

CURRICULUM VITAE

Daniel Lindsley Kastner

Place of Birth Lockport, New York
Marital Status Married, two children

Education

1973 AB, Princeton University, *summa cum laude*
Princeton, New Jersey
Major: Philosophy

1979 PhD, Baylor College of Medicine
Houston, Texas
Department of Microbiology and Immunology
PhD Thesis: "Role of the Qa-1 Region in Cell-Mediated Immune Responses"
Advisor: Robert R Rich, MD

1982 MD, Baylor College of Medicine, with honor
Houston, Texas

Employment

1982-84 Resident in Internal Medicine
Baylor College of Medicine Affiliated Hospitals
Houston, Texas

1985 Chief Resident in Internal Medicine
Baylor College of Medicine
Houston, Texas

1985-87 Medical Staff Fellow in Rheumatology
National Institute of Arthritis, Diabetes, and Digestive and Kidney Diseases
National Institutes of Health
Bethesda, Maryland

1987-90 Arthritis Foundation Fellow
Arthritis and Rheumatism Branch (ARB)
National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS)
National Institutes of Health
Bethesda, Maryland

1990-93 Senior Staff Fellow
ARB/NIAMS/NIH

1992-95 Acting Chief, Genetics Section
ARB/NIAMS/NIH

1993-2001 Senior Investigator
ARB/NIAMS/NIH

1995-2001	Chief, Genetics Section ARB/NIAMS/NIH
2001-2009	Chief, Genetics and Genomics Branch NIAMS/NIH
2005-2010	Clinical Director and Director of Translational Research, Intramural Research Program, NIAMS/NIH
2009-2010	Chief, Laboratory of Clinical Investigation, NIAMS/NIH
2008-2011	Deputy Director for Intramural Clinical Research, NIH
2010-2021	Scientific Director Division of Intramural Research National Human Genome Research Institute (NHGRI)
2019	Acting Clinical Director Division of Intramural Research, NHGRI
2021-present	NIH Distinguished Investigator and Chief, Inflammatory Disease Section Metabolic, Cardiovascular and Inflammatory Disease Genomics Branch Division of Intramural Research, NHGRI

Awards and Honors

1969	National Merit Scholarship Princeton University, Princeton, New Jersey
1973	Alexander Guthrie McCosh Prize in Philosophy Princeton University, Princeton, New Jersey
1981	Alpha Omega Alpha Honor Medical Society Baylor College of Medicine, Houston, Texas
1982	Outstanding Student in Internal Medicine Award Baylor College of Medicine, Houston, Texas
1985	Henry D McIntosh Award to the Outstanding Resident in Internal Medicine Baylor College of Medicine Affiliated Hospitals Houston, Texas
1987	Regina S Loeb Fellowship Award National Arthritis Foundation
1995	Election to the American Society for Clinical Investigation
1997	Harry Heller Award for Outstanding Contribution to the Understanding and Treatment of Patients with Familial Mediterranean Fever
1997	NIH Group Merit Award
1999	NIH Director's Award
2000	Breakthroughs in Arthritis Research Award

Metro Washington Chapter, Arthritis Foundation

- 2000 The Paul Klemperer Award
New York Academy of Medicine
- 2000 Lee C. Howley, Sr. Prize for Research in Arthritis
National Arthritis Foundation
- 2001 NIAMS Mentoring Award
- 2002 Charles W. Thomas Lecture
Medical College of Virginia, Virginia Commonwealth University
- 2003 Ann Hengel Lecture in Rheumatology
Children's Hospital of Wisconsin
- 2003 Kroc Lecture in the Genetics of Immunity and Inflammation
The Scripps Research Institute
- 2003 Top Ten Arthritis Research Advances of 2003
National Arthritis Foundation
- 2004 Mary Jane Keller Lecture
Department of Pediatrics, Yale University School of Medicine
- 2004 Woodard Colby Memorial Lecture
Children's Hospitals and Clinics, Minneapolis, Minnesota
- 2004 Dr. Morris Ziff Distinguished Lecture in Rheumatology
University of Texas Southwestern Medical School
- 2005 Election to the Association of American Physicians
- 2005 ACR Distinguished Investigator Award
American College of Rheumatology
- 2006 Edward W. Boland Lecture in Rheumatology
Mayo Foundation, Rochester, Minnesota
- 2006 Jean S. and Ephraim P. Engleman Visiting Professor of Rheumatology, University of California San Francisco
- 2006 Invited Lecturer, 51st Annual Lowe Conference on Rheumatic Diseases, University of Alabama at Birmingham
- 2007 Promotion to NIH Distinguished Investigator
- 2009 James J. Lane Lecture
University of Washington School of Medicine
- 2009 Herman Beerman Lecture
Society for Investigative Dermatology
- 2009 NIH Director's Award

2009 NIH Astute Clinician Lectureship

2010 Election to the National Academy of Sciences of the United States of America

2010 Lewis Cogen Lecture in Rheumatology
Maine Medical Center, Portland, Maine

2010 Paul M. and Dorothy E. Seebohm Lecture in Allergy
University of Iowa College of Medicine

2011 Walter Bauer Lecture in Rheumatology
Massachusetts General Hospital

2011 Sc.D., *honoris causa*
Elmezzi Graduate School of Molecular Medicine

2011 Harrison Society Visiting Professor in Medicine
Vanderbilt University School of Medicine

2012 Ernest Beutler Lecture
The Scripps Research Institute

2012 Invited Special Lecture
Fifteenth International Conference on Behçet's Disease

2012 NIH Director's Award

2012 Great Teachers Lecture, NIH Clinical Center

2012 Election to the Institute of Medicine of the National Academies

2013 Henry Kunkel Lecture
The Henry Kunkel Society

2013 Keynote Lecture
Summer Frontiers Symposium
Nijmegen Institute of Infection, Inflammation and Immunity

2013 Keynote Lecture
FASEB Science Research Conference on Autoimmunity

2014 David Trentham Lecture in Rheumatology
Beth Israel Deaconess Medical Center, Boston

2014 Keynote Lecture
Convergence Day, University of Texas Southwestern Medical School

2014 Keynote Lecture
Sixteenth Congress of the Asia Pacific League of Associations for Rheumatology

2014 Keynote Lecture
Fourteenth International Symposium on Amyloidosis

2014 Rufus Cole Lecture
The Rockefeller University

- 2014 Opening Keynote Lecture
Federation of Clinical Immunology Societies Annual Meeting
- 2015 The Nelson Lecture
University of California, Davis
- 2015 Distinguished Plenary Lecture
World Congress of Dermatology
- 2015 Thomas Waldmann Award for Excellence in Human Immunology
Foundation for Primary Immunodeficiency Diseases
- 2015 Paul Klemperer Memorial Lecture
American College of Rheumatology
- 2016 Public Lecture Award
Irish Society of Immunology
- 2016 Invited Plenary Lecture
Third Congress of the Turkish Pediatric Rheumatology Society
- 2016 Invited Plenary Lecture
57th Annual Meeting of the Australian Rheumatology Association
- 2016 Arthur and Mary Robinson Family Endowed Lecture
Human Medical Genetics and Genomics Program
University of Colorado School of Medicine
- 2016 Election as Master of the American College of Rheumatology
- 2016 NHGRI Mentorship Award
- 2017 Maimon M. Cohen Endowed Lectureship
The Harvey Institute for Human Genetics
Greater Baltimore Medical Center
- 2017 Invited Lecturer
Fourth International Conference on Primary Immunodeficiency Diseases
Bangalore, India
- 2017 Keynote Lecturer
Keystone Conference on Pattern Recognition Signaling, From Innate Immunity to
Inflammatory Disease
- 2017 Keynote Lecture
Fourth Conference of Translational Medicine on Pathogenesis and Therapy of Immune-
Mediated Diseases, Palermo, Italy
- 2017 Keynote Lecture
American College of Rheumatology Pediatric Rheumatology Symposium (PRSYM)
- 2017 Ruth L. Kirschstein Mentoring Award
National Institutes of Health

- 2018 Opening Keynote Lecture
Advanced Educational Course on Autoinflammatory Syndromes
Paediatric Rheumatology European Society
Jerusalem, Israel
- 2018 Fourth Annual Bobby Alford Distinguished Lecture
Baylor College of Medicine, Houston, Texas
- 2018 Opening Keynote Lecture
Precision Genomics Midwest 2018, Cincinnati, Ohio
- 2018 Keynote Lecture
University of Pennsylvania Rheumatology Trainee Research Day
- 2018 Invited Lecture
Chinese Clinical Immunology Society Annual Meeting, Hangzhou, China
- 2018 Distinguished Lecture Series
Life Sciences Institute, Zhejiang University, Hangzhou, China
- 2018 Samuel J. Heyman Service to America ('Sammie') Medal
2018 Federal Employee of the Year
- 2018 Keynote Lecture
Conference Celebrating 70 Years of FMF Research at the Sheba Medical Center
Tel Hashomer, Israel
- 2019 Gladys J. Fashena Lecture in Pediatrics
University of Texas Southwestern Medical Center
- 2019 Opening Lecture
Tenth International Congress on FMF and Systemic Autoinflammatory Diseases
- 2019 Keynote Lecture
Patient Organizations Meeting
Tenth International Congress on FMF and Systemic Autoinflammatory Diseases
- 2019 Career Achievement Award
U.S. Department of Health and Human Services
- 2019 Ross Prize in Molecular Medicine
The Feinstein Institute for Medical Research
- 2019 Opening Lecture
NIH-WRNMC Symposium on Autoinflammation
- 2019 Keynote Lecture
Novartis Institutes of Biomedical Research Annual Retreat
- 2019 Keynote Lecture
Japan Society of Behçet's Disease Annual Meeting
- 2020 Invited Lecturer
Annual Meeting of the Western Society of Allergy, Asthma, and Immunology

- 2020 Barbara Ansell Lecture
Pediatric Rheumatology European Society (PReS) Annual Meeting
- 2020 Keynote Lecture
Pediatric Dermatology Research Alliance (PeDRA) Annual Meeting
- 2021 American Medical Association Award for Outstanding Member of the Executive Branch
in Career Public Service, for Scientific Achievement
- 2021 Dunlop-Dottridge Lecture
Canadian Rheumatology Association Annual Meeting
- 2021 Crafoord Prize in Polyarthritis
Royal Swedish Academy of Sciences and the Crafoord Foundation
- 2021 Presidential Award
Clinical Immunology Society
- 2021 Plenary Lecture
German Children's and Youth Medicine Conference 2021
- 2021 Keynote Lecture
Lupus Research Alliance Annual Scientific Meeting
- 2021 Keynote Lecture
Fourth Inflammatory Skin Disease Summit
- 2021 NHGRI Director's Distinguished Service Award
- 2022 Nathan J. Zvaifler Lecture
University of California San Diego School of Medicine
- 2023 Great Teachers Lecture, NIH Clinical Center
- 2023 Opening Lecture
Biannual Meeting, International Society of Systemic Autoinflammatory Diseases
- 2023 Rudi Schmid Lecture
University of California San Francisco School of Medicine
- 2023 G. Burroughs Mider Lecture
National Institutes of Health
- 2024 Sudhir Gupta Oration
Indian Society for Primary Immunodeficiency Diseases
- 2024 George M. Kober Medal
Association of American Physicians
- 2024 Keynote Address
43rd European Workshop for Rheumatology Research
- 2024 Plenary Lecture
European Alliance of Organizations for Rheumatology (EULAR) Annual Congress

2024 Plenary Lecture
Peripheral Nerve Society Annual Meeting

Patents

2003 U.S. Patent 6,627,745 B1
Daniel L. Kastner et al., Pyrin Gene and Mutations Thereof, which Cause Familial Mediterranean Fever

Medical Licensure

1983 Texas, #G5242

Board Certification

1985 Diplomate of the American Board of Internal Medicine

1990 Diplomate of the Subspecialty Board in Rheumatology

Memberships

1989 American Association of Immunologists

1989 American College of Rheumatology

1994 American Society of Human Genetics

1995 American Society for Clinical Investigation

2005 Association of American Physicians

2010 National Academy of Sciences of the United States of America

2012 National Academy of Medicine (formerly Institute of Medicine)

Committees

1989-90 Biomedical Chapter Grants Subcommittee
Arthritis Foundation

1991-92 Molecular Biology/Genetics Study Section
Arthritis Foundation

1990-91 Chairman, Institutional Review Board
NIDDK/NIAMS Intramural Research Program

1992-98 Member, Institutional Review Board
NIDDK/NIAMS Intramural Research Program

1995 Chairman, Search Committee for Tenure-Track Scientist
Laboratory of Genetic Disease Research, NHGRI

1995-2010 Member, NIAMS Promotions Panel

1997-1999 Member, Scientific Review Committee

	NIH Intramural Sequencing Center (NISC)
1998-2001	Member, NIH Central Tenure Committee
2000-2005	Member, NIDDK/NIAMS Animal Care and Use Committee
2000-present	Co-Chair, NIH Genetics Interest Group
2003	Member, NIH Director's Blue Ribbon Panel on the Future of Intramural Clinical Research
2003	Member, NINDS Scientific Director's Search Committee
2004	Chair, Genomics and Proteomics Abstract Selection Committee American College of Rheumatology
2005-2006	Basic Science Chair American College of Rheumatology National Meeting
2005	Conference Chair, Fourth International Congress on Systemic Autoinflammatory Diseases, November 6–10, Bethesda, MD
2005-2010	Member, Medical Executive Committee, NIH Clinical Center
2006-2011	Member, NIH Advisory Board for Clinical Research
2007-2009	Member at Large, Scientific Advisory Council, American College of Rheumatology Research and Education Foundation
2008-2011	Chair, NIH Intramural Clinical Research Steering Committee
2008-present	Member, NIH Compensation Committee
2008-present	Member, NIH Clinical Compensation Panel
2010-present	Member, Scientific Advisory Board, Scleroderma Research Foundation
2010-2016	Member, NIAMS Title 42 Pay Committee
2011-2018	Member, NIH Intramural Clinical Research Steering Committee
2012	Chair, NIDDK Scientific Director Search Committee
2013-2016	Member, Advisory Board for Clinical Research Budget Working Group
2014-present	Member, NIH Research Facilities Advisory Committee
2015-2016	Chair, NCI Center for Cancer Research Scientific Director Search Committee
2015-2016	Chair, NIH Clinical Center Budget Working Group
2020	Member, Center for Alzheimer's and Related Dementias Director Search Committee
2020	Chair, NIDCD Scientific Director Search Committee
2020-21	Member, Chief Medical and Scientific Officer Search Committee, <i>All of Us</i> Program

2021	Chair, Center for Alzheimer's and Related Dementias Clinical Director Search Committee
2021	Chair, NEI Scientific Director Search Committee
2022	Chair, NHGRI Metabolic Medicine Branch Chief Search Committee
2022-present	Co-Director, Demystifying Medicine Course, NIH
2022-2023	Member, NIAMS Scientific Director Review Committee
2023	Chair, NIAID Laboratory Chief Search Committee
2023	Chair, NCI/CCR Genomics Branch Director Search Committee
2023	Chair, NIAMS Scientific Director Search Committee
2023	Chair, Chief Scientific Officer of the NIH Clinical Center Search Committee

Publications

1. **Kastner DL**, Rich RR, Chu L, Rich SS (1977) Regulatory mechanisms in cell-mediated immune responses. V. *H-2* homology requirement for the production of a minor locus-induced suppressor factor. *J Exp Med* **146**:1152-1157. PMID:2180814
2. Rich SS, Rich RR, **Kastner DL** (1978) Suppressor factor activity in cell-mediated immune responses. *J Reticuloendothel Soc* **24**:417-425. PMID:152354
3. **Kastner DL**, Rich RR (1978) Regulation of alloantigen-induced cytotoxic responses by concanavalin A-activated lymphoid cells: suppression by antigen elimination. *J Immunol* **121**:1732-1737. PMID:712063
4. **Kastner DL**, Rich RR (1979) *H-2*-nonrestricted cytotoxic responses to an antigen encoded telomeric to *H-2D*. *J Immunol* **122**:196-201. PMID:84017
5. **Kastner DL**, Rich RR, Shen FW (1979) *Qa-1*-associated antigens. I. Generation of *H-2*-nonrestricted cytotoxic T lymphocytes specific for determinants of the *Qa-1* region. *J Immunol* **123**:1232-1238. PMID:89166
6. **Kastner DL**, Rich RR, Chu L (1979) *Qa-1*-associated antigens. II. Evidence for functional differentiation from H-2K and H-2D antigens. *J Immunol* **123**:1239-1244. PMID:469248
7. Rich RR, Sedberry DA, **Kastner DL**, Chu L (1979) Primary *in vitro* cytotoxic response of NZB spleen cells to *Qa-1^b*-associated antigenic determinants. *J Exp Med* **150**:1555-1560. PMID:2185726
8. Nell LJ, **Kastner DL**, Rich RR (1980) *Qa-1*-associated antigens. III. Distribution of *Qa-1*-region antigens on lymphoid subpopulations. *J Immunol* **125**:2597-2603. PMID:6159414
9. Smith HR, Yaffe LJ, **Kastner DL**, Steinberg AD (1986) Evidence that Lyb 5 is a differentiation antigen in normal and *xid* mice. *J Immunol* **136**:1194-1200. PMID:3080520
10. **Kastner DL**, Steinberg AD (1988) Determinants of B cell hyper-activity in murine lupus. *Concepts in Immunopathology* **6**:22-88. PMID:3292051

