

JUDITH L. FRIDOVICH-KEIL, PhD
 Professor, Department of Human Genetics
 Emory University School of Medicine

ADDRESS

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Birthplace Durham, North Carolina
 Citizenship USA

ACADEMIC APPOINTMENTS

2005-present Professor (with tenure), Department of Human Genetics
 Emory University School of Medicine

1998- 2005 Associate Professor (with tenure), Department of Human Genetics
 Emory University School of Medicine

1993-1998 Assistant Professor (tenure track), Department of Genetics
 (and Molecular Medicine) Emory University School of Medicine

1992-1993 Assistant Professor (tenure track), Division of Medical Genetics
 Department of Pediatrics, Emory University School of Medicine

1991-1992 Instructor, Division of Medical Genetics
 Department of Pediatrics, Emory University School of Medicine

BOARD CERTIFICATION

1996- present Diplomate, Clinical Molecular Genetics
 American Board of Medical Genetics

EDUCATION

1988 Ph.D. in Biology
 Massachusetts Institute of Technology, Cambridge MA

1983 AB in Biochemistry (Summa Cum Laude)
 Princeton University, Princeton NJ

POSTGRADUATE TRAINING

1989-91 Training Fellow in Clinical Molecular Genetics
 Harvard Medical School

1988-91 Postdoctoral Research Fellow in Cell Growth and Regulation
 Harvard Medical School and Dana-Farber Cancer Institute

COMMITTEE MEMBERSHIP**National/International**

2009- present Study Section Member, National Institutes of Health Study
 Therapeutic Approaches to Genetic Diseases (TAG)

2010- 2012 Reviewer and Panelist, Howard Hughes Medical Institute
 Medical Fellows Program

2010 Expert Reviewer, Health Research Board, Ireland

2009 Grants Reviewer, Parents of Galactosemic Children, Inc.

2003- 2006 Study Section Member, National Institutes of Health Study
 Special Emphasis Panel on Gene Therapy and Inborn Errors

2004-2006 Member, Information and Education Committee,
 American Society of Human Genetics

2003 Member, Telethon Peer Review System
 Comitato Telethon Fondazione ONLUS, Italy

2003 Ad Hoc Study Section Member, National Institutes of Health

	Medical Biochemistry
Oct 2002	Ad Hoc Study Section Member, National Science Foundation MCB Section on Biochemistry of Gene Expression
Oct 2002	Ad Hoc Study Section Member, National Institutes of Health Medical Biochemistry
Apr 2002	Ad Hoc Study Section Member, National Science Foundation MCB Section on Biochemistry of Gene Expression
Jan 2002	Ad Hoc Study Section Member, American Cancer Society Study Section on Molecular and Cell Biology of Cancer

Regional (non-Emory)

2012- present	Selection Committee, Howard Hughes Medical Institute / Morehouse School of Medicine Medical Student Summer Research Fellowship Program
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Institutional (Emory University/ School of Medicine)

2007-present	Member, Emory Univ. Medical School Admission Committee (Associate Member 1997-2006, full member since 2007)
1997-present	Interviewer, MD/ PhD Program Admissions
2001-03, 05-07, 12-pres	Chair, Faculty Recruiting Committee, Dept. of Human Genetics
2009-2010	Member, Emory University Senate and Faculty Council
2005-2010	Member, School of Medicine (SOM) Faculty Committee on Appointments and Promotions
2007-2008	Member, SOM Institutional Setting Committee (in preparation for 2008 LCME Site Visit)
2002-2008	Member, Editorial Advisory Board of <i>the Academic Exchange</i>
2005	Co-chair, Application of Medical Sciences Sub-committee of the SOM Curriculum Planning Committee
2002-2005	Member, Institutional Review Board (IRB)
2002-2003	Member, S.O.M. Committee to recruit a new Chair of Gyn/Ob
2001-2002	Member, S.O.M. Search Committee for a new Director of the Health Sciences Center Library
2000-2005	Mentor, Passages Program (mentoring junior faculty)
1998-2002	Member, S.O.M. Executive Curriculum Committee
1999	Member, S.O.M. Committee on Teaching Skills
1998-1999	Member, S.O.M. Committee on the Status of Women
1998	Member, Teaching and Faculty Subcommittee S.O.M. Teaching and Education Steering Committee
1995-1999	Coordinator, Genetics Department Seminar Series

