypophosphatemia. *Hum Mol Genet.* 2014; 23(2):397-407.
 23. Levinsohn JL, Teng K, Craiglow BG, Loring EC, Burrow TA, Mane SS, Overton JD, Lifton RP, McNiff JM, Lucky AW, **Choate KA**. Somatic *HRAS* p.G12S Mutation Causes Woolly Hair and Epidermal Nevi. *Journal of Investigative Dermatology.* 2014; 134(4):1149-52.
 24. Zambrano H, Montalvan M, Cabezas J, Lu Y, Yang CS, Milstone LM, **Choate K**. Mutations in *TGM1* in Ecuadorians with autosomal recessive congenital ichthyosis. *Int J Dermatol.* 2014; 53(4):e312-3.
 25. Koochek A, **Choate KA**, Milstone LM. Harlequin ichthyosis: neonatal management and identification of a new *ABCA12* mutation. *Pediatric Dermatology.* 2014; 31(2):e63-4.
 26. Siegel DH, **Choate KA**, Drolet BA, Frieden IJ, Rittenberg S, Teng JM, Tom WL, Williams ML, Eichenfield LF, Paller AS. Proceedings of the Inaugural Pediatric Dermatology Research Alliance (PeDRA) conference. *Journal of Investigative Dermatology.* 2014; 134(11):2671-4.
 27. Boyden LM, Craiglow BG, Zhou J, Hu R, Loring EC, Morel KD, Lauren CT, Lifton RP, Bilguvar K, Paller AS, **Choate KA**. Dominant *De Novo* Mutations in *GJAI* Cause Erythrokeratoderma Variabilis Et Progressiva, without Features of Oculodentodigital Dysplasia. *Journal of Investigative Dermatology.* 2015;135(6):1540-7.
 28. **Choate KA**, Lu Y, Zhou J, Elias PM, Zaidi S, Paller AS, Farhi A, Nelson-Williams C, Crumrine D, Milstone LM, Lifton RP. Frequent somatic reversion of *KRT1* mutations in ichthyosis with confetti. *J Clin Invest.* 2015; 125(4):1703-7.
 29. Levinsohn JL, McNiff JM, Antaya RJ, Choate KA. A Somatic p.G45E *GJB2* Mutant Causing Porokeratotic Eccrine Ostial and Dermal Duct Nevus. *JAMA Dermatology.* 2015; 151(6):638-41.
 30. Lim YH, Douglas SR, Ko CJ, Antaya RJ, McNiff JM, Zhou J, Yale Center for Genome Analysis, **Choate KA***, and Narayan D*. Somatic Activating RAS Mutations Cause Vascular Tumors Including Pyogenic Granuloma. *Journal of Investigative Dermatology.* 2015; 135(6):1698-700.
 31. Levinsohn JL, Sugarman JL, Bilguvar K, McNiff JM, The Yale Center for Mendelian Genomics, **Choate KA**. Somatic V600E BRAF mutation causes linear and sporadic syringocystadenoma pailliferum. *Journal of Investigative Dermatology.* 2015; 135(10):2536-8
 32. Boyden LM, Kam CY, Hernández-Martín A, Zhou J, Craiglow BG, Sidbury R, Mathes EF, Maguiness SM, Crumrine DA, Williams ML, Hu R, Lifton RP, Elias PM, Green KJ, **Choate KA**. Dominant de novo DSP mutations cause erythrokeratoderma-cardiomyopathy syndrome. *Hum Mol Genet.* 2016; 25(2):348-57.
 33. Lim YH, Odell ID, Ko CJ, **Choate KA**. Somatic p.T771R KDR (VEGFR2) Mutation Arising in a Sporadic Angioma During Ramucirumab Therapy. *JAMA Dermatol.* 2015; 151(11):1240-3.
 34. Salas-Alanis JC, Wozniak E, Mein CA, Duran Mckinster CC, Ocampo-Candiani J, Kelsell DP, Hua

- R, Garza-Rodriguez ML, **Choate KA**, Barrera Saldaña HA. Mutations in EDA and EDAR Genes in a Large Mexican Hispanic Cohort with Hypohidrotic Ectodermal Dysplasia. *Ann Dermatol*. 2015; 27(4):474-7.
35. Mirza H, Kumar A, Craiglow BG, Zhou J, Saraceni C, Torbeck R, Ragsdale B, Rehder P, Ranki A, **Choate KA**. Mutations Affecting Keratin 10 Surface-Exposed Residues Highlight the Structural Basis of Phenotypic Variation in Epidermolytic Ichthyosis. *J Invest Dermatol*. 2015; 135(12):3041-50.