11. Steinberg AD, Klinman DM, Krieg AM, Seldin MF, **Kastner DL** (1988) Approach to the use of antigen nonspecific immunosuppression in systemic lupus erythematosus and other rheumatic autoimmune diseases. *J Autoimmunity* **1**:575-592. [doi:10.1016/0896-8411\(88\)90049-2](https://doi.org/10.1016/0896-8411(88)90049-2)
12. Papa MZ, Shiloni E, Vetto JT, **Kastner DL**, McDonald HD (1989) Surgical morbidity in patients with systemic lupus erythematosus. *Am J Surg* **157**:295-298. PMID:2919734
13. **Kastner DL**, McIntyre TM, Mallett CP, Hartman AB, Steinberg AD (1989) Direct quantitative *in situ* hybridization studies of immuno-globulin V_H utilization: a comparison between unstimulated B cells from autoimmune and normal mice. *J Immunol* **143**:2761-2767. PMID:2507638
14. McIntyre TM, Holmes KL, Steinberg AD, **Kastner DL** (1991) CD5⁺ peritoneal B cells express high levels of membrane, but not secretory, C_μ mRNA. *J Immunol* **146**:3639-3645. PMID:1709199
15. Sack GH, Talbot CD Jr, McCarthy BG, Harris EL, **Kastner D**, Gruberg L, Pras M (1991) Exclusion of linkage between familial Mediterranean fever and the human serum amyloid A (SAA) gene cluster. *Hum Genet* **87**:506-508. PMID:1679035
16. Pras E, Aksentijevich I, Gruberg L, Balow JE Jr, Prosen L, Dean M, Steinberg AD, Pras M, **Kastner DL** (1992) Mapping of a gene causing familial Mediterranean fever to the short arm of chromosome 16. *N Engl J Med* **326**:1509-1513. PMID:1579134
17. Gruberg L, Aksentijevich I, Pras E, **Kastner DL**, Pras M (1992) Mapping of the familial Mediterranean fever gene to chromosome 16. *Am J Reprod Immunol* **28**:241-242. PMID:1285890
18. Aksentijevich I, Gruberg L, Pras E, Balow JE Jr, Kovo M, Gazit E, Dean M, Pras M, **Kastner DL** (1993) Evidence for linkage of the gene causing familial Mediterranean fever to chromosome 17q in non-Ashkenazi Jewish families. Second locus or type I error? *Hum Genet* **91**:527-534. PMID:8340105
19. Aksentijevich I, Pras E, Gruberg L, Shen Y, Holman K, Helling S, Prosen L, Sutherland GR, Richards RI, Ramsburg M, Dean M, Pras M, Amos CI, **Kastner DL** (1993) Refined mapping of the gene causing familial Mediterranean fever by linkage and homozygosity studies. *Am J Hum Genet* **53**:451-461. PMC1682355
20. Aksentijevich I, Pras E, Gruberg L, Shen Y, Holman K, Helling S, Prosen L, Sutherland GR, Richards RI, Dean M, Pras M, **Kastner DL** (1993) Familial Mediterranean fever in Moroccan Jews: demonstration of a founder effect by extended haplotype analysis. *Am J Hum Genet* **53**:644-651. PMC1682431
21. Pras E, Arber N, Aksentijevich I, Katz G, Schapiro JM, Prosen L, Gruberg L, Harel D, Liberman U, Weissenbach J, Pras M, **Kastner DL** (1994) Localization of a gene causing cystinuria to chromosome 2p. *Nature Genet* **6**:415-419. PMID:8054985
22. Pras E, Aksentijevich I, Levy E, Gruberg L, Prosen L, Dean M, Pras M, **Kastner DL** (1994) The gene causing familial Mediterranean fever maps to the short arm of chromosome 16 in Druze and Moslem Arab families. *Human Genet* **94**:576-577. PMID:7959700
23. McDermott M, **Kastner DL**, Holloman JD, Schmidt-Wolf G, Lundberg AS, Sinha AA, Hsu C, Cashin P, Molloy MG, Mulcahy B, O'Gara F, McConnell FI, Adams C, Khan MA, Wolfe F, Rubin LA, Clegg DO, Husebye D, Amos CI, Ward RH, McDevitt HO (1995) The role of T-cell receptor β chain genes in the susceptibility to rheumatoid arthritis. *Arthritis Rheum* **38**:91-95. PMID:7645823
24. McDermott M, Hsu C, Molloy MG, Mulcahy B, Phelan M, Shanahan F, O'Gara F, Adams C, Rubin LA, Clegg DO, Husebye D, Amos CI, Ward RH, **Kastner DL** (1995) Nonlinkage of a T-cell receptor β chain

micro-satellite (*D7S485*) to rheumatoid arthritis in multiplex families. *J Autoimmunity* **8**:131-138. PMID:7734033

25. Pras E, Raben N, Golomb E, Arber N, Aksentijevich I, Schapiro JM, Harel D, Katz G, Liberman U, Pras M, **Kastner DL** (1995) Mutations in the *SLC3A1* transporter gene in cystinuria. *Am J Hum Genet* **56**:1297-1303. PMC1801079
26. Pras M, Pras E, **Kastner D** (1995) The origin of the FMF gene. *Isr J Med Sci* **31**:503-4. PMID:7635702
27. Kindy MS, de Beer FC, Markesbery WR, Pras M, Aksentijevich I, **Kastner D**, Kyle R, Skinner M, Solomon A, Woo P (1995) Apolipo-protein E genotypes in AA and AL amyloidoses. *Amyloid* **2**:159-162.
28. Stratakis CA, Carney JA, Lin JP, Papanicolaou DA, Karl M, **Kastner DL**, Pras E, Chrousos GP (1996) Carney complex, a familial multiple neoplasia and lentiginosis syndrome: analysis of 11 kindreds and linkage to the short arm of chromosome 2. *J Clin Invest* **97**:699-705. PMC507106
29. Levy E, Shen Y, Kupelian A, Aksentijevich I, Pras E, Balow JE Jr, Linzer B, Kruglyak L, Pras M, Shohat M, Rotter JI, Fischel-Ghodsian N, Richards RI, **Kastner DL** (1996) Linkage disequilibrium mapping places the gene causing familial Mediterranean fever close to *D16S246*. *Am J Hum Genet* **58**:523-534. PMC1914760
30. Pras E, Sood R, Aksentijevich I, Chen X, **Kastner DL** (1996) Genomic organization of *SLC3A1*, a transporter gene mutated in cystinuria. *Genomics* **36**:163-167. PMID:8812428
31. McDermott MF, Schmidt-Wolf G, Sinha AA, Koo M, Porter MA, Briant L, Cambon-Thomsen A, Maclaren NK, Fiske D, Bertera S, Trucco M, Amos CI, McDevitt HO, **Kastner DL** (1996) No linkage or association of telomeric and centromeric T-cell receptor β -chain markers with susceptibility to type 1 insulin-dependent diabetes in *HLA-DR4* multiplex families. *Eur J Immunogenet* **23**: 361-370. PMID:8909943
32. Pras E, Schumacher HR, **Kastner DL**, Wilder RL (1996) Lack of evidence of mycobacteria in synovial tissue from patients with rheumatoid arthritis. *Arthritis Rheum* **39**: 2080-2081. PMID:8961916
33. Wartenfeld R, Golomb E, Katz G, Bale SJ, Goldman B, Pras M, **Kastner DL**, Pras E (1997) Molecular analysis of cystinuria in Libyan Jews: exclusion of the *SLC3A1* gene and mapping of a new locus on chromosome 19q. *Am J Hum Genet* **60**: 617-624. PMC1712472
34. Sood R, Blake T, Aksentijevich I, Wood G, Chen X, Gardner D, Shelton DA, Mangelsdorf M, Orsborn A, Pras E, Balow JE Jr, Centola M, Deng Z, Zaks N, Chen X, Richards N, Fischel-Ghodsian N, Rotter JI, Pras M, Shohat M, Deaven LL, Gumucio DL, Callen DF, Richards RI, Collins FS, Liu P, **Kastner DL**, Doggett NA (1997) Construction of a 1-Mb restriction-mapped cosmid contig containing the genetic interval for the familial Mediterranean fever locus (*MEFV*) on chromosome 16p13.3. *Genomics* **42**:83-95. PMID:9177779
35. Balow JE Jr, Shelton DA, Orsborn A, Mangelsdorf M, Aksentijevich I, Blake T, Sood R, Gardner D, Liu R, Pras E, Levy EN, Centola M, Deng Z, Zaks N, Wood G, Chen X, Richards N, Shohat M, Livneh A, Pras M, Doggett NA, Collins FS, Liu PP, Rotter JI, Fischel-Ghodsian N, Gumucio D, Richards RI, **Kastner DL** (1997) A high-resolution genetic map of the familial Mediterranean fever candidate region allows identification of haplotype-sharing among ethnic groups. *Genomics* **44**:280-291. PMID:9325049
36. Livneh A, Drenth JP, Klasen IS, Langevitz P, George J, Shelton DA, Gumucio DL, Pras E, **Kastner DL**, Pras M, van der Meer JW (1997) Familial Mediterranean fever and hyperimmunoglobulinemia D syndrome: two diseases with distinct clinical, serologic, and genetic features. *J Rheum* **24**:1558-1563. PMID:9263151

37. International FMF Consortium (**Kastner DL**, corresponding author) (1997) Ancient missense mutations in a new member of the *RoRet* gene family are likely to cause familial Mediterranean fever. *Cell* **90**:797-807. PMID:9288758
38. Pras E, Golomb E, Drake C, Aksentijevich I, Katz G, **Kastner DL** (1998) A splicing mutation (891+4A->G) in *SLC3A1* leads to exon 4 skipping and causes cystinuria in a Moslem Arab family *Human Mutation* Supplement 1:S28-S30. PMID:9452031
39. Pras E, Livneh A, Balow JE Jr, Pras E, **Kastner DL**, Pras M, Langevitz P (1998) Clinical differences between North African and Iraqi Jews with familial Mediterranean fever. *Am J Med Genet* **75**:216-219. PMID:9450890
40. **Kastner DL** (1998) Familial Mediterranean fever: the genetics of inflammation. *Hospital Practice* **33**:131-158. PMID:9562837
41. Samuels J, Aksentijevich I, Torosyan Y, Centola M, Deng Z, Sood R, **Kastner DL** (1998) Familial Mediterranean fever at the millennium: clinical spectrum, ancient mutations, and a survey of 100 American referrals to the National Institutes of Health. *Medicine (Baltimore)* **77**:268-297. PMID:9715731
42. Hirsch R, Lin J-P, Scott WW Jr, Ma LD, Pillemer SR, **Kastner DL**, Jacobson LTH, Bloch DA, Knowler WC, Bennett PH, Bale SJ (1998) Rheumatoid arthritis in the Pima Indians: the intersection of epidemiologic, demographic, and genealogic data. *Arthritis Rheum* **41**:1464-1469. PMID:9704646
43. Centola M, Aksentijevich I, **Kastner DL** (1998) The hereditary periodic fever syndromes: molecular analysis of a new family of inflammatory diseases. *Hum Mol Genet* **7**:1581-1588. PMID:9735379
44. Eisenberg S, Aksentijevich I, Deng Z, **Kastner DL**, Matzner Y (1998) Diagnosis of familial Mediterranean fever by a molecular genetic method. *Ann Intern Med* **129**:539-542. PMID:9758573
45. Deng, Z, Centola M, Chen X, Sood R, Vedula A, Fischel-Ghodsian N, **Kastner DL** (1998) Identification of two *Krüppel*-related zinc finger genes (*ZNF200*, *ZNF210*) from human chromosome 16p13.3. *Genomics* **53**:97-103. PMID:9787081
46. Centola M, Chen X, Sood R, Deng Z, Aksentijevich I, Blake T, Ricke DO, Chen X, Wood G, Zaks N, Richards N, Krizman D, Mansfield E, Apostolou S, Liu J, Shafran N, Vedula A, Hamon M, Cercek A, Kahan T, Gumucio D, Callen DF, Richards RI, Moyzis RK, Doggett NA, Collins FS, Liu PP, Fischel-Ghodsian N, **Kastner DL** (1998) Construction of an ~700 kb transcript map around the familial Mediterranean fever locus on human chromosome 16p13.3. *Genome Res* **8**:1172-1191. PMC310791
47. Shohat M, Lotan R, Magal N, Danon Y, Ogur G, Tokguz G, Schlezinger M, Schwabe A, Halpern G, Fischel-Ghodsian N, **Kastner D**, Shohat T, Rotter JI (1998) Amyloidosis in familial Mediterranean fever is associated with a specific ancestral haplotype in the *MEFV* locus. *Mol Genet Metab* **65**:197-202. PMID:9851884
48. Pras E, Kochba I, Lubetzky A, Pras M, Sidi Y, **Kastner DL** (1998) Biochemical and clinical studies of Libyan Jewish cystinuria patients and their relatives. *Am J Med Genet* **80**:173-176. PMID:9805137
49. Chen X, Hamon M, Deng Z, Centola M, Sood R, Taylor K, **Kastner DL**, Fischel-Ghodsian N (1999) Identification and characterization of a zinc finger gene (*ZNF213*) from 16p13.3. *Biochim Biophys Acta* **1444**:218-230. PMID:10023065
50. Aksentijevich I, Torosyan Y, Samuels J, Centola M, Pras E, Chae JJ, Oddoux C, Wood G, Azzaro MP, Palumbo G, Giustolisi R, Pras M, Ostrer H, **Kastner DL** (1999) Mutation and haplotype studies of

familial Mediterranean fever reveal new ancestral relationships and evidence for a high carrier frequency with reduced penetrance in the Ashkenazi Jewish population. *Am J Hum Genet* **64**:949-962. PMC1377819

51. McDermott MF, Aksentijevich I, Galon J, McDermott EM, Ogunkolade BW, Centola M, Mansfield E, Gadina M, Karenko L, Pettersson T, McCarthy J, Frucht DM, Aringer M, Torosyan Y, Teppo A-M, Wilson M, Karaarslan HM, Wan Y, Todd I, Wood G, Schlimgen R, Kumarajeewa TR, Cooper SM, Vella JP, Amos CI, Mulley J, Quane KA, Molloy MG, Ranki A, Powell RJ, Hitman GA, O'Shea JJ, **Kastner DL** (1999) Germline mutations in the extracellular domains of the 55 kDa TNF receptor, TNFR1, define a family of dominantly inherited autoinflammatory syndromes. *Cell* **97**:133-144. PMID:10199409
52. Livneh A, Langevitz P, Shinar Y, Zaks N, **Kastner DL**, Pras M, Pras E (1999) *MEFV* mutation analysis in patients suffering from familial Mediterranean fever. *Amyloid* **6**:1-6. PMID:10211405
53. Shohat M, Magal N, Shohat T, Chen X, Dagan T, Mimouni A, Danon Y, Lotan R, Ogur G, Sirin A, Schlezinger M, Halpern GJ, Schwabe A, **Kastner D**, Rotter JI, Fischel-Ghodsian N (1999) Phenotype-genotype correlation in familial Mediterranean fever: evidence for an association between Met694Val and amyloidosis. *Eur J Hum Genet* **7**:287-292. PMID:10234504
54. Chae JJ, Kim SH, Kim UK, Han KH, Kim HS, **Kastner DL**, Namkoong Y, Park YB, Lee CC (1999) Three novel deletion mutations of the LDL receptor gene in Korean patients with familial hypercholesterolemia. *Clin Genet* **55**:325-331. PMID:10422802
55. Soares M, Buxbaum J, Sirugo G, Coelho T, Sousa A, **Kastner D**, Saraiva MJ (1999) Genetic anticipation in Portuguese kindreds with familial amyloidotic polyneuropathy is unlikely to be caused by triplet repeat expansions. *Hum Genet* **104**:480-485. PMID:10453736
56. Lin JP, Hirsch R, Jacobsson LT, Scott WW, Ma LD, Pillemer SR, Knowler WC, **Kastner DL**, Bale SJ (1999) Genealogy construction in a historically isolated population: application to genetic studies of rheumatoid arthritis in the Pima Indian. *Genet Med* **1**:187-193. PMID:11256671
57. International Cystinuria Consortium (**Kastner DL**, seventh of eight authors in Group B) (1999) Non-Type I cystinuria caused by mutations in *SLC7A9*, coding for a subunit (b^{0,+} AT) of rBAT. *Nature Genet* **23**:52-57. PMID:10471498
58. Pras E, Kreiss Y, Frishberg Y, Prosen L, Aksentijevich I, **Kastner DL** (1999) Refined mapping of the CSNU3 gene to a 1.8-Mb region on chromosome 19q13.1 using historical recombinants in Libyan Jewish cystinuria patients. *Genomics* **60**:248-250. PMID:10486219
59. Oddoux C, Guillen-Navarro E, Ditivoli C, Dicave E, Cilio MR, Clayton CM, Nelson H, Sarafoglou K, McCain N, Peretz H, Seligsohn U, Luzzatto L, Nafa K, Nardi M, Karpatkin M, Aksentijevich I, **Kastner D**, Axelrod F, Ostrer H (1999) Mendelian diseases among Roman Jews: implications for the origins of disease alleles. *J Clin Endocrinol Metab* **84**:4405-4409. PMID:10599695
60. Centola M, Wood G, Frucht D, Galon J, Aringer M, Farrell C, Kingma DW, Horwitz M, Mansfield E, Holland SM, O'Shea JJ, Rosenberg HF, Malech HL, **Kastner DL** (2000) The gene for familial Mediterranean fever, *MEFV*, is expressed in early leukocyte development and in response to inflammatory mediators. *Blood* **95**:3223-3231. PMID:10807793
61. Chae JJ, Centola M, Aksentijevich I, Dutra A, Tran M, Nagaraju K, Kingma DW, Liu PP, **Kastner DL** (2000) Isolation, genomic organization, and expression analysis of the mouse and rat homologs of *MEFV*, the gene for familial Mediterranean fever. *Mamm Genome* **11**:428-435. PMID:10818206
62. Galon J, Aksentijevich I, McDermott MF, O'Shea JJ, **Kastner DL** (2000) *TNFRSF1A* mutations and autoinflammatory syndromes. *Curr Opin Immunol*, **12**:479-486. PMID:10899034

63. Shinar Y, Livneh A, Langevitz P, Zaks N, Aksentijevich I, Koziol DE, **Kastner DL**, Pras M, Pras E (2000) Genotype-phenotype assessment of common genotypes among patients with familial Mediterranean fever. *J Rheumatol* **27**:1703-1707. PMID:10914855
64. Hooper JD, Bowen N, Marshall H, Cullen LM, Sood R, Daniels R, Stuttgart MA, Normyle JF, Higgs DR, **Kastner DL**, Ogbourne SM, Pera MF, Jazwinska EC, Antalis TM (2000) Localization, expression and genomic structure of the gene encoding the human serine protease testisin. *Biochim Biophys Acta* **1492**:63-71. PMID:11004480
65. Toro JR, Aksentijevich I, Hull K, Dean J, **Kastner DL** (2000) Tumor necrosis factor receptor-associated periodic syndrome: a novel entity with cutaneous manifestations. *Arch Dermatol* **136**:1487-1494. PMID:11115159
66. Aganna E, Aksentijevich I, Hitman GA, **Kastner DL**, Hoepelman AIM, Posma FD, Zweers EJK, McDermott MF (2001) Tumor necrosis factor receptor-associated periodic syndrome (TRAPS) in a Dutch family: evidence for a *TNFRSF1A* mutation with reduced penetrance. *Eur J Hum Genet* **9**:63-66. PMID:11175303
67. Schaner P, Richards N, Wadhwa A, Aksentijevich I, **Kastner D**, Tucker P, Gumucio D (2001) Episodic evolution of pyrin in primates: human mutations recapitulate ancestral amino acid states. *Nature Genet* **27**:318-321. PMID:11242116
68. Jawaheer D, Seldin MF, Amos CI, Chen WV, Shigeta R, Monteiro J, Kern M, Criswell LA, Albani S, Nelson JL, Clegg DO, Pope R, Schroeder HW Jr, Bridges SL Jr, Pisetsky DS, Ward R, **Kastner DL**, Wilder RL, Pincus T, Callahan LF, Flemming D, Wener MH, Gregersen PK (2001) A genome-wide screen in multiplex rheumatoid arthritis families suggests genetic overlap with other autoimmune diseases. *Am J Hum Genet* **68**:927-936. PMC1275647
69. Livneh A, Aksentijevich I, Langevitz P, Torosyan Y, Galon-Shoham N, Shinar Y, Pras E, Zaks N, Padeh S, **Kastner DL**, Pras M (2001) A single mutated MEFV allele in Israeli patients suffering from familial Mediterranean fever and Behcet's disease. *Eur J Hum Genet* **9**:191-196. PMID:11313758
70. Njoroge JM, Mitchell LB, Centola M, **Kastner D**, Raffeld M, Miller JL (2001) Characterization of viable autofluorescent macrophages among cultured peripheral blood mononuclear cells. *Cytometry* **44**:38-44. PMID:11309807
71. Mansfield E, Chae JJ, Komarow HD, Brotz T, Frucht DM, Aksentijevich I, **Kastner DL** (2001) The FMF protein, pyrin, associates with microtubules and actin filaments. *Blood* **98**:851-859. PMID:11468188
72. Aksentijevich I, Galon J, Soares M, Mansfield E, Hull K, Oh H-H, Goldbach-Mansky R, Dean J, Athreya B, Reginato AJ, Henrickson M, Pons-Estel B, **Kastner DL** (2001) The TNF receptor-associated periodic syndrome (TRAPS): new mutations in *TNFRSF1A*, ancestral origins, genotype-phenotype studies, and evidence for further genetic heterogeneity of periodic fevers. *Am J Hum Genet* **69**:301-314. PMC1235304
73. Lahat H, Eldar M, Levy-Nissenbaum E, Bahan T, Friedman E, Khoury A, Lorber A, **Kastner DL**, Goldman B, Pras E (2001) Autosomal recessive catecholamine induced polymorphic tachycardia: clinical features and assignment of the disease gene to chromosome 1p 13-21. *Circulation* **103**:2822-2827. PMID:11401939
74. Pras E, Pras E, Bakhan T, Levy-Nissenbaum E, Lahat H, Assisa EI, Gorzozzi HJ, **Kastner DL**, Goldman B, Frydman M (2001) A gene causing autosomal recessive cataract maps to the short arm of chromosome 3. *Isr Med Assoc J* **3**:559-562. PMID:11519376