EDITORIAL RESPONSIBILITIES

2003-2008	Editorial Board Member, <i>Journal of Biological Chemistry</i>
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Manuscript reviewer (1988-present)

American Journal of Human Genetics, Biochemistry, BioTechniques, Cellular and Molecular Life Sciences, Experimental Cell Research, FEMS Yeast Research, Genetics, Genomics, Human Molecular Genetics, Human Mutation, Journal of Biological Chemistry, Journal of Inherited Metabolic Disease, Molecular and Cellular Biology, Molecular Genetics and Metabolism, Nucleic Acids Research, Pediatric Research, Proceedings of the Natl. Acad. of Sciences USA, Wiener klinische Wochenschrift (a European journal)

HONORS and AWARDS

2011	One in One Hundred Mentorship Award (Emory University)
1991	Inclusion in Marquis Who's Who in Science and Engineering
1990	Aid for Cancer Research Postdoctoral Fellowship

1986	American Association of Univ. Women Dissertation Fellowship
1983	National Science Foundation Graduate Fellowship
1983	Phi Beta Kappa (Princeton University)
1979	National Merit Scholarship

SOCIETY MEMBERSHIPS

American College of Medical Genetics, Founding Fellow
 American Society of Human Genetics

ORGANIZATION OF NATIONAL/ INTERNATIONAL CONFERENCES

2003 American Society of Human Genetics, Annual Meeting
 Invited Speakers Session "Yeast and Human Genetic Disease"

RESEARCH FOCUS

The focus of our research is galactose metabolism and the role(s) galactose metabolites play in normal development and disease. Specifically, we are working to understand the role(s) of galactose metabolism in normal embryogenesis and homeostasis using a fly model system, and the pathophysiology of transferase- and epimerase-deficiency galactosemias using a combination of model systems and patient studies. We are also working to identify modifiers of outcome in galactosemia. Finally, we are exploring novel strategies for improved intervention in galactosemia.

GRANT AWARDS

Current

P.I., National Institutes of Health, "Bases of pathophysiology in galactosemia." R01 DK059904, 08/01/01-06/30/13 (NCE)
 P.I., National Institutes of Health, "A *D. melanogaster* model of galactosemia." R01 DK046403, 08/01/93-04/30/13 (NCE)

Pending

P.I., National Institutes of Health, "A *D. melanogaster* model of classic galactosemia." R01 DK046403 (competitive renewal)
 P.I., National Institutes of Health, "Bases of pathophysiology in galactosemia." R01 DK059904 (competitive renewal)
 P.I., National Institutes of Health, "Outcomes in Duarte Galactosemia." New R03 application.

Previous

P.I., subcontract with the University of Texas Health Science Center at San Antonio (National Newborn Screening and Genetics Resource Center) as part of a cooperative agreement with the Health Resources and Services Administration (Grant Number U32MC 00148, PI Therrell), "Toward evidence-based recommendations for the management of Duarte galactosemia" January 2012 - May 2012.
 P.I., Parents of Galactosemic Children, Inc "Improved intervention for galactosemia" 2008-9
 P.I., National Institutes of Health, "Molecular Studies of Human Enzymes Underlying Galactosemia." RO1 DK046403, 2002-07
 P.I., National Institutes of Health, "Studies of Galactose Toxicity in Yeast and Human Cells." RO1 DK059904, 2002-2006
 P.I., Emory University Research Committee, "Functional analysis of Scp160p: a polyribosome-associated multiple KH domain protein in yeast." \$30K, 2005-06
 P.I., National Science Foundation, "Functional analysis of Scp160p: a polyribosome-associated multiple KH domain protein in yeast." \$345K. total costs 2001-04.
 P.I., National Institutes of Health, "Molecular Studies of Human Enzymes Underlying Galactosemia." RO1 DK046403, \$529,035 total direct costs, 1998-2002
 P.I., Emory University Research Committee, "Functional analysis of the yeast protein Scp160p." \$30K, 02/01/01- 08/31/01.

