

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 Musculo-
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-present Clinical Director and Director of Translational Research, Intramural Research
Program, NIAMS/NIH
- 2008-present Deputy Director for Intramural Clinical Research, NIH
- 2009-present Chief, Laboratory of Clinical Investigation, NIAMS/NIH

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 membership in 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 Lectureship in Rheumatology
University of Texas Southwestern Medical School
- 2005 Election to membership in the Association of American Physicians
- 2005 ACR Distinguished Investigator Award
American College of Rheumatology
- 2006 Edward W. Boland Lectureship 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 Lectureship
University of Washington School of Medicine
- 2009 Herman Beerman Lectureship
Society for Investigative Dermatology
- 2009 NIH Astute Clinician Lectureship
- 2010 Election to membership in the National Academy of Sciences

Patents

- 2003 U.S. Patent 6,627,745 B1
Daniel L. Kastner et al., Pypin 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

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
 National Center for Human Genome Research

1995- Member, NIAMS Promotions Panel

1997- 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- 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
- 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-present Chair, NIH Intramural Clinical Research Steering Committee
- 2010 Member, Scientific Advisory Board, Scleroderma Research Foundation

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.
2. Rich SS, Rich RR, **Kastner DL** (1978) Suppressor factor activity in cell-mediated immune responses. *J Reticuloendothel Soc* **24**:417-425.
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.
4. **Kastner DL**, Rich RR (1979) *H-2*-nonrestricted cytotoxic responses to an antigen encoded telomeric to *H-2D*. *J Immunol* **122**:196-201.
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.
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.
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.
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.
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.

10. **Kastner DL**, Steinberg AD (1988) Determinants of B cell hyper-activity in murine lupus. *Concepts in Immunopathology* **6**:22-88.
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.
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.
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.
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.
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.
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.
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.
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.
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.
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.

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.
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.
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.
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 γ chain micro-satellite (*D7S485*) to rheumatoid arthritis in multiplex families. *J Autoimmunity* **8**:131-138.
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.
26. Pras M, Pras E, **Kastner D** (1995) The origin of the FMF gene. *Isr J Med Sci* **31**:503-4.
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.
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.
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.
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.
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.
 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.
 34. Sood R, Blake T, Akseptijevich 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.
 35. Balow JE Jr, Shelton DA, Orsborn A, Mangelsdorf M, Akseptijevich 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.
 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.
 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.
 38. Pras E, Golomb E, Drake C, Akseptijevich 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.
 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.
 40. **Kastner DL** (1998) Familial Mediterranean fever: the genetics of inflammation. *Hospital Practice* **33**:131-158.

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.
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.
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.
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.
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.
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.
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.
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.
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.
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.
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.
 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.
 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.
 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.
 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.
 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.
 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 (b⁰⁺ AT) of rBAT. *Nature Genet* **23**:52-57.
 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.
 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.
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.
 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.
 62. Galon J, Aksentijevich I, McDermott MF, O'Shea JJ, **Kastner DL** (2000) *TNFRSF1A* mutations and autoinflammatory syndromes. *Curr Opin Immunol*, **12**:479-486.
 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.
 64. Hooper JD, Bowen N, Marshall H, Cullen LM, Sood R, Daniels R, Stuttgen 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.
 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.
 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.
 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.
 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.

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.
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.
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.
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.
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.
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.
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.
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