75. **Kastner D** (2001) Pathogenesis and diagnostic tools in the hereditary periodic fevers. *Riv Ital Pediatr* **27**:618-621.
76. **Kastner DL**, O'Shea JJ (2001) A fever gene comes in from the cold. *Nature Genet* **29**:241-242. PMID:11687785
77. Pras E, Aksentijevich I, Shinar Y, **Kastner DL**, Achiron A (2001) Lack of evidence for an association between two genetic polymorphisms in the tumor necrosis factor receptor 1 gene and multiple sclerosis in Ashkenazi Jews. *Eur Neurol* **46**:153-155. PMID:11598334
78. Shinar Y, Ben-Zeev B, Brand N, Lahat H, Gross-Zur V, MacGregor D, Bahan T, **Kastner DL**, Pras E (2002) A common ancestral haplotype in carrier chromosomes from different ethnic backgrounds in vacuolating megalencephalic leukoencephalopathy with subcortical cysts. *J Med Genet* **39**:54-57. PMC1734951
79. Harley JB, Trent J, **Kastner DL** (2002) American College of Rheumatology Basic Research Conference: Genetics and genomics in rheumatic disease. *Arthritis Rheum* **47**:93-98. PMID:11932884
80. Hull KM, **Kastner DL**, Balow JE. Hereditary periodic fever [letter]. *N Engl J Med* **346**:1415. PMID:11986423
81. Hull KM, Wong K, Wood GM, Chu W-S, **Kastner DL** (2002) Monocytic fasciitis: a new clinical feature of TNF receptor dysfunction. *Arthritis Rheum* **46**:2189-2194. PMID:12209524
82. Hull KM, Drewe E, Aksentijevich I, Singh H, McDermott EM, Dean J, Powell RJ, **Kastner DL** (2002) The TNF receptor-associated periodic syndrome: emerging concepts of an autoinflammatory disorder. *Medicine (Baltimore)* **81**:349-368. PMID:12352631
83. Ozen S, Balci B, Ozkara S, Ozcan A, Yilmaz E, Besbas N, Ozguc M, **Kastner DL**, Bakkaloglu A (2002) Is there a heterozygote advantage for familial Mediterranean fever carriers against tuberculosis infections: speculations remain. *Clin Exp Rheumatol* **20 (Suppl 26)**: S57-58. PMID:12371639
84. Aksentijevich I, Nowak M, Mallah M, Chae JJ, Watford WT, Hoffman SR, Stein L, Russo R, Goldsmith D, Dent P, Rosenberg HF, Austin F, Remmers EF, Balow JE Jr, Rosenzweig S, Komarow H, Shoham NG, Wood G, Jones J, Mangra N, Carrero H, Adams BS, Moore TL, Schikler K, Hoffman H, Lovell DJ, Lipnick R, Barron K, O'Shea JJ, **Kastner DL**, Goldbach-Mansky R (2002) De novo *CIAS1* mutations, cytokine activation, and evidence for genetic heterogeneity in patients with neonatal-onset multisystem inflammatory disease (NOMID): a new member of the expanding family of pyrin-associated autoinflammatory diseases. *Arthritis Rheum* **46**:3340-3348. PMC4556432
85. Hull KM, Shoham N, Chae JJ, Aksentijevich I, **Kastner DL** (2003) The expanding spectrum of systemic autoinflammatory disorders and their rheumatic manifestations. *Curr Opin Rheumatol* **15**:61-69. PMID:12496512
86. Chae JJ, Komarow H, Cheng J, Wood G, Raben N, Liu PP, **Kastner DL** (2003) Targeted disruption of pyrin, the FMF protein, causes heightened sensitivity to endotoxin and a defect in macrophage apoptosis. *Mol Cell* **11**:591-604. PMID:12667444
87. Jawaheer D, Seldin MF, Amos CI, Chen WV, Shigeta R, Etzel C, Damle A, Xiao X, Chen D, Lum RF, Monteiro J, Kern M, Criswell L, Albani S, Nelson JL, Clegg DO, Pope R, Schroeder HW Jr, Bridges SL Jr, Pisetsky DS, Ward R, **Kastner DL**, Wilder RL, Pincus T, Callahan LF, Fleming D, Wener MH, Gregersen PK, for the North American Rheumatoid Arthritis Consortium (2003) Screening the genome for rheumatoid arthritis susceptibility genes: a replication study and combined analysis of 512 multicase families. *Arthritis Rheum* **48**: 906-916. PMID:12687532

88. Soares ML, Centola M, Chae JJ, Saraiva MJ, **Kastner DL** (2003) Human transthyretin intronic open reading frames are not independently expressed in vivo or part of functional transcripts. *Biochem Biophys Acta* **1626**:65-74. PMID:12697331
89. Aringer M, Hofmann SR, Frucht DM, Chen M, Centola M, Morinobu A, Visconti R, **Kastner DL**, Smolen JS, O'Shea JJ (2003) Characterization and analysis of the proximal *Janus Kinase 3* promoter. *J Immunol* **170**:6057-6064. PMID:12794134
90. Touitou I, Ben-Chetrit E, Gershoni-Baruch R, Grateau G, **Kastner DL**, Kone-Paut I, Livneh A, Manna R, Mansour I, Ozdogan H, Ozen S, Sarkisian T, Tunca M, Yalcinkaya F (2003) Allogeneic bone marrow transplantation: not a treatment yet for familial Mediterranean fever. *Blood* **102**:409. PMID:12814918
91. Takada K, Aksentijevich I, Mahadevan V, Dean JA, Kelley RI, **Kastner DL** (2003) Favorable preliminary experience with etanercept in two patients with hyperimmunoglobulinemia D and periodic fever syndrome. *Arthritis Rheum* **48**:2645-2651. PMID:13130485
92. Shoham NG, Centola M, Mansfield E, Hull KM, Wood G, Wise CA, **Kastner DL** (2003) Pyrin binds the PSTPIP1/CD2BP1 protein, defining PAPA syndrome and familial Mediterranean fever as disorders in the same pathway. *Proc Natl Acad Sci USA* **100**:13501-13506. PMC263843
93. Soares ML, Coelho T, Sousa A, Holmgren G, Saraiva MJ, **Kastner DL**, Buxbaum JN (2004) Haplotypes and DNA sequence variation within and surrounding the transthyretin gene: genotype-phenotype correlations in familial amyloid polyneuropathy (V30M) in Portugal and Sweden. *Eur J Hum Genet* **12**:225-237. PMID:14673473
94. Begovich AB, Carlton VEH, Honigberg LA, Schrodi SJ, Chokkalingam AP, Alexander HC, Ardlie KG, Huang Q, Smith AM, Spoerke JM, Conn MT, Chang M, Chang S-YP, Saiki RK, Catanese JJ, Leong DU, Garcia VE, McAllister LB, Jeffery DA, Lee AT, Batliwalla F, Remmers E, Criswell LA, Seldin MF, **Kastner DL**, Amos CI, Sninsky JJ, Gregersen PK (2004) A missense single-nucleotide polymorphism in a gene encoding a protein tyrosine phosphatase (*PTPN22*) is associated with rheumatoid arthritis. *Am J Hum Genet* **75**:330-337. PMC1216068
95. Diaz A, Hu C, **Kastner DL**, Schaner P, Reginato AM, Richards N, Gumucio DL (2004) Expression of multiple alternatively spliced *MEFV* transcripts in human synovial fibroblasts induced by lipopolysaccharide. *Arthritis Rheum* **50**:3679-3689. PMID:15529356
96. Stojanov S, **Kastner DL** (2005) Familial autoinflammatory diseases: update on genetics, pathogenesis, and treatment. *Curr Opin Rheumatol* **17**:586-599. PMID:16093838
97. Addo A, Le J, Aksentijevich I, Balow J Jr, Lee A, Gregersen PK, **Kastner DL**, Remmers EF (2005) Analysis of *CARD15/NOD2* haplotypes fails to identify common variants associated with rheumatoid arthritis susceptibility. *Scand J Rheumatol* **34**:198-203. PMID:16134725
98. Carlton VE, Hu X, Chokkalingam AP, Schrodi SJ, Brandon R, Alexander HC, Chang M, Catanese JJ, Leong DU, Ardlie KG, **Kastner DL**, Seldin MF, Criswell LA, Gregersen PK, Beasley E, Thomson G, Amos CI, Begovich AB (2005) *PTPN22* genetic variation: evidence for multiple variants associated with rheumatoid arthritis. *Am J Hum Genet* **77**:567-581. PMC1275606
99. Lokuta MA, Cooper KM, Aksentijevich I, **Kastner DL**, Huttenlocher A (2005) Abnormal neutrophil chemotaxis in a patient with neonatal onset multisystem inflammatory disease/Muckle-Wells syndrome. *Ann Allergy Asthma Immunol* **95**:394-399. PMID:16279571
100. **Kastner DL** (2005) Hereditary periodic fever syndromes. *Hematology (Am Soc Hematol Educ Program)* 2005:74-81. PMID:16304362

101. Irogoyen P, Lee AT, Wener MH, Li W, Kern M, Batliwalla F, Lum RF, Massarotti E, Weisman M, Bombardier C, Remmers EF, **Kastner DL**, Seldin MF, Criswell LA, Gregersen PK (2005) Regulation of anti-cyclic citrullinated peptide antibodies in rheumatoid arthritis: contrasting effects of HLA-DR3 and the shared epitope alleles. *Arthritis Rheum* **52**:3813-3818. PMID:16320316
102. Plenge RM, Padukov L, Remmers E, Purcell S, Lee AT, Wolfe F, **Kastner DL**, Alfredsson L, Altschuler D, Gregersen PK, Klareskog L, Rioux JD (2005) Replication of putative candidate gene associations with rheumatoid arthritis in over 4,000 clinical samples from North America and Sweden: association of susceptibility with *PTPN22*, *CTLA4*, and *PADI4*. *Am J Hum Genet* **77**:1044-1060. PMC1285162
103. Jacob SE, Cowen EW, Goldbach-Mansky R, **Kastner D**, Turner ML (2006) A recurrent rash with fever and arthropathy. *J Am Acad Dermatol* **54**:318-321. PMID:16443062
104. Ting JP, **Kastner DL**, Hoffman HM (2006) CATERPILLERS, pyrin, and hereditary immunologic disorders. *Nat Rev Immunol* **6**:183-195. PMID:16498449
105. Hu X, Chang M, Saiki RK, Cargill MA, Begovich AB, Ardlie KG, Criswell LA, Seldin MF, Amos CI, Gregersen PK, **Kastner DL**, Remmers EF (2006) The functional -169T→C single-nucleotide polymorphism in FCRL3 is not associated with rheumatoid arthritis in white North Americans. *Arthritis Rheum* **54**:1022-1025. PMID:16508985
106. Ozen S, Hoffman HM, Frenkel J, **Kastner D** (2006) Familial Mediterranean fever and beyond: a new horizon. Fourth International Congress on the Systemic Autoinflammatory Diseases held in Bethesda, USA, 6–10 November 2006. *Ann Rheum Dis* **65**:961-964. PMC1798229
107. Amos CI, Chen WV, Lee A, Li W, Kern M, Lundsten R, Batliwalla F, Wener F, Remmers E, **Kastner D**, Criswell LA, Seldin MF, Gregersen PK (2006) High-density SNP analysis of 642 Caucasian families with rheumatoid arthritis identified two new linkage regions on 11p12 and 2q33. *Genes Immun* **7**:277-286. PMID:16691188
108. Lobito AA, Kimberley FC, Muppidi JR, Komarow H, Jackson AJ, Hull KM, **Kastner DL**, Sreaton GR, Siegel RM (2006) Abnormal disulfide-linked oligomerization results in ER retention and altered signaling by TNFR1 mutants in the TNFR1 associated periodic fever syndrome (TRAPS). *Blood* **108**:1320–1327. PMC1895878
109. Chae JJ, Wood G, Masters SL, Richard K, Park G, Smith BJ, **Kastner DL** (2006) The B30.2 domain of pyrin, the familial Mediterranean fever protein, interacts directly with caspase-1 to modulate IL-1 β production. *Proc Natl Acad Sci USA* **103**:9982–9987. PMC1479864
110. Aksentijevich I, Remmers EF, Goldbach-Mansky R, **Kastner DL** (2006) Mutational analysis in neonatal-onset multisystem inflammatory disease: comment on the articles by Frenkel et al and Saito et al. *Arthritis Rheum* **54**:2703–2704. PMID:16871551
111. Goldbach-Mansky R, Dailey NJ, Canna SW, Gelabert A, Jones J, Rubin BI, Kim HJ, Brewer C, Zelewski C, Wiggs E, Hill S, Turner ML, Karp BI, Aksentijevich I, Pucino F, Penzak S, Haverkamp MH, Stein L, Adams BS, Moore TL, Fuhlbrigge RC, Shaham B, Jarvis JN, O'Neill K, Vehe RK, Beitz LO, Gardner G, Hannan WP, Warren RW, Horn W, Cole JL, Paul SM, Hawkins PN, Pham TH, Snyder C, Wesley RA, Hoffman SC, Holland SM, Butman JA, **Kastner DL** (2006) Neonatal onset multisystem inflammatory disease responsive to IL-1 β inhibition. *N Engl J Med* **355**:581–592. PMC4178954
112. Masters SL, Lobito AA, Chae JJ, **Kastner DL** (2006) Recent advances in the molecular pathogenesis of hereditary recurrent fevers. *Curr Opin Allergy Clin Immunol* **6**:428–433. PMID:17088647
113. Aksentijevich I, Putnam CD, Remmers EF, Mueller JL, Kolodner RD, Moak Z, Chuang M, Austin F, Goldbach-Mansky R, Hoffman HM, **Kastner DL** (2007) The clinical continuum of cryopyrinopathies:

novel *CIAS1* mutations in the North American cohort of patients and a new cryopyrin model. *Arthritis Rheum* **56**:1273–1285. PMC4321998

114. Chitkara P, Stojanov S, **Kastner DL** (2007) The hereditary autoinflammatory syndromes. *Pediatr Infect Dis J* **26**:353–354. PMID:17414402
115. Touitou I, Sarkisian T, Medlej-Hashim M, Tunca M, Livneh A, Cattan D, Yalcinkaya F, Ozen S, Majeed H, Ozdogan H, **Kastner D**, Booth D, Ben-Chétrit E, Pugnère D, Michelon C, Séguret F, Gershoni-Baruch R, for the International Study Group for Phenotype-Genotype Correlation in FMF. (2007) Country as the primary risk factor for renal amyloidosis in familial Mediterranean fever (FMF). *Arthritis Rheum* **56**:1706–1712. PMID:17469185
116. Goldbach-Mansky R, Pucino F, **Kastner DL** (2007) Treatment of patients with neonatal-onset multisystem inflammatory disease (NOMID): comment on the article by Masubara et al. *Arthritis Rheum* **56**:2099–2101. PMID:17530657
117. Wright DG, **Kastner DL**, Pollen GB, and Members of the Systemic Amyloidosis Advisory Group (2007) Challenges and opportunities for systemic amyloidosis research: summary of an advisory workshop sponsored by the NIH Office of Rare Diseases. *Amyloid* **14**:103–112. PMID:17577683
118. Remmers EF, Plenge RM, Lee AT, Graham RR, Hom G, Behrens TW, de Bakker PIW, Le JM, Lee H-S, Batliwalla F, Li W, Masters SL, Booty MG, Carulli JP, Padyukov L, Alfredsson L, Klareskog L, Chen WV, Amos CI, Criswell LA, Seldin MF, **Kastner DL**, Gregersen PK (2007) *STAT4* and the risk of rheumatoid arthritis and systemic lupus erythematosus. *N Engl J Med* **357**:977–986. PMC2630215
119. Plenge RM, Seielstad M, Padyukov L, Lee AT, Remmers EF, Ding B, Liew A, Khalili H, Chandrasekaran A, Davies LRL, Li W, Tan AKS, Bonnard C, Ong RTH, Thalamuthu A, Pettersson S, Liu C, Tian C, Chen WV, Carulli JP, Beckman EM, Altshuler D, Alfredsson L, Criswell LA, Amos CI, Seldin MF, **Kastner DL**, Klareskog L, Gregersen PK (2007) *TRAF1–C5* as a risk locus for rheumatoid arthritis — a genomewide study. *N Engl J Med* **357**:1199–1209. PMC2636867
120. Lee H-S, Remmers EF, Le JM, **Kastner DL**, Bae S-C, Gregersen PK (2007) Association of *STAT4* with rheumatoid arthritis in the Korean population. *Mol Med* **13**:455–460. PMC2014726
121. Ryan JG, de Koning HD, Beck LA, Booty MG, **Kastner DL**, Simon A (2007) IL-1 blockade in Schnitzler syndrome: *ex vivo* findings correlate with clinical remission. *J Allergy Clin Immunol* **121**:260–262. PMID:17936890
122. Amos CI, Chen WV, Remmers E, Siminovitch KA, Seldin MF, Criswell LA, Lee AT, John S, Shephard ND, Worthington J, Cornelis F, Plenge RM, Begovitch AB, Dyer TD, **Kastner DL**, Gregersen PK (2007) Data for Genetic Analysis Workshop (GAW) 15 Problem 2, genetic causes of rheumatoid arthritis and associated traits. *BMC Proc.* 2007 **1 Suppl 1**:S3. PMC2367518
123. Korman BD, Alba MI, Le JM, Alevizos I, Smith JA, Nikolov NP, **Kastner DL**, Remmers EF, Illei GG (2008) Variant form of *STAT4* is associated with primary Sjögren’s syndrome. *Genes Immun* **9**:267–270. PMID:18273036
124. Lee H-S, Lee AT, Criswell LA, Seldin MF, Amos CI, Carulli JP, Navarrete C, Remmers EF, **Kastner DL**, Plenge RM, LI W, Gregersen PK (2008) Several regions of the major histocompatibility complex confer risk for anti-CCP-antibody positive rheumatoid arthritis, independent of the *DRB1* locus. *Mol Med* **14**:293–300. PMC2255558
125. Taylor KE, Remmers EF, Lee AT, Ortmann WA, Plenge RM, Tian C, Chung SA, Nititham J, Hom G, Kao AH, Demirci FY, Kamboh MI, Petri M, Manzi S, **Kastner DL**, Seldin MF, Gregersen PK, Behrens