- P.I., National Institutes of Health, "Molecular studies of FMRP function in yeast." PO1 component (Dr. Steve Warren, Program Director for "A Program of Investigation into Fragile X Syndrome") \$256,918 total direct costs, 1997-2000
- P.I., Emory University Research Committee, "Molecular studies of Scp160p, a candidate yeast homologue of the FMRP protein associated with Fragile X syndrome", \$15K declined 6/97 when overlapping NIH funds secured
- P.I., Batten Disease Support and Research Association, "Functional analysis of *yCLN3*, a yeast homologue of the human *CLN3* gene underlying Batten disease." \$10K, 1996- 97.
- P.I., Emory University Research Committee, "Structure/function analysis of dimer formation and subunit interaction in the human enzyme galactose-1-phosphate uridylyltransferase: studies using a yeast-based cotransformation expression system." \$15K, 1994- 95.
- P.I., National Institutes of Health, "Molecular analysis of human gal-1-P uridylyltransferase." R29 DK046403, \$350K total direct costs, 1993- 98.
- P.I., Emory University Research Committee, "Structure/function analysis of the human galactose-1-phosphate uridylyltransferase sequence: mutagenesis and selection using a yeast-based expression system." \$10K, 1992-93.
- P.I., Emory Children's Research Center, "Molecular analysis of variant alleles of human galactose-1-phosphate uridylyltransferase." \$23K, 1992- 93.

TEACHING

Administrative roles (at Emory)

- Graduate Program in Biochemistry, Cell and Dev Biology
- | | |
|-----------|-----------------------------------|
| 2008-2011 | Member, Qualifying Exam Committee |
| 2008-2009 | Member, Rotations Committee |
| 1998-2000 | Program Director |
| 1997-1998 | Director of Graduate Studies |
| 1995-1997 | Member, Executive Committee |
- Graduate Program in Genetics and Molecular Biology
- | | |
|--------------|---|
| 2011-present | Member, Executive Com and Chair, Orals Exam Committee |
| 2010-2012 | Member, Graduate Recruiting Committee |
| 2010-2012 | Member, Curriculum Committee |
| 2009-2011 | Member, Orals Exam Committee |
| 1994-1996 | Member, Executive Committee |

Classroom Teaching

Medical Students (at Emory)

- M1 Foundation: Human Genetics and Evolution
- | | |
|--------------|----------------------|
| 2007-present | Teacher/ Facilitator |
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- Human and Molecular Genetics (MEDI 545, IBS 505)
- | | |
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| 2002-2007 | Lecturer, facilitator, and coordinator for clinical correlations |
| 1994-2001 | Course Director, lecturer and facilitator |
| 1992-1993 | Lecturer and facilitator |
- Medical Biochemistry (MEDI 515, IBS 528)
- | | |
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| 1997, 2004-2005 | Lecturer |
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- Human Embryology: Development and Disease (MEDI 510, IBS 518)
- | | |
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| 1993 | Lecturer |
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PhD Students (at Emory)

- Graduate Human Genetics (IBS746)
- | | |
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| 2003- present | Course Director and lecturer |
|---------------|------------------------------|
- Principles of Basic Biomedical and Biological Sciences (IBS555/556, replaced IBS520/501)
- | | |
|---------------|---|
| 2008- present | Genetics Block Coordinator and Lecturer |
| 2004- 2007 | Lecturer |
- Foundations of Biochemistry, Cell, and Developmental Biology (BCDB 501)
- Molecular Approaches to Biological Problems

2011- present Mini-course co-director
(with Dave Katz 2011 and with Grace Pavlath 2012)

Values in Science (for GMB students)
2011, 2012 Course Co-Director and lecturer

Glycoscience (IBS500R)
2010, 2012 Lecturer

Genetic Model Systems (IBS560)
1995, 2003-present Lecturer

Pathophysiology (BMED8121/6793 joint Emory/ Georgia Tech program)
2004-present Lecturer