36. Lim YH, Fisher JM, Bosenberg MW, **Choate KA**, Ko CJ. Keratoacanthoma Shares Driver Mutations with Cutaneous Squamous Cell Carcinoma. *J Invest Dermatol*. 2016; 136(8):1737-41.
37. Levinsohn JL, Sugarman JL; Yale Center for Mendelian Genomics, McNiff JM, Antaya RJ, **Choate KA**. Somatic Mutations in *NEK9* Cause Nevus Comedonicus. *Am J Hum Genet*. 2016; 98(5):1030-7.
38. Lim YH, Qiu J, Saraceni C, Burrall BA, **Choate KA**. Genetic Reversion via Mitotic Recombination in Ichthyosis with Confetti due to a *KRT10* Polyalanine Frameshift Mutation. *J Invest Dermatol*. 2016; 136(8):1725-8.
39. Lim YH, Bacchiocchi A, Qiu J, Straub R, Bruckner A, Bercovitch L, Narayan D; Yale Center for Mendelian Genomics, McNiff J, Ko C, Robinson-Bostom L, Antaya R, Halaban R, **Choate KA**. *GNA14* Somatic Mutation Causes Congenital and Sporadic Vascular Tumors by MAPK Activation. *Am J Hum Genet*. 2016; 99(2):443-50.
40. Paller AS, Renert-Yuval Y, Suprun M, Esaki H, Oliva M, Huynh TN, Ungar B, Kunjrvia N, Friedland R, Peng X, Zheng X, Estrada YD, Krueger JG, **Choate KA**, Suárez-Fariñas M, Guttman-Yassky E. An IL-17-dominant immune profile is shared across the major orphan forms of ichthyosis. *J Allergy Clin Immunol*. 2016 ePub ahead of press.
41. Marukian NV, Levinsohn JL, Craiglow BG, Milstone LM, **Choate KA**. Palmoplantar Keratoderma in Costello Syndrome Responsive to Acitretin. *Pediatr Dermatol*. 2017 Mar;34(2):160-162. PMID: 28008647.
42. Marukian NV, Hu RH, Craiglow BG, Milstone LM, Zhou J, Theos A, Kaymakcalan H, Akkaya DA, Uitto JJ, Vahidnezhad H, Youssefian L, Bayliss SJ, Paller AS, Boyden LM, **Choate KA**. Expanding the Genotypic Spectrum of Bathing Suit Ichthyosis. *JAMA Dermatol*. 2017 Jun 1;153(6):537-543. PMID: 28403434.
43. Boyden LM, Craiglow BG, Hu RH, Zhou J, Browning J, Eichenfield L, Lim YL, Luu M, Randolph LM, Ginarte M, Fachal L, Rodriguez-Pazos L, Vega A, Kramer D, Yosipovitch G, Vahidnezhad H, Youssefian L, Uitto J, Lifton RP, Paller AS, Milstone LM, **Choate KA**. Phenotypic spectrum of autosomal recessive congenital ichthyosis due to PNPLA1 mutation. *Br J Dermatol*. 2017 Jul;177(1):319-322. PMID: 28403545.
44. Boyden LM, Vincent NG, Zhou J, Hu R, Craiglow BG, Bayliss SJ, Rosman IS, Lucky AW, Diaz LA, Goldsmith LA, Paller AS, Lifton RP, Baserga SJ, **Choate KA**. Mutations in *KDSR* Cause Recessive Progressive Symmetric Erythrokeratoderma. *Am J Hum Genet*. 2017 Jun 1;100(6):978-984. PMID: 28575652.
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Chapters, Books, and Reviews:

46. **Choate KA**, Lu Y, and Lifton RP. Claudins mediate specific paracellular fluxes in vivo: Paracellin-1 is required for paracellular Mg²⁺ flux. In Tight Junctions, 2nd edition, eds. Cereijido M and Anderson J. CRC Press, New York, NY, 2001, pp. 483-492.
47. Lifton RP, Wilson FH, **Choate KA**, Geller DS. Salt and blood pressure: new insight from human genetic studies. *Cold Spring Harbor Symposia on Quantitative Biology*. 2002; 67:445-50.
48. Milstone LM, Choate KA. Improving outcomes for harlequin ichthyosis. *J Am Acad Dermatol*. 2013; 69(5):808-9.
49. Choate KA, Milstone LM. Phenotypic Expansion in Ichthyosis With Confetti. *JAMA Dermatology*. 2014; 151(1):15-6.
50. Ovejero D, Lim YH, Boyce AM, Gafni RI, McCarthy E, Nguyen TA, Eichenfield LF, DeKlotz CM, Guthrie LC, Tosi LL, Thornton PS, **Choate KA**, Collins MT. Cutaneous skeletal hypophosphatemia syndrome: clinical spectrum, natural history, and treatment. *Osteoporos Int*. 2016
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