- TW, Criswell LA (2008) Specificity of the *STAT4* genetic association for severe manifestations of systemic lupus erythematosus. *PLoS Genet* **4**:e1000084. PMC2377340
126. Chang M, Saiki RK, Cantanes JJ, Lew D, van der Helm-van Mil AH, Toes RE, Huizinga TW, Ardlie KG, Criswell LA, Seldin MF, Amos CI, **Kastner DL**, Gregersen PK, Schrodi SJ, Begovitch AB (2008) The inflammatory disease-associated variants in IL12B and IL23R are not associated with rheumatoid arthritis. *Arthritis Rheum* **58**:1877–1881. PMC2996314
 127. Chang M, Rowland CM, Garcia VE, Schrodi SJ, Catanese JJ, van der Helm-van Mil AH, Ardlie KG, Amos CI, Criswell LA, **Kastner DL**, Gregersen PK, Kurreeman FA, Toes RE, Huizinga TW, Seldin MF, Begovitch AB (2008) A large-scale rheumatoid arthritis genetic study identifies association at chromosome 9q33.2. *PLoS Genet* **4**:e1000107. PMC2481282
 128. Chae JJ, Wood G, Richard K, Jaffe H, Colburn NT, Masters SL, Gumucio DL, Shoham NG, **Kastner DL** (2008) The familial Mediterranean fever protein, pyrin, is cleaved by caspase-1 and activates NF- κ B through its N-terminal fragment. *Blood* **112**:1794–1803. PMC2518886
 129. Goldbach-Mansky R, Shroff S, Wilson M, Snyder C, Barham B, Pham TH, Wesley R, Papadopoulos J, Weinstein S, Mellis S, **Kastner DL** (2008) A pilot study to evaluate the safety and efficacy of the long-acting IL-1 inhibitor, riloncept, in patients with familial cold autoinflammatory syndrome (FCAS). *Arthritis Rheum* **58**:2432–2442. PMC2875198
 130. Korman BD, **Kastner DL**, Gregersen PK, Remmers EF, (2008) STAT4: genetics, mechanisms, and implications for autoimmunity. *Curr Allergy Asthma Rep* **8**:398–403. PMC2562257
 131. Ryan JG, **Kastner DL** (2008) Fevers, genes, and innate immunity. *Current Topics in Microbiology and Immunology* **321**:169–184. PMID:18727492
 132. Raychaudhuri S, Remmers EF, Lee AT, Hackett R, Guiducci C, Burt NP, Gianniny L, Korman BD, Padyukov L, Kurreeman FAS, Chang M, Catanese JJ, Ding B, Wong S, van der Helm-van Mil AHM, Neale BM, Coby J, Cui J, Tak PP, Wolbink GJ, Crusius JBA, van der Horst-Bruinsma IE, Criswell LA, Amos CI, Seldin MF, **Kastner DL**, Ardlie KG, Alfredsson L, Costenbader KH, Altshuler D, Huizinga TWJ, Shadick NA, Weinblatt ME, de Vries N, Worthington J, Seielstad M, Toes REM, Karlson EW, Begovitch AB, Klareskog L, Gregersen PK, Daly MJ, Plenge RM (2008) Common variants at *CD40* and other loci confer risk of rheumatoid arthritis. *Nature Genet* **40**:1216–1223. PMC2757650
 133. Korman BD, Seldin MF, Taylor KE, Le JM, Lee AT, Plenge RM, Amos CI, Criswell LA, Gregersen PK, **Kastner DL**, Remmers EF (2009) The chromosome 7q region association with rheumatoid arthritis in families in a British population is not replicated in a North American case-control series. *Arthritis Rheum* **60**:47–51. PMC2741408
 134. Lee HS, Korman BD, Le JM, **Kastner DL**, Remmers EF, Gregersen PK, Bae SC (2009) Genetic risk factors for rheumatoid arthritis differ in Caucasian and Korean populations. *Arthritis Rheum* **60**:364–371. PMC2770844
 135. Masters SL, Simon A, Aksentijevich I, **Kastner DL** (2009) *Horror Autoinflammaticus*: the molecular pathophysiology of autoinflammatory disease. *Annu Rev Immunol* **27**:621–668. PMC2996236
 136. Aksentijevich I, Masters SL, Ferguson PJ, Dancey P, Frenkel J, van Royen-Kerkhoff A, Laxer R, Tedgård U, Cowen EW, Pham T-H, Booty M, Estes JD, Sandler NG, Plass N, Stone D, Turner ML, Hill S, Butman JA, Schneider R, Babyn P, El-Shanti HI, Pope E, Barron K, Bing X, Laurence A, Lee C-CR, Chapelle D, Clarke GI, Ohson K, Nicholson M, Gadina M, Yang B, Korman BD, Gregersen PK, van Hagen M, Hak AE, Huizinga M, Rahman P, Douek DC, Remmers EF, **Kastner DL**, Goldbach-Mansky R (2009) Deficiency of the interleukin-1 receptor-antagonist (DIRA) – a systemic autoinflammatory disease of skin and bone. *N Engl J Med* **360**:2426–2437. PMC2876877

137. Booty MG, Chae JJ, Masters SL, Remmers EF, Barham B, Le JM, Barron KS, **Kastner DL**, Aksentijevich I (2009) Familial Mediterranean fever with a single *MEFV* mutation: where is the second hit? *Arthritis Rheum* **60**:1851–1861. PMC2753538
138. Chae JJ, Aksentijevich I, **Kastner DL** (2009) Advances in the understanding of familial Mediterranean fever and possibilities for targeted therapy. *Br J Haematol*, **146**:467 – 478. PMC2759843
139. Brydges SD, Mueller JL, McGeough MD, Pena CA, Misaghi A, Gandhi C, Boyle DL, Firestein GS, Horner AA, Soroosh P, Watford WT, O’Shea JJ, **Kastner DL**, Hoffman HM (2009) Inflammasome-mediated disease animal models reveal roles for innate but not adaptive immunity. *Immunity* **30**:875–887. PMC2759865
140. Gregersen PK, Amos CI, Lee AT, Lu E, Remmers EF, **Kastner DL**, Seldin MF, Criswell LA, Plenge RM, Holers VM, Mikuls TR, Sokka T, Moreland LW, Bridges SL Jr, Xie G, Begovich AB, Siminovitch KA (2009) *REL*, encoding a member of the NF- κ B family of transcription factors, is a newly defined risk locus for rheumatoid arthritis. *Nature Genet* **41**:820–823. PMC2705058
141. Waite AL, Schaner P, Richards N, Balci-Peynircioglu B, Masters SL, Brydges SD, Fox M, Hong A, Yilmaz E, **Kastner DL**, Reinherz EL, Gumucio DL (2009) Pyrin modulates the intracellular distribution of PSTPIP1. *PLoS One* **4**:e6147. PMC2702820
142. Raychaudhuri S, Thomson BP, Remmers EF, Eyre S, Hinks A, Guiducci C, Catanese JJ, Xie G, Shahl EA, Chen R, Alfredsson L, Amos CI, Ardlie KG; BIRAC Consortium; Barton A, Bowes J, Burt NP, Chang M, Coblyn J, Costenbader KH, Criswell LA, Crusius JB, Cui J, De Jager PL, Ding B, Emery P, Flynn E, Harrison P, Hocking LJ, Huizinga TW, **Kastner DL**, Ke X, Kurreeman FA, Lee AT, Liu X, Li Y, Martin P, Morgan AW, Padyukov L, Reid DM, Seielstad M, Seldin MF, Shadick NA, Steer S, Tak PP, Thomson W, van der Helm-van Mil AH, van der Horst-Bruinsma IE, Weinblatt ME, Wilson AG, Wolbink GJ, Wordsworth P; YEAR Consortium; Altshuler D, Karlson EW, Toes RE, de Vries N, Begovich AB, Siminovitch KA, Worthington J, Klareskog L, Gregersen PL, Daly MJ, Plenge RM (2009) Genetic variants at CD28, PRDM1, and CD2/CD58 are associated with rheumatoid arthritis risk. *Nature Genet* **41**:1313-1318. PMC3142887
143. Goldbach-Mansky R, **Kastner DL** (2009) Autoinflammation: the prominent role of IL-1 in monogenic autoinflammatory diseases and implications for common illnesses. *J Allergy Clin Immunol* **124**:1141-1149. PMC2995268
144. Amos CI, Chen WV, Seldin MF, Remmers EF, Taylor KE, Criswell LA, Lee AT, Plenge RM, **Kastner DL**, Gregersen PK (2009) Data for Genetic Analysis Workshop 16 Problem 1, association analysis of rheumatoid arthritis data. *BMC Proc* **3 Suppl 7**:S2. PMC2795916
145. **Kastner DL**, Aksentijevich I, Goldbach-Mansky R (2010) Autoinflammatory disease reloaded: a clinical perspective. *Cell* **140**:784-790. PMC3541025
146. Lehmann P, Salzberger B, Haerle P, Aksentijevich I, **Kastner D**, Schmoelmerich J, Rosenfeld S, Mueller-Ladner U (2010) Variable intrafamilial expressivity of the rare tumor necrosis factor-receptor associated periodic syndrome-associated mutation I170N that affects the TNFR1A cleavage site. *Mod Rheumatol* **20**:311-315. PMC3074501
147. Stahl EA, Raychaudhuri S, Remmers EF, Xie G, Eyre S, Thomson BP, Yonghong L, Kurreeman FAS, Zhernakova A, Hinks A, Guiducci C, Chen R, Alfredsson L, Amos CI, Ardlie KG, BIRAC Consortium, Barton A, Bowes J, Brouwer E, Burt NP, Catanese JJ, Coblyn J, Coenen MJH, Costenbader KH, Criswell LA, Crusius JBA, Cui J, de Bakker PIW, De Jager PL, Ding B, Emery P, Flynn E, Harrison P, Hocking LJ, Huizinga TWJ, **Kastner DL**, Ke X, Lee AT, Liu X, Martin P, Morgan AW, Padyukov L, Posthumus MD, Radstake TRDJ, Reid DM, Seielstad M, Seldin MF, Shadick NA, Steer S, Tak PP,

- Thomson W, van der Helm-van Mil AHM, van der Horst-Bruinsma IE, van der Schoot CE, van Riel PLCM, Weinblatt ME, Wilson AG, Wolbink GJ, Wordsworth BP, YEAR Consortium, Wijmenga C, Karlson EW, Toes REM, de Vries N, Begovich AB, Worthington J, Siminovitch KA, Gregersen PK, Klareskog L, Plenge RM (2010) Genome-wide association study meta-analysis identifies seven new rheumatoid arthritis risk loci. *Nature Genet* **42**:508-514. PMC4243840
148. Simon A, Park H, Maddipati R, Lobito AA, Bulua AC, Jackson AL, Chae JJ, Ettinger R, de Koning HD, Cruz AC, **Kastner DL**, Komarow H, Siegel RM (2010) Concerted action of wild-type and mutant TNF receptors enhances inflammation in TNF receptor 1-associated periodic fever syndrome. *Proc Natl Acad Sci USA* **107**:9801-9806. PMC2906866
149. Ryan JG, Masters SL, Booty MG, Habal N, Alexander JD, Barham BK, Remmers EF, Barron KS, **Kastner DL**, Aksentijevich I (2010) Clinical features and functional significance of the P369S/R408Q variant in pyrin, the familial Mediterranean fever protein. *Ann Rheum Dis* **69**:1383-8. PMC3570240
150. Cortesio CL, Cooper KM, Wernimont SA, **Kastner DL**, Huttenlocher A (2010) Impaired podosome formation and invasive migration of macrophages from patients with a *PSTPIP1* mutation and PAPA syndrome. *Arthritis Rheum* **62**:2556 – 2558. PMC2921034
151. Remmers EF, Cosan F, Kirino Y, Ombrello MJ, Abaci N, Satorius C, Le JM, Yang B, Korman BD, Cakris A, Aglar O, Emrence Z, Azakli H, Ustek D, Tugal-Tutkun I, Akman-Demir G, Chen W, Amos CI, Dizon MB, Kose AA, Azizlerli G, Erer B, Brand OJ, Kaklamani VG, Kaklamanis P, Ben-Chetrit E, Stanford M, Fortune F, Ghabra M, Ollier WER, Cho Y-H, Bang D, O'Shea J, Wallace GR, Gadina M, **Kastner DL**, Gül A (2010) Genome-wide association study identifies variants in the MHC class I, *IL10*, and *IL23R-IL12RB2* regions associated with Behçet's disease. *Nature Genet* **42**:698 – 702. PMC2923807
152. Wang HY, Gopalan V, Aksentijevich I, Yeager M, Ma CA, Mohamoud YA, Quinones M, Matthews C, Boland J, Niemela JE, Torgerson TR, Giliani S, Uzel G, Orange JS, Shapiro R, Notarangelo L, Ochs HD, Fleisher T, **Kastner D**, Chanock SJ, Jain A (2010) A custom 148 gene-based resequencing chip and the SNP explorer software: new tools to study antibody deficiency. *Human Mutat* **31**:1080 – 1088. PMC2945728
153. Smith EJ, Allantaz F, Bennett L, Zhang D, Gao X, Wood G, **Kastner DL**, Punaro M, Aksentijevich I, Pascual V, Wise CA (2010) Clinical, molecular, and genetic characteristics of PAPA syndrome: a review. *Curr Genomics* **11**:519 – 527. PMC3048314
154. Ombrello M, **Kastner DL** (2011) Autoinflammation 2010: Expanding clinical spectrum and broadening therapeutic horizons. *Nature Rev Rheumatol* **7**:82 – 84. PMC3393888
155. Bulua AC, Simon A, Maddipati R, Pelletier M, Park H, Kim K-Y, Sack MN, **Kastner DL**, Siegel RM (2011) Mitochondrial reactive oxygen species promote production of pro-inflammatory cytokines and are elevated in a TNFR1-associated periodic syndrome (TRAPS). *J Exp Med* **208**:519 – 533.
156. Yang Y, Remmers EF, Ogunwole CB, **Kastner DL**, Gregersen PK, Li W (2011) Effective sample size: quick estimation of the effect of related samples in genetic case-control association analyses. *Comput Biol Chem* **35**:40 – 49. PMC3119257
157. Olsson M, Meadows JRS, Truvé K, Pielberg GR, Puppo F, Mauceli E, Quilez J, Tonomura N, Zanna G, Docampo MJ, Bassols A, Avery AC, Karlsson EK, Thomas A, **Kastner DL**, Bongcam-Rudloff E, Webster MT, Sanchez A, Hedhammar Å, Remmers EF, Andersson L, Ferrer L, Tintle L, Lindblad-Toh K (2011) A novel unstable duplication upstream of *HAS2* predisposes to a breed-defining skin phenotype and a periodic fever syndrome in Chinese Shar-Pei dogs. *Plos Genet* **7**:e1001332. PMC3060080

158. Stojanov S, Lapidus S, Chitkara P, Feder H, Salazar JC, Fleisher TA, Brown MR, Edwards KM, Ward MM, Colbert RA, Sun H-W, Wood GM, Barham BK, Jones A, Aksentijevich I, Goldbach-Mansky R, Athreya BH, Barron KS, **Kastner DL** (2011) Periodic fever, aphthous stomatitis, pharyngitis, and adenitis (PFAPA) is a disorder of innate immunity and Th1 activation responsive to IL-1 blockade, *Proc Natl Acad Sci USA* **108**:7148 – 7153. PMC3084055
159. Chae JJ, Cho Y-H, Lee G-S, Cheng J, Liu PP, Feigenbaum L, Katz SI, **Kastner DL** (2011) Pypin mutations induce *NLRP3*-independent IL-1 β activation and severe autoinflammation in mice. *Immunity* **34**:755 – 768. PMC3129608
160. Aksentijevich I, **Kastner DL** (2011) Genetics of monogenic autoinflammatory diseases: past successes, future challenges. *Nat Rev Rheumatol* **7**:469 – 478. PMID:21727933
161. Demidowich AP, Freeman AF, Kuhns DB, Aksentijevich I, Gallin JI, Turner ML, **Kastner DL**, Holland SM (2012) Genotype, phenotype, and clinical course in five patients with pyogenic arthritis, pyoderma gangrenosum, and acne (PAPA) syndrome. *Arthritis Rheum* **64**:2022-2027. PMC3737487
162. Ombrello MJ, Remmers EF, Sun G, Freeman AF, Datta S, Torabi-Parizi P, Subramanian N, Bunney TD, Baxendale RW, Martins MS, Romberg N, Komarow H, Aksentijevich I, Kim HS, Ho J, Cruse G, Jung M-Y, Gilfillan AM, Metcalfe DD, Nelson C, O'Brien M, Wisch L, Stone K, Douek DC, Gandhi C, Wanderer AA, Lee H, Nelson SF, Shianna KV, Cirulli ET, Goldstein DB, Long EO, Moir S, Meffre E, Holland SM, **Kastner DL**, Katan M, Hoffman HM, Milner JD (2012) Cold urticaria, immunodeficiency, and autoimmunity related to *PLCG2* deletions. *N Engl J Med*, **366**:330-338. PMC3298368
163. Bulua AC, Mogul DB, Aksentijevich I, Singh H, He D, Muenz L, Ward MM, Yarboro C, **Kastner DL**, Siegel RM, Hull KM (2012) Efficacy of etanercept in the tumor necrosis factor receptor-associated periodic syndrome (TRAPS). *Arthritis Rheum* **64**:908-913. PMC3882089
164. Bonar SL, Brydges SD, Mueller JL, McGeough MD, Pena C, Chen D, Grimston SK, Hickman-Brecks CL, Ravindran S, McAlinden A, Novack DV, **Kastner DL**, Civitelli R, Hoffman HM, Mbalaviele G (2012) Constitutively activated *NLRP3* inflammasome causes inflammation and abnormal skeletal development in mice. *PLoS One* **7**:e35979. PMC3338787
165. Park H, Bourla AB, **Kastner DL**, Colbert RA, Siegel RM (2012) Lighting the fires within: the cell biology of autoinflammatory diseases. *Nat Rev Immunol* **12**:570-580. PMC4165575
166. Hashkes PJ, Spalding SJ, Giannini EH, Huang B, Johnson A, Park G, Barron KS, Weisman MH, Pashinian N, Reiff AO, Samuels J, Wright D, **Kastner DL**, Lovell DJ (2012) Riloncept for colchicine resistant or intolerant familial Mediterranean fever: a randomized controlled trial. *Ann Intern Med* **157**:533-541. PMID:23070486
167. Zhou Q, Lee G-S, Brady J, Sheikh A, Katan M, Martins MS, Bunney TD, Datta S, Milner J, Ombrello A, Stone D, Ombrello MJ, Khan J, **Kastner DL**, Aksentijevich I (2012) Exome sequencing identifies a hypermorphic missense mutation in the *PLCG2* gene as the cause of a dominantly inherited autoinflammatory disease with immunodeficiency. *Am J Hum Genet* **91**:713-720. PMC3484656
168. Ombrello MJ, **Kastner DL**, Milner JD (2012) HOIL and water: the two faces of HOIL-1 deficiency. *Nature Immunol* **13**:1133-1135. PMID:23160206
169. Lee G-S, Subramanian N, Kim A, Aksentijevich I, Goldbach-Mansky R, Sacks DB, Germain RN, **Kastner DL**, Chae JJ (2012) The calcium-sensing receptor regulates the *NLRP3* inflammasome through intracellular Ca²⁺ and cAMP. *Nature* **492**:123-127. PMC4175565
170. Masters SL, Gerlic M, Metcalf D, Preston S, Pellegrini M, O'Donnell JA, McArthur K, Baldwin TM, Chevrier S, Nowell CJ, Cengia LH, Henley KJ, Collinge JE, **Kastner DL**, Feigenbaum L, Hilton DJ,