Teaching Assistant Training and Teaching Opportunity (TATTO)
2007-present Lecturer

Values in Science (IBS606)
2005- 2009 Lecturer and/or faculty participant

Responsible Conduct in Science (GMB)
2004-05 Course Co-director and lecturer

Hypothesis design and scientific writing, BCDB "Grants class" (IBS522)
2004 Lecturer
2005, 2006, 2009, 2010 Study Section member

Introductory Biochemistry and Molecular Biology (IBS520, course renamed IBS555 in 2004)
1993-2003 Lecturer

Introduction to Laboratory Methods (IBS 559)
2000 Course Director and lecturer

Molecular Approaches to Questions of Structure and Function (IBS 706)
1994-1997 Course Director and lecturer

BMB Graduate Student Seminar Series (BMB570R/ BMB790)
1994-1996 Course Director and lecturer

RNA binding proteins (GMB570R)
1995 Course Co-director and lecturer

Introductory Graduate Cell Biology (IBS 501)
1994 Lecturer

Residents and postdocs (at Emory)

Postdoc Ethics
2009-present facilitator for "Responsible Authorship" section

Pediatric Chairman's Conference, Grady Hospital
1997 Lecturer

Pediatric Fellows Applied Basic Science Course
1992 Lecturer

Other Teaching (local or national)

Galactosemia Foundation Continuing Medical Education Course 2012 sponsored by the University of Texas Health Sciences Center (at San Antonio)
2012 Course Director and Lecturer

Health Sciences Workshop for High School Teachers (organized by the Center for Science Education at Emory)
2010 Lecturer (on Human Genetics)

National Youth Leadership Forum on Medicine (NYLF-MED) Summer Program
2003-04 Lecturer on "Careers in Human Genetics"

Research Coordinators' Meetings for Studies Involving Human Subjects
06/2003 Lecturer on "Genetic Studies and Informed Consent" (Emory)
07/2003 Lecturer on "Genetic Studies and Informed Consent" (VA)

CME Course for the Society for Gynecological Investigation
1998 Lecturer on "How to Find a Gene and Beyond"

Harvard University Extension School, "Human Genetics"

1988-1991 Lecturer
 Harvard University Extension School, "Principles of Genetics"
 1991 Course Co-director and lecturer
 1988-1990 Lecturer

Supervisory Teaching

Postdoctoral Trainees

Current Ying Liu, PhD (2010-present)
 Brook Pyhtila, PhD (2012-present)
 Emily Ryan, PhD (2013-present)
 Jenna Daenzer, PhD (2013-present)

Past Patricia P Jumbo Lucioni, PhD (2009-2012)
 Kerry (Ross) Garza, PhD (2002-2009)
 Ai-min Li, MD PhD (2000-2004)
 Jane Odhiambo Mumma, PhD (2005-2006)
 Weining Tang, PhD (2004-2006)

PhD Dissertation Advisees

Current Kelly A. Shaw [GMB, rotation student 2012]
 Emily A. Nickoloff [BCDB, rotation student 2012]

Past J. Patrick Elsevier [BMB, Ph.D. 1996]
 B. Booth Quimby [GMB, Ph.D. 1997]
 R. Lance Wells [BMB, Ph.D. 1998]
 Travis M. Wohlers [GMB, Ph.D. 2000]
 Brian D. Lang [BMB, Ph.D. 2000]
 Nicole Christacos [GMB, Ph.D. 2002]
 Charity (Crews) Davis [GMB, Ph.D. 2003]
 Jenny (Henderson) Schulz [NHS, Ph.D. 2004]
 Jamie Wasilenko [GMB, Ph.D. 2006]
 Melissa Brykailo [GMB, PhD 2007]
 Rebekah Kushner [BCDB, PhD 2009]
 Rebecca Sanders [BCDB, completed PhD 2009, MD-PhD 2011]
 Emily L. Ryan [BCDB, Ph.D. 2012]
 Jenna (Sefton) Daenzer [GMB, Ph.D. 2012]
 Darwin Hang [GMB, MS 2012]