Alexander WS, Kile BT, Croker BA (2012) NLRP1 activation induces pyroptosis of hematopoietic progenitor cells. *Immunity* **37**:1009-1023. PMC4275304

171. Balow JE Jr, Ryan J, Chae JJ, Booty MG, Bulua A, Sun H-W, Greene J, Barham B, Goldbach-Mansky R, **Kastner DL**, Aksentijevich I (2012) Microarray-based gene expression profiling in patients with CAPS defines a disease-related signature and IL-1-responsive transcripts. *Ann Rheum Dis* epub ahead of print, doi:10.1136/annrheumdis-2012-20282. PMC4174357
172. Kirino Y, Bertias G, Ishigatsubo Y, Mizuki N, Tugal-Tutkun I, Seyahi E, Ozyazgan Y, Sacli FS, Erer B, Inoko H, Emrence Z, Cakar A, Abaci N, Ustek D, Satorius C, Ueda A, Takeno M, Kim Y, Wood GM, Ombrello MJ, Meguro A, Gül A, Remmers EF, **Kastner DL** (2013) Genome-wide association analysis identifies new susceptibility loci for Behçet's disease and epistasis between *HLA-B*51* and *ERAP1*. *Nature Genet* **45**:202-207. PMC3810947
173. Kirino Y, Zhou Q, Ishigatsubo Y, Mizuki N, Tugal-Tutkun I, Seyahi E, Ozyazgan Y, Ugurlu S, Erer B, Abaci N, Ustek D, Meguro A, Ueda A, Takeno M, Inoko H, Ombrello MJ, Satorius C, Maskeri B, Mullikin JC, Sun H-W, Gutierrez-Cruz G, Kim Y, Wilson AF, **Kastner DL**, Gül A, Remmers EF (2013) Targeted resequencing implicates the familial Mediterranean fever gene *MEFV* and the toll-like receptor 4 gene *TLR4* in Behçet's disease. *Proc Natl Acad Sci USA* **110**:8134-8139. PMC3657824
174. Zhou Q, Yang D, Ombrello AK, Zavialov AV, Toro C, Zavialov AV, Stone DL, Chae JJ, Rosenzweig SD, Bishop K, Barron K, Kuehn HS, Hoffmann P, Negro A, Tsai WL, Cowen EW, Pei W, Milner JD, Silvin C, Heller T, Chin DT, Patronas NJ, Barber JS, Lee C-CR, Wood GM, Ling A, Kelly SJ, Kleiner DE, Mullikin J, Ganson NJ, Kong HH, Hambleton S, Candotti F, Quezado MM, Calvo K, Alao H, Barham BK, Jones A, Meschia JF, Worrall BB, Kasner SE, Rich SS, Goldbach-Mansky R, Abinum M, Chalom E, Gotte AC, Punaro M, Pascual V, Verbsky J, Torgerson TR, Singer NG, Gershon TR, Ozen S, Karadag O, Fleisher TA, Remmers EF, Burgess SM, Moir SL, Gadina M, Sood R, Hershfield M, Boehm M, **Kastner DL**, Aksentijevich I (2014) Early-onset stroke and vasculopathy associated with mutations in *ADA2*. *N Engl J Med* **370**:911-920. PMC4193683
175. Tsang JS, Schwartzberg PL, Kotliarov Y, Biancotto A, Xie Z, Germain RN, Wang E, Olnes MJ, Narayanan M, Golding H, Moir S, Dickler HB, Perl S, Cheung F, The Baylor HIPC Center, The CHI Consortium (2014) Global analyses of human immune variation reveal baseline predictors of postvaccination responses. *Cell* **157**:499-513. PMC4139290
176. Ombrello MJ, Kirino Y, de Bakker PI, Gül A, **Kastner DL**, Remmers EF (2014) Behçet disease-associated MHC class I residues implicate antigen binding and regulation of cell-mediated cytotoxicity. *Proc Natl Acad Sci USA* **111**:8867-8872. PMC4066484
177. Ombrello MJ, Sikora KA, **Kastner DL**. Genetics, genomics, and their relevance to pathology and therapy (2014) *Best Practice and Research Clinical Rheumatology* **28**:175-189. PMC4149217
178. **Kastner DL**, Zhou Q, Aksentijevich I (2014) Mutant *ADA2* in vasculopathies. *N Engl J Med* **371**:480-481. PMID:25075844
179. Liu Y, Jesus AA, Marrero B, Yang D, Ramsey SE, Montealegre Sanchez GA, Tenbrock K, Wittkowski H, Jones OY, Kuehn HS, Lee C-CR, DiMattia MA, Cowen EW, Gonzalez B, Palmer I, DiGiovanna JJ, Biancotto A, Kim H, Tsai WL, Trier AM, Huang Y, Stone DL, Hill S, Kim HJ, St. Hilaire C, Gurprasad S, Plass N, Chapelle D, Horkayne-Szakaly I, Foell D, Barysenka A, Candotti F, Holland SM, Hughes JD, Mehmet H, Issekutz AC, Raffeld M, McElwee J, Fontana JR, Minniti CP, Moir S, **Kastner DL**, Gadina M, Steven AC, Wingfield PT, Brooks SR, Rosenzweig SD, Fleisher TA, Deng Z, Boehm M, Paller AS, Goldbach-Mansky R (2014) Activated STING in a vascular and pulmonary syndrome. *N Engl J Med* **371**:507-518. PMC4174543

180. Chang Z, Spong C, de Jesus AA, Davis M, Plass N, Stone DL, Chapelle D, Hoffmann P, **Kastner DL**, Barron K, Goldbach-Mansky RT, Stratton P (2014) Anakinra use during pregnancy in patients with cryopyrin-associated periodic syndromes. *Arthritis Rheum* **66**:3227-3232. PMC4323990
181. Bakir-Gungor B, Remmers EF, Meguro A, Mizuki N, **Kastner DL**, Gül A, Sezerman OU (2014) Identification of possible pathogenic pathways in Behçet's disease using genome-wide association study data from two different populations. *Eur J Hum Genet*, epub ahead of print. (2015 May; 23 (5):678-87.doi: 10.1038/ejhg.2014.158). PMC4402634
182. Giannelou A, Zhou Q, **Kastner DL** (2014) When less is more: primary immunodeficiency with an autoinflammatory kick (2014) *Curr Opin Allergy Clin Immunol* **14**:491-500. PMC4212813
183. Chae JJ, Park YH, Park C, Hwang I-Y, Hoffmann P, Kehrl JH, Aksentijevich I, **Kastner DL** (2015) Connecting two pathways through Ca²⁺ signaling: NLRP3 inflammasome activation induced by a hypomorphic *PLCG2* mutation. *Arthritis Rheum* **67**:563-567. PMC4369162
184. Bakir-Gungor B, Remmers EF, Meguro A, Mizuki N, **Kastner DL**, Gül A, Sezerman OU (2015) Reply to Stoimenis et al. *Eur J Hum Genet* **23**:1280. PMC 4592084
185. Coll RC, Robertson AAB, Chae JJ, Higgins SC, Muñoz-Planillo R, Inserra MC, Vetter I, Dungan LS, Monks BG, Stutz A, Croker DE, Butler MS, Haneklaus M, Sutton CE, Núñez G, Latz E, **Kastner DL**, Mills KHG, Masters SL, Schroder K, Cooper MA, O'Neill LAJ (2015) A small molecule inhibitor of the NLRP3 inflammasome for the treatment of inflammatory diseases. *Nat Med* **21**:248-255. PMC4392179
186. Sokolowska M, Chen L-Y, Liu Y, Martinez-Anton A, Qi H-Y, Logun C, Alsaaty S, Park YH, **Kastner DL**, Chae JJ, Shelhamer JH (2015) Prostaglandin E₂ inhibits NLRP3 inflammasome activation through EP4 receptor and intracellular cyclic AMP in human macrophages. *J Immunol* **194**:5472-5487. PMC4433768
187. Zhou Q, Aksentijevich I, Wood GM, Walts AD, Hoffmann P, Remmers EF, **Kastner DL**, Ombrello AK (2015) Cryopyrin-associated periodic syndrome caused by a myeloid-restricted somatic *NLRP3* mutation. *Arthritis Rheum* **67**:2482-2486. PMC4551575
188. Kim ML, Chae JJ, Park YH, de Nardo D, Stirzaker RA, Ko H-J, Tye H, Cengia L, DiRago L, Metcalf D, Roberts AW, **Kastner DL**, Lew AM, Lyras D, Kile BT, Croker BA, Masters SL (2015) Aberrant actin depolymerization triggers the pyrin inflammasome and autoinflammatory disease that is dependent on IL-18, not IL-1β. *J Exp Med* **212**:927-938. PMC4451132
189. Ombrello MJ, Remmers EF, **Kastner DL** (2015) Endoplasmic reticulum-associated amino-peptidase 1 and rheumatic disease: genetics. *Curr Opin Rheumatol* **27**:349-356. PMC4565054
190. Lubick KJ, Robertson SJ, McNally KL, Freedman BA, Rasmussen AL, Taylor RT, Walts AD, Tsuruda S, Sakai M, Ishizuka M, Boer EF, Foster EC, Chiramel AI, Addison CB, Green R, **Kastner DL**, Katze MG, Holland SM, Forlino A, Freeman AF, Boehm M, Yoshii K, Best SM (2015) Flavivirus antagonism of type I interferon signaling reveals prolidase as a regulator of IFNAR1 surface expression. *Cell Host Microbe* **18**:61-74. PMC4505794
191. Takeuchi M, **Kastner DL**, Remmers EF (2015) The immunogenetics of Behçet's disease: a comprehensive review. *J Autoimmun* **64**:137-148. PMC4628864
192. Brehm A, Liu Y, Sheikh A, Marrero, B, Omoyinmi E, Zhou Q, Montealegre Sanchez MA, Biancotto A, Reinhardt A, Jesus AA, Pelletier M, Tsai WL, Remmers EF, Kardava L, Hill S, Kim H, Lachmann HL, Megarbane A, Chae JJ, Brady J, Castillo RD, Brown D, Casano AV, Ling G, Chapelle D, Huang Y, Stone D, Chen Y, Sotzny F, Lee C-CR, **Kastner DL**, Torrelo A, Zlotogorski A, Moir S, Gadina M, McCoy P, Wesley R, Rother K, Hildebrand PW, Brogan P, Krüger E, Aksentijevich I, Goldbach-Mansky

- R. (2015) Additive loss-of-function proteasome subunit mutations in CANDLE/PRAAS patients promote type I IFN production. *J Clin Invest*, **125**(11):4196-211. PMC4639987
193. Ombrello MJ, Remmers EF, Tachmazidou I, Grom A, Foell D, Haas J-P, Martini A, Gattorno M, Özen S, Prahallad S, Zeff AS, Bohnsack JF, Mellins ED, Ilowite NT, Russo R, Len C, Hilario MOE, Oliveira S, Yeung RSM, Rosenberg A, Wedderburn LR, Anton J, Schwarz T, Hinks A, Bilginer Y, Park J, Cobb J, Satorius C, Han B, Baskin E, Signa S, Duerr R, Achkar JP, Kamboh MI, Kaufman K, Kottyan LC, Pinto D, Scherer SW, Alarcón-Riquelme ME, Docampo E, Estivill X, Gül A, British Society of Pediatric and Adolescent Rheumatology (BSPAR) Study Group, Childhood Arthritis Prospective Study (CAPS) Group, Randomized Placebo Phase Study of Rilonacept in sJIA (RAPPORT) Investigators, Sparks-Childhood Arthritis Response to Medication Study (CHARMS) Group, Biologically Based Outcome Predictors in JIA (BBOP) Group, de Bakker PIW, Raychaudhuri S, Langefeld CD, Thompson SD, Zeggini E, Thomson W, **Kastner DL**, Woo P, on behalf of the International Childhood Arthritis Genetics (INCHARGE) Consortium (2015 Dec 29) *HLA-DRB1*11* and variants of the MHC class II locus are strong risk factors for systemic juvenile idiopathic arthritis. *Proc Natl Acad Sci USA*, **112**:15970-15975. PMC4702958
194. Zhou Q, Wang H, Schwartz DM, Stoffels M, Park YH, Zhang Y, Yang D, Demirkaya E, Takeuchi M, Tsai WL, Layons JJ, Yu X, Ouyang C, Chen C, Chin DT, Zaal K, Chandrasekharappa SC, Hanson EP, Yu Z, Mullikin JC, Hasni SA, Wertz IE, Ombrello AK, Stone DL, Hoffmann P, Jones A, Barham BK, Leavis HL, van Royen-Kerkof A, Sibley C, Batu ED, Gül A, Siegel RM, Boehm M, Milner JD, Ozen S, Gadina M, Chae J, Laxer RM, **Kastner DL**, Aksentijevich I (2016) Loss-of-function mutations in *TNFAIP3* leading to A20 haploinsufficiency cause an early-onset autoinflammatory disease. *Nature Genet* **48**:67-73. PMC4777523
195. Boyden SE, Desai A, Cruse G, Young ML, Bolan HC, Scott LM, Eisch AR, Long RD, Lee C-CR, Satorius CL, Pakstis AJ, Olivera A, Mullikin JC, Chouery E, Mégarbané, Medlej-Hashim M, Kidd KK, **Kastner DL**, Metcalfe DD, Komarow HD (2016) Vibratory urticaria associated with a missense variant in *ADGRE2*. *N Engl J Med* **374**:656-663. PMC4782791
196. Ozen S, Demirkaya E, Erer B, Livneh A, Ben-Chetrit E, Giancane G, Ozdogan H, Abu I, Gattorno M, Hawkins PN, Yuce S, Kallinich T, Bilginer Y, **Kastner D**, Carmona L (2016) EULAR recommendations for the management of familial Mediterranean fever. *Ann Rheum Dis*: **75**:644-651. PMID:26802180
197. Park YH, Wood G, **Kastner DL**, Chae JJ (2016) Pyrin inflammasome activation and RhoA signaling in the autoinflammatory diseases FMF and HIDS. *Nat Immunol* **17**:914-921. PMID: 27270401, PMC4955684
198. Akula MK, Shi M, Jiang Z, Foster CE, Miao D, Li AS, Zhang X, Gavin RM, Forde SD, Germain G, Carpenter S, Rosadini CV, Gritsman K, Chae JJ, Hampton R, Silverman N, Gravallese EM, Kagan JC, Fitzgerald KA, **Kastner DL**, Golenbock DT, Bergo MO, Wang D (2016) Control of the innate immune response by the mevalonate pathway. *Nat Immunol* **17**:922-929. PMID:27270400, PMC4955724
199. Stoffels M, **Kastner DL** (2016) Old dogs, new tricks: monogenic autoinflammatory disease unleashed. *Annu Rev Genom Hum Genet* **17**:18.1-18.28. PMID:27362340
200. Takeuchi M, Ombrello MJ, Kirino Y, Erer B, Tugal-Tutkun I, Seyahi E, Özyazgan Y, Watts NR, Gül A, **Kastner DL**, Remmers E (2016) A single endoplasmic reticulum aminopeptidase-1 protein allotype is a strong risk factor for Behçet's disease in *HLA-B*51* carriers. *Ann Rheum Dis* **75**:2208-2211. PMID:27217550
201. Bhattacharyya T, Jha S, Wang H, **Kastner DL**, Remmers EF (2016) Hypophosphatasia and the risk of atypical femur fractures: a case-control study. *BMC Musculoskelet Disord* **17**:332. PMID:27507156, PMC4977896

202. Zhou Q, Yu X, Demirkaya E, Deutch N, Stone D, Tsai WL, Kuehn HS, Wang H, Yang D, Park YH, Ombrello AK, Blake M, Romeo T, Remmers EF, Chae JJ, Mullikin JC, Güzel F, Milner JD, Boehm M, Rosenzweig SD, Gadina M, Welch SB, Özen S, Topaloglu R, Abinum M, **Kastner DL**, Aksentijevich I (2016) Biallelic hypomorphic mutations in a linear deubiquitinase define otulipenia, an early-onset autoinflammatory disease. *Proc Natl Acad Sci USA* **113**:10127-10132. PMID:27559085, PMC5018768
203. Chung LK, Park YH, Zheng Y, Brodsky IE, Hearing P, **Kastner DL**, Chae JJ, Bliska JB (2016) The *Yersinia* virulence factor YopM hijacks host kinases to inhibit type III effector-triggered activation of the pyrin inflammasome. *Cell Host Microbe* **20**:296-306. PMID:27569559, PMC5025386
204. Erer B, Takeuchi M, Ustek D, Tugal-Tutkin I, Seyahi E, Özyazgan Y, Duymaz-Tozgir J, Gül A, **Kastner DL**, Remmers EF, Ombrello MJ (2016) Evaluation of KIR3DL1/KIR3DS1 polymorphism in Behçet's disease. *Genes Immun* **17**:396-399. PMID:27708262
205. Takeuchi M, Mizuki N, Meguro A, Ombrello MJ, Kirino Y, Satorius C, Le J, Blake M, Erer B, Kawagoe T, Ustek D, Tugal-Tutkun I, Seyahi E, Ozyazgan Y, Sousa I, Davatchi F, Francisco V, Shahram F, Abdollahi BS, Nadji A, Shafiee NM, Ghaderibarmi F, Ohno S, Ueda A, Ishigatsubo Y, Gadina M, Oliviera SA, Gül A, **Kastner DL**, Remmers EF (2017) Dense genotyping of immune-related loci implicates host responses to microbial exposure in Behçet's disease susceptibility. *Nat Genet* **49**:438-443. PMID:28166214
206. Ombrello MJ, Arthur VL, Remmers EF, Hinks A, Tachmazidou I, Grom AA, Foell D, Martini A, Gattorno M, Özen S, Prahalad S, Zeff AS, Bohnsack JF, Ilowite NT, Mellins ED, Russo R, Len C, Hilario MO, Oliveira S, Yeung RS, Rosenberg AM, Wedderburn LR, Anton J, Haas JP, Rosen-Wolff A, Minden K, Tenbrock K, Demirkaya E, Cobb J, Baskin E, Signa S, Shuldiner E, Duerr RH, Achkar JP, Kamboh MI, Kaufman KM, Kottyan LC, Pinto D, Scherer SW, Alarcón-Riquelme ME, Docampo E, Estivill X, Gül A; British Society of Pediatric and Adolescent Rheumatology (BSPAR) Study Group, Inception Cohort of Newly Diagnosed Patients with Juvenile Idiopathic Arthritis (ICON-JIA) Study Group, Childhood Arthritis Prospective Study (CAPS) Group, Randomized Placebo Phase of Riloncept in sJIA (RAPPORT) Investigators, Sparks-Childhood Arthritis Response to Medication Study (CHARMS) Group, Biologically Based Outcome Predictors in JIA (BBOP) Group, Langefeld CD, Thompson S, Zeggini E, **Kastner DL**, Woo P, Thomson W (2017) Genetic architecture distinguishes systemic juvenile idiopathic arthritis from other forms of juvenile idiopathic arthritis: clinical and therapeutic implications. *Ann Rheum Dis* **76**:906-913. PMID:27927641
207. Demirkaya E, Zhou Q, Smith CK, Ombrello MJ, Deutch N, Tsai WL, Hoffmann P, Remmers EF, Takeuchi M, Park YH, Chae JJ, Barut K, Simsek D, Adrovic A, Sahin S, Caliskan S, Chandrasekharappa SC, Hasni SA, Ombrello AK, Gadina M, **Kastner DL**, Kaplan MJ, Kasapcopur O, Aksentijevich I (2017) Deficiency in complement 1r subcomponent in early onset systemic lupus erythematosus: the role for disease-modifying alleles in a monogenic disease. *Arthritis Rheumatol* **69**:1832-1839. PMID:28544690
208. Oda H and **Kastner DL** (2017) Genomics, biology, and human illness: advances in the monogenic autoinflammatory diseases. *Rheum Dis Clin North Am* **43**:327-345. PMID:28711137
209. Netea MG, Balkwill F, Chonchol M, Cominelli F, Donath MY, Giamarellos-Bourboulis EJ, Golenbock D, Gresnigt M, Heneka MT, Hoffman HM, Hotchkiss R, Joosten LAB, **Kastner DL**, Korte M, Latz E, Libby P, Mandrup-Poulsen T, Montovani A, Mills KHG, Novak KL, O'Neill LA, Pickkers P, van der Poll T, Ridker PM, Schalkwijk J, Schwartz DA, Siegmund B, Steer CJ, Tilg H, van der Meer JWM, van de Veerdonk FL, Dinarello CA (2017) A guiding map for inflammation. *Nat Immunol* **18**:826-831. PMID:28722720
210. Manthiram K, Zhou Q, Aksentijevich I, **Kastner DL** (2017) The monogenic autoinflammatory diseases define new pathways in human innate immunity and inflammation. *Nat Immunol* **18**:832-842. PMID:28722725

211. Nakanishi H, Kawashima Y, Kurima K, Chae JJ, Ross AM, Pinto-Patarroyo G, Patel SK, Muskett JA, Ratay JS, Chattaraj P, Park YH, Grevich S, Brewer CC, Hoa M, Kim HJ, Butman JA, Broderick L, Hoffman HM, Aksentijevich I, **Kastner DL**, Goldbach-Mansky R, Griffith AJ (2017) *NLRP3* mutation and cochlear autoinflammation cause syndromic and nonsyndromic hearing loss DFNA34 responsive to anakinra treatment. *Proc Natl Acad Sci USA* **114**:E7766-E7775. PMID:28847925
212. Morgan ND, Shah AA, Mayes MD, Domsic RT, Medsger TA Jr., Steen VD, Varga J, Carns M, Ramos PS, Silver RM, Schiopu E, Khanna D, Hsu V, Gordon JK, Gladue H, Saketkoo LA, Criswell LA, Derk CT, Trojanowski MA, Shanmugam VK, Chung L, Valenzuela A, Jan R, Goldberg A, Remmers EF, **Kastner DL**, Wigley FM, Gourh P, Boin F (2017) Clinical and serologic features of systemic sclerosis in a multicenter African American cohort: analysis of the genome research in African American scleroderma patients clinical database. *Medicine* **96**:51(e8980). PMID:29390428
213. Giannelou A, Wang H, Zhou Q, Park YH, Abu-Asab MS, Ylaya K, Stone DL, Sediva A, Sleiman R, Sramkova L, Bhatla D, Serti E, Tsai WL, Yang D, Bishop K, Carrington B, Pei W, Deutch N, Brooks S, Edwan JH, Joshi S, Prader S, Kaiser D, Owen WC, Sonbul AA, Zhang Y, Niemela JE, Burgess SM, Boehm M, Rehermann B, Chae JJ, Quezado MM, Ombrello AK, Buckley RH, Grom AA, Remmers EF, Pachlopnik JM, Su HC, Gutierrez-Cruz G, Hewitt SM, Sood R, Risma K, Calvo KR, Rosenzweig SD, Gadina M, Hafner M, Sun H-W, **Kastner DL**, Aksentijevich I (2018) Aberrant tRNA processing causes an autoinflammatory syndrome responsive to TNF inhibitors. *Ann Rheum Dis* **77**:612-619. PMID:29358286
214. Aeschlimann FA, Batu ED, Canna SW, Go E, Gül A, Hoffmann P, Laevis HL, Ozen S, Schwartz DM, Stone DL, van Royen-Kerkof A, **Kastner DL**, Aksentijevich I, Laxer RM (2018) A20 haploinsufficiency (HA20): clinical phenotypes and disease course of patients with a newly recognized NF- κ B-mediated autoinflammatory disease. *Ann Rheum Dis* **77**:728-735. PMID:29317407
215. Arthur VL, Shuldiner E, Remmers EF, Hinks A, Grom AA, Foell D, Martini A, Gattorno M, Özen S, Prahallad S, Zeff AS, Bohnsack JF, Ilowite NT, Mellins ED, Russo R, Len C, Oliveira S, Yeung RSM, Rosenberg AM, Wedderburn LR, Anton J, Haas JP, Rösen-Wolff A, Minden K, Szymanski AM; INCHARGE Consortium, Thomson W, **Kastner DL**, Woo P, Ombrello MJ (2018) *IL1RN* variation influences both disease susceptibility and response to recombinant human interleukin 1 receptor antagonist therapy in systemic juvenile idiopathic arthritis. *Arthritis Rheumatol* **70**:1319-1330. PMID:29609200
216. Richards RI, Robertson SA, **Kastner DL** (2018) Neurodegenerative diseases have genetic hallmarks of autoinflammatory disease. *Hum Mol Genet* **27**:R108-R118. PMID:29684205
217. Gourh P, Remmers EF, Boyden SE, Alexander T, Morgan ND, Shah AA, Mayes MD, Doumatey A, Bentley AR, Shriner D, Domsic RT, Medsger TA Jr, Steen VD, Ramos PS, Silver RM, Korman B, Varga J, Schiopu E, Khanna D, Hsu V, Gordon JK, Saketkoo LA, Gladue H, Kron B, Criswell LA, Derk CT, Bridges SL Jr, Shanmugam VK, Kolstad KD, Chung L, Jan R, Bernstein EJ, Goldberg A, Trojanowski M, Kafaja S, Maksimowicz-McKinnon KM, Mullikin JC; NISC Comparative Sequencing Program, Adeyemo A, Rotimi C, Boin F, **Kastner DL**, Wigley FM (2018) Whole-exome sequencing to identify rare variants and gene networks that increase susceptibility to scleroderma in African Americans. *Arthritis Rheumatol* **70**:1654-1660. PMID:29732714
218. Ben-Chetrit E, Gattorno M, Gul A, **Kastner DL**, Lachmann HJ, Touitou I, Ruperto N; Paediatric Rheumatology International Trials Organization (PRINTO) and the AIDs Delphi study participants (2018) Consensus proposal for taxonomy and definition of the autoinflammatory diseases (AIDs): a Delphi study. *Ann Rheum Dis* **77**:1558-1565. PMID:30100561
219. Mistry P, Carmona-Rivera C, Ombrello AK, Hoffmann P, Seto NL, Jones A, Stone DL, Naz F, Carlucci P, Dell'Orso S, Gutierrez-Cruz G, Sun HW, **Kastner DL**, Aksentijevich I, Kaplan MJ (2018)

Dysregulated neutrophil responses and neutrophil extracellular trap formation and degradation in PAPA syndrome. *Ann Rheum Dis* **77**:1825-1833. PMID:30131320

220. Oda H, Beck DB, Kuehn HS, Sampaio Moura N, Hoffmann P, Ibarra M, Stoddard J, Tsai WL, Gutierrez-Cruz G, Gadina M, Rosenzweig S, **Kastner DL**, Notorangelo LD, Aksentijevich I (2019) Second Case of HOIP deficiency expands clinical features and defines inflammatory transcriptome regulated by LUBAC. *Front Immunol* **10**:479. PMID:30936877.
221. Ombrello AK, Qin J, Hoffmann P, Kuman P, Stone D, Jones A, Romeo T, Barham B, Pinto-Patarroyo G, Toro C, Soldatos A, Zhou Q, Deutch N, Aksentijevich I, Sheldon SL, Kelly S, Man A, Barron K, Hershfield M, Flegel WA, **Kastner DL** (2019) Treatment strategies for the deficiency of adenosine deaminase 2. *N Engl J Med* **380**:1582-1584. PMID:30995379.
222. Carmona-Rivera C, Khaznadar SS, Shwin KW, Irizarry-Caro JA, O'Neil LJ, Liu Y, Jacobson KA, Ombrello AK, Stone DL, Tsai WL, **Kastner DL**, Aksentijevich I, Kaplan MJ, Graysoon PC (2019) Deficiency of adenosine deaminase 2 triggers adenosine-mediated NETosis and TNF production in patients with DADA2. *Blood* **134**:395-406. PMID:31015188.
223. Gattorno M, Hofer M, Federici S, Vanoni F, Bovis F, Aksentijevich I, Anton J, Arostegui JI, Barron K, Ben-Chetrit E, Brogan PA, Cantarini L, Ceccherini I, De Benedetti F, Dedeoglu F, Demirkaya E, Frenkel J, Goldbach-Mansky R, Gul A, Hentgen V, Hoffman H, Kallinich T, Kone-Paut I, Kuemmerle-Deschner J, Lachmann HJ, Laxer RM, Livneh A, Obici L, Ozen S, Rowczenio D, Russo R, Shinar Y, Simon A, Toplak N, Touitou I, Uziel Y, van Gijn M, Foell D, Garassino C, **Kastner D**, Martini A, Sormani MP, Ruperto N; Eurofever Registry and the Paediatric Rheumatology International Trials Organization (PRINTO) (2019) Classification criteria for autoinflammatory recurrent fevers. *Ann Rheum Dis* **78**:1025-1032. PMID:31018962.
224. Horita N, Gül A, Aksentijevich I, **Kastner D**, Remmers EF (2019) Pseudodominance of autoinflammatory disease in a single Turkish family explained by co-inheritance of haploinsufficiency of A20 and familial Mediterranean fever. *Clin Exp Rheumatol*, epub ahead of print. PMID:31376265.
225. Schnappauf O, Chae JJ, **Kastner DL**, Aksentijevich I (2019) The pyrin inflammasome in health and disease. *Front Immunol* **10**:1745. eCollection 2019. PMID:31456795.
226. **Kastner D**, O'Shea J (2019) Henry Metzger, M.D. (1932-2018), The American Association of Immunologists President, 1991-1992. *J Immunol* **203**:1679-1680. PMID:31551398.
227. Schwartz DM, Blackstone SA, Sampaio-Moura N, Rosenzweig S, Burma AM, Stone D, Hoffmann P, Jones A, Romeo T, Barron KS, Waldman MA, Aksentijevich I, **Kastner DL**, Milner JD, Ombrello AK (2020) Type I interferon signature predicts response to JAK inhibition in haploinsufficiency of A20. *Ann Rheum Dis* **79**:429-431. PMID:31767699.
228. Lalaoui N, Boyden SE, Oda H, Wood GM, Stone DL, Chau D, Liu L, Stoffels M, Kratina T, Lawlor KE, Zaal KJM, Hoffmann PM, Etemadi N, Shield-Artin K, Biben C, Tsai WL, Blake MD, Kuehn HS, Yang D, Anderton H, Silke N, Wachsmuth L, Zheng L, Sampaio Moura N, Beck DB, Gutierrez-Cruz G, Ombrello AK, Pinto-Patarroyo GP, Kueh AJ, Herold MJ, Hall C, Wang H, Chae JJ, Dmitrieva NI, McKenzie M, Light A, Barham BK, Jones A, Romeo TM, Zhou Q, Aksentijevich I, Mullikin JC, Gross AJ, Shum AK, Hawkins ED, Masters S, Lenardo MJ, Boehm M, Rosenzweig SD, Pasparakis M, Voss AK, Gadina M, **Kastner DL**, Silke J (2020) Mutations that prevent caspase cleavage of RIPK1 cause inflammatory disease. *Nature* **577**:103-108. PMID:31827281.
229. Gourh P, Safran SA, Alexander T, Boyden SE, Morgan ND, Shah AA, Mayes MD, Doumatey A, Bentley AR, Shriner D, Domsic RT, Medsger TA Jr, Ramos PS, Silver RM, Steen VD, Varga J, Hsu V, Saketkoo LA, Schiopu E, Khanna D, Gordon JK, Kron B, Criswell LA, Gladue H, Derk CT, Bernstein EJ, Bridges SL Jr, Shanmugam VK, Kolstad KD, Chung L, Kafaja S, Jan R, Trojanowski M, Goldberg

- A, Korman BD, Steinbach PJ, Chandrasekharappa SC, Mullikin JC, Adeyemo A, Rotimi C, Wigley FM, **Kastner DL**, Boin F, Remmers EF (2020) *HLA* and autoantibodies define scleroderma subsets and risk in African and European Americans and suggest a role for molecular mimicry. *Proc Natl Acad Sci USA* **117**:552-562. PMID:31871193.
230. Naranjo AN, Bandara G, Bai Y, Smelkinson MG, Tobio A, Komarow HD, Boyden SE, **Kastner DL**, Metcalfe DD, Olivera A (2020) Critical signaling events in the mechanoactivation of human mast cells via p.C492Y-ADGRE2. *J Invest Dermatol*, epub ahead of print. PMID:32222457.
231. Manthiram K, Preite S, Dedeoglu F, Demir S, Ozen S, Edwards KE, Lapidus S, Katz AE, The Genomic Ascertainment Cohort, Feder HM Jr, Lawton M, Licameli GR, Wright PF, Le J, Barron KS, Ombrello AK, Barham B, Romeo T, Jones A, Srinivasalu H, Mudd PA, DeBiasi RL, Gul A, Marshall GS, Jones OY, Chandrasekharappa C, Stepanovskiy Y, Ferguson PJ, Schwartzberg PL, Remmers EF, **Kastner DL** (2020) Common genetic susceptibility loci link PFAPA syndrome, Behçet's disease, and recurrent aphthous stomatitis. *Proc Natl Acad Sci USA* **117**:14405-14411. PMID:32518111.
232. Schnappauf O, Ombrello AK, **Kastner DL** (2020) Deficiency of adenosine deaminase 2: is it really an elephant after all? *J Allergy Clin Immunol* **145**:1560-1561. PMID:32353490.
233. Park YH, Remmers EF, Lee W, Ombrello AK, Chung LK, Shilei Z, Stone DL, Ivanov MI, Loeven NA, Barron KS, Hoffmann P, Nehrebecky M, Akkaya-Ulum YZ, Sag E, Balci-Peynircioglu B, Aksentijevich I, Gül A, Rotimi CN, Chen H, Bliska JB, Ozen S, **Kastner DL**, Shriner D, Chae JJ (2020) Ancient familial Mediterranean fever mutations in human pyrin and resistance to *Yersinia pestis*. *Nat Immunol* **21**:857-867. PMID:32601469.
234. Schnappauf O, Zhou Q, Moura NS, Ombrello AK, Michael DG, Deutch N, Barron K, Stone DL, Hoffmann P, Hershfield M, Applegate C, Bjornsson HT, Beck DB, Witmer PD, Sobreira N, Wohler E, Chiorini JA, The American Genome Center, Dalgard CL, NIH Intramural Sequencing Center, **Kastner DL**, Aksentijevich I (2020) Deficiency of adenosine deaminase 2 (DADA2): hidden variants reduced penetrance, and unusual inheritance. *J Clin Immunol* **40**:917-926. PMID:32638197.
235. Meguro A, Ishihara M, Petrek M, Yamamoto K, Takeuchi M, Mrazek F, Kolek V, Benicka A, Yamane T, Shibuya E, Yoshino A, Isomoto A, Ota M, Yatsu K, Shijubo N, Nagai S, Yamaguchi E, Yamaguchi T, Namba K, Kaburaki T, Takase H, Morimoto SI, Hori J, Kono K, Goto H, Suda T, Ikushima S, Ando Y, Takenaka S, Takeuchi M, Yuasa T, Sugisaki K, Ohguro N, Hiraoka M, Kitaichi N, Sugiyama Y, Horita N, Asukata Y, Kawagoe T, Kimura I, Ishido M, Inoko H, Mochizuki M, Ohno S, Bahram S, Remmers EF, **Kastner DL**, Mizuki N (2020) Genetic control of *CCL24*, *POR*, and *IL23R* contributes to the pathogenesis of sarcoidosis. *Commun Biol* **3**:465. PMID:32826979.
236. Green ED, Gunter C, Biesecker LG, DiFrancesco V, Easter CL, Felsenfeld AL, Kaufman DJ, Ostrander EA, Pavan WJ, Phillippy AM, Wise AL, Dayal JG, Kish BJ, Mandich A, Wellington CR, Wetterstrand KA, Bates SA, Leja D, Vasquez S, Gahl WA, Graham BJ, **Kastner DL**, Liu P, Rodriguez LL, Solomon BD, Bonham VL, Brody LC, Hutter CM, Manolio TA (2020) Strategic vision for improving human health at The Forefront of Genomics. *Nature* **586**:683-692. PMID:33116284.
237. Stone DL, Beck DB, Manthiram K, Park YH, Chae JJ, Remmers E, **Kastner DL** (2020) The systemic autoinflammatory diseases: coming of age with the human genome. *J Allergy Clin Immunol*, epub ahead of print. PMID:32987090.
238. Beck DB, Ferrada MA, Sikora KA, Ombrello AK, Collins JC, Pei W, Balanda N, Ross DL, Ospina Cardona D, Wu Z, Patel B, Manthiram K, Groarke EM, Gutierrez-Rodriguez F, Hoffmann P, Rosenzweig S, Nakabo S, Dillon LW, Hourigan CS, Tsai WL, Gupta S, Carmona-Rivera C, Asmar AJ, Xu L, Oda H, Goodspeed W, Barron KS, Nehrebecky M, Jones A, Laird RS, Deutch N, Rowczenio D, Rominger E, Wells K, Lee C-CR, Wang W, Trick M, Mullikin J, Wigerblad G, Brooks S, Dell'Orso S, Deng Z, Chae JJ, Dulau-Florea A, Malicdan MCV, Novacic D, Colbert RA, Kaplan MJ, Gadina M, Savic