PhD Rotation Advisees

1992-93	1 student	1993-94	4 students	1994-95	lab full
1995-96	1 student	1996-97	4 students	1997-98	2 students
1998-99	5 students	1999-00	5 students	2000-01	3 students
2001-02	4 students	2002-03	4 students	2003-04	4 students
2004-05	4 students	2005-06	5 students	2006-07	3 students
2007-08	5 students	2010-11	3 students	2011-12	1 student
2012-13	2 students				

Emory Undergraduate or School of Public Health or Post-Bac Research Assistants

1999-00 Beth Belletete
 2000-02 Linh Nguyen, Bryce Mendelsohn
 2002-03 Mindy Deason
 2003-04 Juliet Chhay
 2005-06 Corey Orton
 2007-08 Uriel Castaneda
 2008-09 Ambreen Dharani, John Kiel (SPH), James Yoon (SPH)
 2009-10 Uriel Castaneda, Xiwen Zheng, Caroline Wilson
 2010-11 Marquise Hopson, Bianca Copello

- 2011-12 Marquise Hopson, Erica Ditkoff
 2012-13 Marquise Hopson, Erica Ditkoff, Samantha Neumann

Summer Research Students (undergrads, high school or visiting students)

- | | | | |
|------|--|------|--------------------------------|
| 1993 | Brooks Moore | 1997 | Shannon Hook |
| 1998 | Long Le, Margaret Park | 1999 | Adi Sunderam |
| 2000 | Linh Nguyen | 2001 | Bryce Mendelsohn, Joshua Tusin |
| 2003 | Mindy Deason, Juliet Chhay | 2004 | Anjan Deka |
| 2005 | Corey Orton | 2006 | Amanda Carney |
| 2007 | Uriel Castaneda | 2008 | Ambreen Dharani |
| 2009 | Caroline Wilson, Ambreen Dharani, Ana Isabel Cruz Coelho (from Portugal) | | |
| 2010 | Bianca Copello, Avery Davis | 2011 | Susan Lee |
| 2012 | Samantha Neumann, Heather Bishop | | |

Long-distance mentoring

- 2003-04 Mr. Justice Lebea, School of Chemistry and Biochemistry, Potchefstroom University for Christian Higher Education, Potchefstroom, South Africa
- 2011-12 Mentor for PhD student Ms. Diana Koch (Univ of Cincinnati College of Medicine) through MentorNet (www.mentornet.net)

SEMINARS (local non-Emory)

- 2012 Georgia Glycoscience Symposium at UGA in Athens, GA (Keynote presentation "Impaired galactose metabolism in yeast, flies, and people" 10/3/12)
- 2012 Centers for Disease Control and Prevention (Topic: Outcomes in Duarte Galactosemia, Host: R. Olney, Aug 2012)
- 2008 Foundation for Genetic Technology (Decatur, GA Sept 2008)
- 2004 University of Georgia (Complex Carbohydrate Research Center)
- 2004 Clayton State University (Frontiers of Biology Seminar Series)
- 2002 Spelman College (Founders' Day Celebration)
- 1994 Georgia Institute of Technology

NATIONAL OR INTERNATIONAL PRESENTATIONS

- 2012 Galactosemia Foundation (formerly PGC, bi-annual meeting, I gave four presentations) (Dallas, Texas, July 2012)
- 2010 Meeting, the Portuguese Society of Metabolic Disorders (Algarve, Portugal, Nov 2010)
- 2010 Parents of Galactosemic Children (PGC, Bloomington, MN, July 2010)
- 2009 Galactosemia Workshop (Maastricht, The Netherlands May 2009)
- 2008 NIH/NICHD "Orphan Mechanisms of Primary Ovarian Insufficiency" (Bethesda, MD Oct 2008)
- 2008 Parents of Galactosemic Children (PGC, Chicago, July 2008)
- 2006 Parents of Galactosemic Children (PGC, Philadelphia, July 2006)
- 2005 Int. Symposium on Galactosemia: Facts & Unresolved Issues (Germany)
- 2003 Annual Meeting of the American Society of Human Genetics "Yeast and Human Genetic Disease" Invited Speaker Session
- 1997 Annual Meeting, Batten Disease Support and Research Association
- 1992 Annual Meeting, Society for Inherited Metabolic Disorders
- 1991 Gordon Research Conference