- S, Lachmann HJ, Mones A-A, Solomon BD, Retterer K, Gahl WA, Burgess SM, Akstentijevich I, Young NS, Calvo KR, Werner A, **Kastner DL**, Grayson PC (2020) Somatic mutations in *UBA1* and severe adult-onset autoinflammatory disease. *N Engl J Med* **383**:2628-2638. PMID:33108101.
239. Beck DB, Basar MA, Asmar AJ, Thompson JJ, Oda H, Uehara DT, Saida K, Pajusalu S, Talvik I, D'Souza P, Bodurtha J, Mu W, Barañano KW, Miyaka N, Wang R, Kempers M, Tamada T, Nishimura Y, Okada S, Kosho T, Dale R, Mitra A, Macnamara E, Undiagnosed Diseases Network, Matsumoto N, Inazawa J, Walkiewicz, Ōunap K, Tifft CJ, Akstentijevich I, **Kastner DL**, Rocha PP, Werner A (2021) Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. *Sci Adv* **7**:eabe2116. PMID:33523931.
240. Schnappauf O, Sampaio Moura N, Akstentijevich I, Stoffels M, Ombrello AK, Hoffmann P, Barron K, Remmers EF, Hershfield M, Kelly SJ, NISC Comparative Sequencing Program, Cuthbertson D, Carette S, Chung SA, Forbess L, Khalidi NA, Koenig CL, Langford CA, McAlear CA, Monach PA, Moreland L, Pagnoux C, Seo P, Springer JM, Sreih AG, Warrington KJ, Ytterberg SR, **Kastner DL**, Grayson PC, Merkel PA, Vasculitis Clinical Research Consortium (2021) Sequence-based screening of patients with idiopathic polyarteritis nodosa, granulomatosis with polyangiitis, and microscopic polyangiitis for deleterious genetic variants in *ADA2*. *Arthritis Rheumatol* **73**:512-519. PMID:33021335.
241. Schwartz DM, Kitakule MM, Dizon B, Gutierrez-Huerta C, Blackstone SA, Burma AM, Son A, Deutch N, Rosenzweig S, Komarow H, Stone D, Jones A, Nehrebecky M, Hoffmann P, Romeo T, Barron K, Akstentijevich I, Ombrello AK, Goldbach-Mansky R, **Kastner DL**, Milner JD, Frischmeyer-Guerrero P (2021) Systematic evaluation of nine monogenic autoinflammatory diseases reveals common and disease-specific correlations with allergy-related features. *Ann Rheum Dis*, epub ahead of print. PMID:33619160.
242. Ferrada MA, Sikora KA, Luo Y, Wells KV, Patel B, Groarke EM, Ospina Cardona D, Rominger E, Hoffmann P, Le MT, Deng, Z, Quinn KA, Rose E, Tsai WL, Wigerblad G, Goodspeed W, Jones A, Wilson L, Goodspeed W, Schnappauf O, Laird RS, Kim J, Allen C, Sirajuddin A, Chen M, Gadina M, Calvo KR, Kaplan MJ, Colbert RA, Akstentijevich I, Young NS, Savic S, **Kastner DL**, Ombrello AK, Beck DB, Grayson PC (2021) Somatic mutations in *UBA1* define a distinct subset of patients with relapsing chondritis. *Arthritis Rheumatol* **73**:1886-1895. PMID:33779074.
243. Schnappauf O, Heale L, Dissanayake D, Tsai WL, Gadina M, Leto TL, **Kastner DL**, Malech HL, Kuhns DB, Akstentijevich I, Laxer RM (2021) Homozygous variant p.Arg90His in *NCF1* is associated with early-onset interferonopathy: a case report. *Pediatr Rheumatol Online J* **19**:54. PMID:33892719.
244. Beck DB, Grayson PC, **Kastner DL** (2021) Mutant *UBA1* and severe adult-onset autoinflammatory disease. Reply. *N Engl J Med* **384**:2164-2165. PMID:34077654.
245. Taft J, Markson M, Legarda D, Patel R, Chan M, Malle L, Richardson A, Gruber C, Martin-Fernández M, Mancini GMS, van Laar JAM, van Pelt P, Buta S, Wokke BHA, Sabli IKD, Sancho-Shimizu V, Pimpale Chavan P, Schnappauf O, Khubchandani R, Cüceoglu MK, Özen S, **Kastner DL**, Ting AT, Akstentijevich I, Hollink IHIM, Bogunovic D (2021) Human *TBK1* deficiency leads to autoinflammation driven by TNF-induced cell death. *Cell* **184**:4447-4463.e20. PMID:34363755.
246. Obiorah IE, Patel BA, Groarke EM, Wang W, Trick M, Ombrello AK, Ferrada MA, Wu Z, Gutierrez-Rodrigues F, Lotter J, Wilson L, Hoffmann P, Ospina Cardona D, Patel N, Florea AD, **Kastner DL**, Grayson PC, Beck DB, Young NS, Calvo KR (2021) Benign and malignant hematologic manifestations in patients with VEXAS syndrome due to somatic mutations in *UBA1*. *Blood Adv* **5**:3203-3215. PMID:34427584.
247. Barron KS, Akstentijevich I, Deutch NT, Stone DL, Hoffmann P, Videgar-Laird R, Soldatos A, Bergerson J, Toro C, Cudrici C, Nehrebecky M, Romeo T, Jones A, Boehm M, Kanakry JA, Dimitrova D, Calvo KR, Alao H, Kapuria D, Ben-Yakov G, Pichard DC, Hathaway L, Brofferio A, McRae E, Sampaio Moura

- N, Schnappauf O, Rosenzweig S, Heller T, Cowen EN, **Kastner DL**, Ombrello AK (2022) The spectrum of the deficiency of adenosine deaminase 2: an observational analysis of a 60-patient cohort. *Front Immunol* **12**:811473. PMID:35095905.
248. Stone DL, Ombrello A, Arostegui JI, Schneider C, de Jesus A, Girard-Guyonvarc'h C, Gabay C, Lee W, Chae JJ, Aksentijevich I, Goldbach-Mansky RT, **Kastner DL**, Canna SW (2022) Excess serum interleukin-18 distinguishes patients with pathogenic mutations in *PSTPIP1*. *Arthritis Rheumatol* **74**:353-357. PMID:34492165.
249. Saper VE, Ombrello MJ, Tremoulet AH, Montero-Martin G, Prahalad S, Canna S, Shimizu C, Deutsch G, Tan SY, Remmers EF, Monos D, Hahn T, Phadke OK, Cassidy E, Ferguson I, Mallajosyula V, Xu J, Rosa Duque JS, Chua GT, Ghosh D, Szymanski AM, Rubin D, Burns JC, Tian L, Fernandez-Vina MA, Mellins ED, Hollenbach JA; Drug Hypersensitivity Consortium; **INCHARGE Consortium** (2022) Severe delayed hypersensitivity reactions to IL-1 and IL-6 inhibitors link to common HLA-DRB1*15 alleles. *Ann Rheum Dis* **81**:406-415. PMID:34789453.
250. Deutch NT, Yang D, Lee PY, Yu X, Sampaio Moura N, Schnappauf O, Ombrello AK, Stone D, Kuehn HS, Rosenzweig SD, Hoffmann P, Cudrici C, Levy DM, Kessler E, Soep JB, Hay AD, Dalrymple A, Zhang Y, Sun L, Zhang Q, Tang X, Wu Y, Rao K, Li H, Luo H, Zhang Y, Burnham JM, Boehm M, Barron K, **Kastner DL**, Aksentijevich I, Zhou Q (2022) TNF-inhibition in vasculitis management in adenosine deaminase 2 deficiency (DADA2). *J Allergy Clin Immunol* **149**:1812-1816.e6. PMID: 34780847.
251. Romano M, Arici ZS, Piskin D, Alehashemi S, Aletaha D, Barron K, Benseler S, Berard RA, Broderick L, Dedeoglu F, Diebold M, Durrant K, Ferguson P, Foell D, Hausmann JS, Jones OY, **Kastner D**, Lachmann HJ, Laxer RM, Rivera D, Ruperto N, Simon A, Twilt M, Frenkel J, Hoffman HM, de Jesus AA, Kuemmerle-Deschner JB, Ozen S, Gattorno M, Goldbach-Mansky R, Demirkaya E (2022) The 2021 EULAR/American College of Rheumatology Points to Consider for Diagnosis, Management and Monitoring of the Interleukin-1 Mediated Autoinflammatory Diseases: Cryopyrin-Associated Periodic Syndromes, Tumour Necrosis Factor Receptor-Associated Periodic Syndrome, Mevalonate kinase Deficiency, and Deficiency of the Interleukin-1 Receptor Antagonist. *Arthritis Rheumatol* **74**:1102-1121. PMID:35621220.
252. Beck DB, Werner A, **Kastner DL**, Aksentijevich I (2022) Disorders of ubiquitylation: unchained inflammation. *Nat Rev Rheumatol* **18**:435-447. PMID:35523963.
253. Romano M, Arici ZS, Piskin D, Alehashemi S, Aletaha D, Barron KS, Benseler S, Berard R, Broderick L, Dedeoglu F, Diebold M, Durrant KL, Ferguson P, Foell D, Hausmann J, Jones OY, **Kastner DL**, Lachmann HJ, Laxer RM, Rivera D, Ruperto N, Simon A, Twilt M, Frenkel J, Hoffman H, de Jesus AA, Kuemmerle-Deschner JB, Ozen S, Gattorno M, Goldbach-Mansky R, Demirkaya E (2022) The 2021 EULAR/American College of Rheumatology points to consider for diagnosis, management and monitoring of the interleukin-1 mediated autoinflammatory diseases: cryopyrin-associated periodic syndromes, tumour necrosis factor receptor-associated periodic syndrome, mevalonate kinase deficiency, and deficiency of the interleukin-1 receptor antagonist. *Ann Rheum Dis* **81**:907-921. PMID: 35623638.
254. Kozycki CT, Kodati S, Huryh L, Wang H, Warner BM, Jani P, Hammoud D, Abu-Asab MS, Jittayasothorn Y, Mattapallil MJ, Tsai WL, Ullah E, Zhou P, Tian X, Soldatos A, Moutsopoulos N, Kao-Hsieh M, Heller T, Cowen EW, Lee C-C R, Toro C, Kalsi S, Khavandgar Z, Baer A, Beach M, Long Priel D, Nehrebecky M, Rosenzweig S, Romeo T, Deutch N, Brenchley L, Pelayo E, Zein W, Sen N, Yang AH, Farley G, Sweetser DA, Briere L, Yang J, de Oliveira Poswar F, Schwartz I, Silva Alves T, Dusser P, Koné-Paut I, Touitou I, Titah SM, van Hagen PM, van Wijck RTA, van der Spek PJ, Yano H, Benneche A, Apalset EM, Jansson RW, Caspi RR, Kuhns DB, Gadina M, Takada H, Ida H, Nishikomori R, Verrecchia E, Sangiorgi E, Manna R, Brooks BP, Sobrin L, Hufnagel R, Beck D, Shao F, Ombrello AK, Aksentijevich I, **Kastner DL**, Undiagnosed Diseases Network (2022) Gain-of-function mutations in

ALPK1 cause an NF- κ B mediated autoinflammatory disease: functional assessment, clinical phenotyping, and disease course of patients with ROSAH syndrome. *Ann Rheum Dis* **81**:1453-1464. PMID:35868845.

255. Spaan AN, Neehus A-L, Laplantine E, Staels F, Ogishi M, Seeleuthner Y, Rapaport F, Lacey KA, van Nieuwenhove E, Chrabieh M, Hum D, Migaud M, Izmiryan A, Lorenzo L, Kochetkov T, Heesterbeek DAC, Bardoel BW, DuMont AL, Dobbs K, Chardonnet S, Heissel S, Baslan T, Zhang P, Yang R, Bugunovic D, Wunderink HF, Haas P-J A, Molina H, van Buggenhout G, Lyonnet S, Notarangelo LD, Seppänen MRJ, Weil R, Seminario G, Gomez-Tello H, Wouters C, Mesdaghi M, Shahrooei M, Bossuyt X, Sag E, Topaloglu R, Ozen S, Leavis HL, van Eijk MMJ, Bezrodnik L, Blancas Galicia L, Havnanian A, Nassif A, Bader-Meunier B, Neven B, Meyts I, Schrijvers R, Puel A, Bustamante J, Aksentijevich I, **Kastner DL**, Torres VJ, Humblet-Baron S, Liston A, Abel L, Boisson B, Casanova JL (2022) Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal α -toxin. *Science*, epub ahead of print. PMID:35587511.
256. Grayson PC, Beck DB, Ferrada MA, Nigrovic PA, **Kastner DL** (2022) Notes from the field: VEXAS syndrome and disease taxonomy in rheumatology. *Arthritis Rheumatol*, epub ahead of print. PMID: 35696333.
257. Ferrada MA, Savic S, Ospina Cardona D, Collins JC, Alessi H, Gutierrez-Rodrigues F, Uthaya Kumar DB, Wilson L, Goodspeed W, Topilow JS, Paik JJ, Poulter JA, Kermani TA, Koster MJ, Warrington K, Cargo CA, Tattersall RS, Duncan CJ, Hoffmann P, Payne EM, Bonnekoh H, Krause K, Cowen EW, Calvo KR, Patel BA, Ombrello AK, **Kastner DL**, Werner A, Grayson PC, Beck DB. Translation of cytoplasmic UBA1 contributes to VEXAS syndrome pathogenesis. *Blood*, epub ahead of print. PMID: 35793467.
258. Magnotti F, Chirita D, Dalmon S, Martin A, Bronnec P, Sousa J, Helynck O, Lee W, **Kastner DL**, Chae JJ, McDermott MF, Belot A, Popoff M, Sève P, Georgin-Lavialle S, Munier-Lehrmann H, Tran TA, De Langhe E, Wouters C, Jamilloux Y, Henry T (2022) Steroid hormone catabolites activate the pyrin inflammasome through a non-canonical mechanism. *Cell Rep.* **41**:111472. PMID:36223753.
259. Hury LA, Kozycki C, Serpen JY, Zein WM, Ullah E, Iannaccone A, Williams L, Sobrin L, Brooks BP, Sen HN, Hufnagel RB, **Kastner DL**, Kodati S (2022) Ophthalmic manifestations of ROSAH syndrome, an inherited NF- κ B mediated autoinflammatory disease with retinal dystrophy. *Ophthalmology* S01161. PMID:36332842.
260. Mughrabi IT, Ochani M, Tanovic M, Wang P, Diamond B, Sherry B, Pavlov VA, Ozen S, **Kastner DL**, Chae JJ, Al-AbedY (2022) Galantamine attenuates autoinflammation in a mouse model of familial Mediterranean fever. *Mol Med* **28**:148. PMID:36494621.
261. Beck DB, Bodian DL, Shah V, Mirshahi UL, Kim J, Ding Y, Magaziner SJ, Strande NT, Cantor A, Haley JS, Cook A, Hill W, Schwartz AL, Grayson PC, Ferrada MA, **Kastner DL**, Carey DJ, Stewart DR (2023) Estimated prevalence and clinical manifestations of UBA1 variants associated with VEXAS syndrome in a clinical population. *JAMA* **329**:318-324. PMID:36692560.
262. An JW, Pimpale-Chavan P, Stone DL, Bandiera M, Dedeoglu F, Lo J, Bohnsack J, Rosenzweig S, Schnappauf O, Dissanayake D, Hiraki LT, **Kastner DL**, Pelajo C, Laxer RM, Aksentijevich I (2023) Case report: novel variants in *RELA* associated with familial Behçet's-like disease. *Front Immunol* **14**:1127085. PMID:36926348.
263. Soldatos A, Toro C, Hoffmann P, Romeo T, Deutch N, Brofferio A, Aksentijevich I, **Kastner DL**, Ombrello AK (2023) TNF-blockade for primary stroke prevention in adenosine deaminase deficiency: a case series. *Neurol Neuroimmunol Neuroinflamm* **10**:e200073. PMID:36941081.

264. Gutierrez-Rodriguez F, Kusne Y, Fernandez J, Lasho TL, Shalhoub RN, Ma X, Alessi H, Finke CM, Koster MJ, Mangaonkar AA, Warrington KJ, Begna K, Xie Z, Ombrello AK, Viswanatha DS, Ferrada MA, Wilson L, Go RS, Kourelis TV, Reichard KK, Olteanu H, Darden I, Hironaka D, Alemu L, Kajigaya S, Calado RT, Groarke EM, Rosenzweig S, **Kastner DL**, Calvo KR, Wu CO, Grayson PC, Young NS, Beck DB, Patel BA, Patnaik MM (2023) Spectrum of clonal hematopoiesis in VEXAS syndrome. *Blood*, online ahead of print. PMID:37084382.
265. Lee PY, Davidson BA, Abraham RS, Alter B, Arostegui JI, Bell K, Belot A, Bergerson JRE, Bernard TJ, Brogan PA, Berkun Y, Deutch NT, Dimitrova D, Georgin-Lavialle SA, Gattorno M, Grimbacher B, Hashem H, Hershfield MS, Ichord RN, Izawa K, Kanakry JA, Khubchandani RP, Klouwer FCC, Luton EA, Man AW, Meyts I, Van Montfrans JM, Ozen S, Saarela J, Santo GC, Sharma A, Soldatos A, Sparks R, Torgerson TR, Uriarte IL, Youngstein TAB, Zhou Q, Aksentijevich I, **Kastner DL**, Chambers EP, Ombrello AK, for the DADA2 Foundation (2023) Evaluation and management of deficiency of adenosine deaminase 2: an international consensus statement. *JAMA Network Open* **6**:e2315894. PMID:37256629.
266. Baghdassarian H, Blackstone SA, Clay OS, Philips R, Matthiasardottir B, Nehrebecky M, Hua VK, McVicar R, Liu Y, Tucker SM, Randazzo D, Deutch N, Rosenzweig S, Mark A, Sasik R, Fisch KM, Pimpale-Chavan P, Eren E, Watts NR, Ma CA, Gadina M, Schwartz DM, Sanyal A, Werner G, Murdock DR, Horita N, Chowdhury S, Dimmock D, Jepsen K, Remmers EF, Goldbach-Mansky R, Gahl WA, O'Shea JJ, Milner JD, Lewis NE, Chang J, **Kastner DL**, Torok K, Oda H, Putnam CD, Broderick L (2023) Variant *STAT4* and response to ruxolitinib in an autoinflammatory syndrome. *N Engl J Med* **388**:2241-2252. PMID:37256972.
267. Wu Z, Gao S, Gao Q, Patel BA, Groarke EM, Feng X, Manley AL, Li H, Ospina Cardona D, Kajigaya S, Alemu L, Quinones Raffo D, Ombrello AK, Ferrada MA, Grayson PC, Calvo KR, **Kastner DL**, Beck DB, Young NS (2023) Early activation of inflammatory pathways in UBA1-mutated hematopoietic stem and progenitor cells in VEXAS. *Cell Rep Med*. **4**:101160. PMID:37586319.
268. Broderick L, **Kastner DL** (2023) Variant *STAT4* and treatment of an autoinflammatory syndrome. Reply. *N Engl J Med* **389**:1151-1152. PMID:37733321.
269. Luo Y, Ferrada MA, Sikora KA, Rankin C, Alessi HD, **Kastner DL**, Deng Z, Zhang M, Merkel PA, Kraus VB, Allen AS, Grayson PC (2023) Ultra-rare genetic variation in relapsing polychondritis: a whole-exome sequencing study. *Ann Rheum Dis* **83**:253-260. PMID:37918895.
270. Blackstone SA, **Kastner DL**, Broderick L (2023) Autoinflammatory syndromes: updates in management. *J Allergy Clin Immunol* **153**:85-89. PMID:37926121.
271. Kusne Y, Ghorbanzadeh A, Dulau Florea A, Shalhoub RN, Alcedo Andrade PE, Nghiem K, Ferrada MA, Hines A, Quinn KA, Panicker SR, Ombrello AK, Reichard KK, Darden I, Goodspeed W, Durrani J, Wilson L, Olteanu H, Lasho TL, **Kastner DL**, Warrington KJ, Mangaonkar AA, Go RS, Braylan RC, Beck DB, Patnaik MM, Young NS, Calvo KR, Casanegra A, Grayson PC, Koster MJ, Wu CO, Kanthi Y, Patel BA, Houghton DE, Groarke EM (2024) Venous and arterial thrombosis in patients with VEXAS syndrome. *Blood* **143**:2190-2200. PMID:38306657.
272. Lee W, Stone DL, Hoffmann P, Rosenzweig S, Tsai WL, Gadina M, Romeo T, Lee CR, Randazzo D, Pimpale Chavan P, Manthiram K, Canna S, Park YH, Ombrello AK, Aksentijevich I, **Kastner DL**, Chae JJ (2024) Interrupting an IFN- γ -dependent feedback loop in the syndrome of pyogenic arthritis with pyoderma gangrenosum and acne. *Ann Rheum Dis* **83**:787-798. PMID: 38408849.
273. Oda H, Manthiram K, Pimpale Chavan P, Reiser E, Öney V, Kaya Ö, Rauch C, Nakabo S, Kuehn HS, Swart M, Wang Y, Çelik NI, Xu Q, Preite S, Ziaee V, Movahedi N, Shahrooei M, Parvaneh N, Alipour-olyei, Molitor A, Carapito R, Beck DB, Chae JJ, Nehrebecky M, Ombrello AK, Romeo T, Deutch N,

Matthiasardóttir B, Mullikin J, Komarow H, Stoddard J, Niemela J, Dobbs K, Sweeney CL, Anderton H, Lawlor KE, Yoshitomi H, Yang D, Boehm M, Davis J, Mudd P, Randazzo D, Tsai WL, Gadina M, Kaplan MJ, Toguchida J, Mayer CT, Rosenzweig SD, Notarangelo LD, Iwai K, Silke J, Schwartzberg PL, Boisson B, Casanova J-L, Seiamak B, Rao A, Peltzer N, Walczak H, Lalaoui N, Aksentijevich I, **Kastner DL** (2024) Biallelic human SHARPIN loss-of-function induces autoinflammation and immunodeficiency. *Nat Immunol* **25**:764-777. PMID: 38609546.

274. Kozycki C, **Kastner D**, Huryl L, Kodati S, Warner BM (2024) *ALPK1*-related autoinflammatory disease. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. PMID: 38935811.
275. Correia Marques M, Rubin D, Shuldiner EG, Datta M, Schmitz E, Gutierrez Cruz G, Patt A, Bennett E, Grom A, Foell D, Gattorno M, Bohnsack J, Yeung RSM, Prahalad S, Mellins E, Anton J, Len CA, Oliveira S, Woo P, Ozen S; INCHARGE Consortium; Deng Z, Ombrello MJ (2024) Enrichment of rare variants of hemophagocytic lymphohistiocytosis genes in systemic juvenile idiopathic arthritis. *Arthritis Rheumatol*, online ahead of print. PMID: 38937141.
276. Sparks R, Rachmaninoff N, Lau WW, Hirsch DC, Bansal N, Martins AJ, Chen J, Liu CC, Cheung F, Failla LE, Biancotto A, Fantoni G, Sellers BA, Chawla DG, Howe KN, Mostaghimi D, Farmer R, Kotliarov Y, Calvo KR, Palmer C, Daub J, Foruraghi L, Kreuzburg S, Treat JD, Urban AK, Jones A, Romeo T, Deutch NT, Moura NS, Weinstein B, Moir S, Ferrucci L, Barron KS, Aksentijevich I, Kleinstein SH, Townsley DM, Young NS, Frischmeyer-Guerrero PA, Uzel G, Pinto-Patarroyo GP, Cudrici CD, Hoffmann P, Stone DL, Ombrello AK, Freeman AF, Zerbe CS, **Kastner DL**, Holland SM, Tsang JS. A unified metric of human immune health. *Nat Medicine*, online ahead of print. PMID: 38961223.

Book Chapters

1. Steinberg AD, Klinman DM, **Kastner DL**, Seldin MF, Gause WC, Scribner CL, Britten JL, Siegel JN, Mountz JD (1987) Genetic and molecular genetic studies of murine and human lupus, in *Proceedings of First International Symposium on SLE, J Rheumatol (Supplement 13)* **14**:166-176.
2. Steinberg AD, Krieg AM, Gause WC, **Kastner DL**, Mountz JD, Klinman DM (1988) Towards an understanding of lupus, in *Cellular Basis of Immune Modulation, Proceedings of the Nineteenth International Leukocyte Culture Conference, Banff*, JG Kaplin and DR Green, eds., Alan R. Liss Co., pp. 423-442.
3. **Kastner DL**, McIntyre TM, Mallett CP, Hartman AB, Steinberg AD (1989) Studies of the murine immunoglobulin V_H repertoire by *in situ* hybridization in unstimulated spleen cells, in *The Immune Response to Structurally Defined Proteins*, S Smith-Gill and E Sercarz, eds., Adenine Press, Schenectady, New York, pp. 291-301.
4. Steinberg AD, Klinman DM, McIntyre TM, **Kastner DL** (1989) Cellular and molecular basis of SLE. I. Genesis of autoantibodies, in *Proceedings of the Second International Conference on Systemic Lupus Erythematosus*, Professional Postgraduate Services, International, Singapore, pp. 15-18.
5. **Kastner DL** (1997) Intermittent and periodic arthritic syndromes, in *Arthritis and Allied Conditions*, 13th ed., WJ Koopman, ed., Williams and Wilkins, Baltimore, pp.1279-1306.
6. Pras E, Livneh A, Balow JE Jr, Pras E, **Kastner DL**, Pras M, Langevitz P (1997) Clinical heterogeneity in familial Mediterranean fever, in *Familial Mediterranean Fever*, E Sohar, J Gafni, and M Pras, eds., Freund, London and Tel Aviv, pp. 62-65.

7. Lotan R, Danon Y, Magal N, Ogur G, Tokguz G, Schwabe A, Fischel-Ghodsian N, **Kastner D**, Rotter JI, Schlezinger M, Shohat M (1997) Frequencies of the various FMF core haplotypes in the *MEFV* locus, *ibid.*, pp. 104-108.
8. Shohat M, Lotan R, Magal N, Ogur G, Tokguz G, Schwabe A, Fischel-Ghodsian N, **Kastner D**, Rotter JI, Schlezinger M, Danon Y (1997) Association between ancestral haplotype in the *MEFV* locus and FMF-amyloidosis, *ibid.*, pp. 115-119.
9. Pras E, Aksentijevich I, Balow JE Jr, Pras M, **Kastner D** (1997) Lack of evidence for an increase in survival rate among heterozygote sibs from families with familial Mediterranean fever, *ibid.*, pp. 120-121.
10. Zaks N, **Kastner DL** (1997) Clinical syndromes resembling familial Mediterranean fever, *ibid* pp. 211-215.
11. Deng Z, **Kastner DL**, and the International FMF Consortium (1997) Genomic structure and sequence analysis of the FMF gene and its protein product, *ibid.*, pp. 239-245.
12. Aksentijevich I, **Kastner DL**, and the International FMF Consortium (1997) Microsatellite haplotypes and *MEFV* mutations: exploring the genealogy of FMF, *ibid.*, pp. 246-251.
13. Centola M, **Kastner DL**, and the International FMF Consortium (1997) Cloning of *MEFV*: implications for the pathophysiology of familial Mediterranean fever, *ibid.*, pp. 252-259.
14. Pras E, Langevitz P, Livneh A, Zemer D, Migdal A, Padeh S, Lubetzky A, Aksentijevich I, Centola M, Zaks N, Deng Z, Sood R, **Kastner DL**, Pras M (1997) Genotype-phenotype correlation in familial Mediterranean fever (a preliminary report), *ibid.*, pp. 260-264.
15. Eisenberg S, Urieli-Shoval S, Azar Y, Centola M, Deng Z, **Kastner DL**, and Matzner Y (1997) C5a inhibitor and pyrin/marenostrin: possible relationship, *ibid.*, pp. 275-278.
16. **Kastner DL**, and the International FMF Consortium (1997) Positional cloning of the gene causing familial Mediterranean fever: an overview, *ibid.*, Suppl., pp. 3-11.
17. Pras M, **Kastner DL** (1997) Familial Mediterranean fever, in *Rheumatology*, 2nd ed., JH Klippel and PA Dieppe, eds., Mosby, St. Louis, pp. 5.23.1-5.23.4.
18. **Kastner DL**, O'Shea JJ, Aksentijevich I (1999) Molecular basis of the hereditary periodic fever syndromes, in *Harrison's Online*, AS Fauci, E Braunwald, KJ Isselbacher, DL Kasper, SL Hauser, DL Longo, JL Jameson, eds., McGraw Hill, New York.
19. **Kastner DL** (2001) Intermittent and periodic arthritic syndromes, in *Arthritis and Allied Conditions*, 14th ed., WJ Koopman, ed., Williams and Wilkins, Baltimore, pp. 1400-1441.
20. **Kastner DL** (2001) Periodic syndromes, in *Primer on the Rheumatic Diseases*, 12th ed., JH Klippel, LJ Crofford, JH Stone, CM Weyand, eds., Arthritis Foundation, Atlanta, pp. 194-201.
21. Hull KM, **Kastner DL** (2003) Hereditary recurrent fever syndromes [Perspectives, Contemporary Topics]. MD Consult Infectious Disease. Available at: <http://www.mdconsult.com>.
22. Hull KM, **Kastner DL** (2003) Rheumatology and signal transduction, in *Signal Transduction in Human Disease*, T Finkel, S Gutkind, eds., John Wiley and Sons, Hoboken, pp. 357-375.
23. **Kastner DL** (2003) The hereditary periodic fevers, in *Rheumatology*, 3rd ed., MC Hochberg, AJ Silman, JS Smolen, ME Weinblatt, MH Weisman, eds., pp. 1717-1734.

24. Morinobu A, Remmers E, **Kastner D**, O'Shea JJ (2003) Principles and techniques in molecular biology, in *Rheumatology*, 3rd ed., MC Hochberg, AJ Silman, JS Smolen, ME Weinblatt, MH Weisman, eds., pp. 85-97.
25. **Kastner DL** (2004) The systemic autoinflammatory diseases, in *Medical Knowledge Self-Assessment Program 13 Rheumatology*, ED Harris, Jr., ed., pp. 52-57.
26. **Kastner DL** (2005) Familial Mediterranean fever and other hereditary recurrent fevers, in *Harrison's Principles of Internal Medicine, Sixteenth Edition*, DL Kasper, E Braunwald, AS Fauci, SL Hauser, DL Longo, JL Jameson, eds., pp. 1793-1795.
27. **Kastner DL**, Aksentijevich I (2005) Intermittent and periodic arthritis syndromes, in *Arthritis and Allied Conditions*, 15th ed., WJ Koopman and LW Moreland, eds., pp. 1411-1461.
28. Brydges S, Athreya B, **Kastner DL** (2005) Periodic fever syndromes in children, in *Textbook of Pediatric Rheumatology, Fifth Edition*, JT Cassidy and RE Petty, eds., pp. 657-670.
29. Brydges S, Kastner DL (2006) The systemic autoinflammatory diseases: inborn errors of the innate immune system, in *Current Topics in Microbiology and Immunology*, vol. 305, A. Radbruch and P.E. Lipsky, eds., pp. 127-160.
30. **Kastner DL**, Brydges S, Hull KM (2007) Periodic fever syndromes, in *Primary Immunodeficiency Diseases: A Molecular and Genetic Approach, Second Edition*, HD Ochs, CIE Smith, JM Puck, eds., pp. 367-389.
31. Ryan JG, **Kastner DL** (2007) Periodic syndromes, in *Primer on the Rheumatic Diseases, 13th ed.*, JH Klippel, JH Stone, LJ Crofford, PH White, eds., pp. 460-469.
32. **Kastner DL** (2007) The systemic autoinflammatory diseases, in *The Cecil Textbook of Medicine, 23rd Edition*, D Ausiello, L Goldman, eds., pp. 1988 – 1993.
33. Remmers EF, Kanno Y, Siegel RM, **Kastner DL**, O'Shea JJ (2008) Principles and techniques in molecular biology, in *Rheumatology*, 4th ed., MC Hochberg, AJ Silman, JS Smolen, ME Weinblatt, MH Weisman, eds., pp. 103-116.
34. Colburn NT, **Kastner DL** (2008) The hereditary periodic fevers, in *Rheumatology*, 4th ed., MC Hochberg, AJ Silman, JS Smolen, ME Weinblatt, MH Weisman, eds., pp. 1619-1640.
35. **Kastner DL** (2008) Familial Mediterranean fever, in *Harrison's Principles of Internal Medicine, 17th ed.*, AS Fauci, DL Kasper, E Braunwald, SL Hauser, DL Longo, JL Jameson, J Loscalzo, eds., pp. 2142-2145.
36. Ryan JG, **Kastner DL** (2008) The systemic autoinflammatory diseases, in *Fitzpatrick's Dermatology in General Medicine, 7th ed.*, K Wolff, LA Goldsmith, SI Katz, B Gilchrist, AS Paller, DJ Leffell, eds., pp. 1265-1271.
37. Barron K, Athreya B, **Kastner DL** (2011) Periodic fever syndromes and other inherited autoinflammatory diseases, in *Textbook of Pediatric Rheumatology, Sixth Edition*, JT Cassidy, RE Petty, RM Laxer, CB Lindsley, eds., pp. 642-660.
38. Remmers EF, Ombrello MJ, Kanno Y, Siegel RM, **Kastner DL** (2011) Principles and techniques in molecular biology, in *Rheumatology*, 5th ed., MC Hochberg, AJ Silman, JS Smolen, ME Weinblatt, MH Weisman, eds., pp. 105-118.

39. Kim PW, Aksentijevich I, Colburn NT, **Kastner DL** (2011) The hereditary recurrent fevers, in *Rheumatology*, 5th ed., MC Hochberg, AJ Silman, JS Smolen, ME Weinblatt, MH Weisman, eds., pp. 1637-1657.
40. **Kastner DL** (2011) Systemic autoinflammatory diseases, in *Goldman's Cecil Medicine*, 24th ed., L Goldman, A Schafer, eds., pp. 1667-1672.
41. **Kastner DL** (2012) Familial Mediterranean fever and other hereditary recurrent fevers, in *Harrison's Principles of Internal Medicine*, 18th ed., AS Fauci, DL Kasper, E Braunwald, SL Hauser, DL Longo, JL Jameson, J Loscalzo, eds., pp. 2814-2817.
42. Broderick L, **Kastner DL**, Hoffman HM (2014) Recurrent fever syndromes, in *Primary Immunodeficiency Diseases: A Molecular and Genetic Approach, Third Edition*, HD Ochs, CIE Smith, JM Puck, eds., pp. 414-447.
43. Goldbach-Mansky R, de Jesus AA, McDermott MF, Kastner DL (2015) Monogenic autoinflammatory diseases, in *Rheumatology*, 6th ed., MC Hochberg, AJ Silman, JS Smolen, ME Weinblatt, MH Weisman, eds., pp. 1369-1391.
44. **Kastner DL** (2015) Familial Mediterranean fever and other hereditary recurrent fevers, in *Harrison's Principles of Internal Medicine*, 19th ed., DL Kasper, AS Fauci, SL Hauser, DL Longo, JL Jameson, J Loscalzo, eds., pp. 2212-2215.
45. Chae JJ, **Kastner DL** (2015) Pathogenesis, in *Familial Mediterranean Fever*, M Gattorno, ed., pp. 13-30.
46. Siegel RM, **Kastner DL** (2016) The systemic autoinflammatory diseases, in *Goldman's Cecil Medicine*, 25th ed., L Goldman, A Schafer, eds., pp. 1739-1744.
47. Ombrello AK, **Kastner DL** (2016) Hereditary periodic fever syndromes and other systemic autoinflammatory diseases, in *Nelson Textbook of Pediatrics*, 20th ed., RM Kliegman, BF Stanton, JW St. Geme, NF Schor, eds. pp. 1193-1204.
48. Barron KS, **Kastner DL** (2016) Periodic fever syndromes and other inherited autoinflammatory diseases, in *Textbook of Pediatric Rheumatology, Seventh Edition*, RE Petty, RM Laxer, CB Lindsley, LR Wedderburn, eds., pp. 609-626.
49. **Kastner DL** (2018) Familial Mediterranean fever and other hereditary autoinflammatory diseases, in *Harrison's Principles of Internal Medicine*, 20th ed., JL Jameson, AS Fauci, DL Kasper, SL Hauser, DL Longo, J Loscalzo, eds., pp. 2610-2614.
50. de Jesus AA, McDermott MF, **Kastner DL**, Goldbach-Mansky R (2019) Monogenic autoinflammatory diseases, in *Rheumatology* 7th ed., MC Hochberg, EM Gravallese, AJ Silman, JS Smolen, ME Weinblatt, MH Weisman, eds., pp. 1445-1469.
51. **Kastner DL** (2019) Autoinflammation: past, present, and future, in *Textbook of Autoinflammation*, PL Hashkes, RM Laxer, A Simon, eds., pp. 3-15.
52. **Kastner DL** (2019) Foreword, in *Auto-inflammatory syndromes: pathophysiology, diagnosis, and management*. P Efthimiou, ed., pp. vii-x.
53. Stone DL and **Kastner DL** (2019) Pyogenic arthritis, pyoderma gangrenosum, and acne (PAPA) syndrome, in *Encyclopedia of Medical Immunology*, I Mackay and NR Rose, eds., https://doi.org/10.1007/978-1-4614-9209-2_128-1.

54. **Kastner DL** (2020) Foreword, in *Periodic and Non-Periodic Fevers (Rare Diseases of the Immune System)*, R Cimaz, ed., pp. v-vi.
55. Siegel RM, **Kastner DL** (2020) The systemic autoinflammatory diseases, in *Goldman's Cecil Medicine, 26th ed.*, L. Goldman, AI Schafer, eds., pp. 1691-1697.
56. Barron KS, **Kastner DL** (2021) Periodic fever syndromes and other inherited autoinflammatory diseases, in *Textbook of Pediatric Rheumatology, Eighth Edition*, RE Petty, RM Laxer, CB Lindsley, LR Wedderburn, eds., pp. 525-543.
57. **Kastner DL** (2022) Familial Mediterranean fever and other hereditary autoinflammatory diseases, in *Harrison's Principles of Internal Medicine, 21st ed.*, J Loscalzo, AS Fauci, DL Kasper, SL Hauser, DL Longo, JL Jameson, eds., pp. 2840-2844.
58. de Jesus A, McDermott MF, **Kastner DL**, Goldbach-Mansky R (2023) Monogenic autoinflammatory diseases, in *Rheumatology 8th ed.*, MC Hochberg, EM Gravallese, JS Smolen, D van der Heijde, ME Weinblatt, MH Weisman, eds., pp. 1519-1546.
59. **Kastner DL** (2024) The systemic autoinflammatory diseases, in *Goldman-Cecil Medicine, 27th edn.*, L Goldman, ed., *in press*.
60. Hansen SKE, **Kastner DL**, Hashkes PJ (2024) Autoinflammatory diseases, in *Oxford Textbook of Rheumatology, 5th edn.*, RA Watts, ed., *in press*.
61. **Kastner DL** (2025) Familial Mediterranean fever and other hereditary autoinflammatory diseases, in *Harrison's Principles of Internal Medicine, 22nd ed.*, DL Longo, AS Fauci, DL Kasper, SL Hauser, JL Jameson, J Loscalzo, SM Holland, CA Langford, eds.