BIBLIOGRAPHY**Original research papers (published or accepted in peer-reviewed journals)**

1. Serhan, C.N., J. Fridovich, E. Goetzl, P. Dunham, and G. Weissmann. (1982). Leukotriene B₄ and Phosphatidic Acid are Calcium Ionophores. (Studies Employing Arsenazo III in Liposomes). *J. Biol. Chem.* 257:4746-4752.
2. Bond, J.F., J.L. Fridovich-Keil, L. Pillus, R. Mulligan, and F. Solomon. (1986). A Chicken-Yeast Chimeric Beta-Tubulin Protein is Incorporated into Mouse Microtubules *In Vivo*. *Cell* 44:461-468.
3. Fridovich-Keil, J.L., J.F. Bond, and F. Solomon. (1987). Domains of Beta-Tubulin Essential for Conserved Functions *In Vivo*. *Mol. Cell. Biol.* 7:3792-3798.
4. Dezube, B.J., J.L. Fridovich-Keil, I. Bouvard, R. Lange, and A.B. Pardee. (1990). Pentoxifylline and wellbeing in patients with cancer. *The Lancet* 335:662 (letter).
5. Bradley, D.W., Q.-P. Dou, J.L. Fridovich-Keil, and A.B. Pardee. (1990). Transformed and Non-Transformed Cells Differ in Stability and Cell Cycle Regulation of a Binding Activity to the Thymidine Kinase Promoter. *Proc. Natl. Acad. Sci. USA.* 87:9310-9314.
6. Fridovich-Keil, J.L., J. Gudas, Q.-P. Dou, I. Bouvard, and A.B. Pardee. (1991). Growth-Responsive Expression from the Murine Thymidine Kinase Promoter: Genetic Analysis of DNA Sequences. *Cell Growth and Differentiation.* 2:67-76.
7. Dou, Q.-P., J.L. Fridovich-Keil, and A.B. Pardee. (1991). Inducible Proteins Binding to the Murine Thymidine Kinase Promoter in Late G₁/ S Phase. *Proc. Natl. Acad. Sci. USA.* 88:1157-1161.
8. Fridovich-Keil, J.L., J.M. Gudas, I.B. Bryan, and A.B. Pardee. (1991). Improved Expression Vectors for Eukaryotic Promoter/Enhancer Studies. *BioTechn.* 11:572-9.
9. Gudas, J.M., J.L. Fridovich-Keil, M.W. Datta, J. Bryan, and A.B. Pardee. (1992) Molecular characterization of the murine thymidine kinase-encoding gene and analysis of transcription start point heterogeneity. *Gene* 118:205-216.
10. Leslie, N.D., E.B. Immerman, J.E. Flach, M. Florez, J.L. Fridovich-Keil, and L.J. Elsas. (1992) The Human Galactose-1-P Uridyltransferase Gene. *Genomics* 14:474-80.
11. Dezube, B.J., M.L. Sherman, J.L. Fridovich-Keil, J. Allen-Ryan, and A.B. Pardee. (1993). Down-regulation of tumor necrosis factor expression by pentoxifylline in cancer patients: a pilot study. *Cancer Immunol. Immunother.* 36:57-60.
12. Fridovich-Keil, J.L. and S. Jinks-Robertson. (1993). A yeast expression system for human galactose-1-phosphate uridyltransferase. *Proc. Natl. Acad. Sci. USA.* 90:398-402.
13. Gudas, J.M., J.L. Fridovich-Keil, and A.B. Pardee. (1993). Posttranscriptional Control of Thymidine Kinase mRNA during the Cell Cycle. *Cell Growth and Diff.* 4:421-30.
14. Fridovich-Keil, J.L., P.J. Markell, J.M. Gudas, and A.B. Pardee. (1993). DNA sequences required for serum-responsive regulation of the mouse thymidine-kinase promoter. *Cell Growth and Differentiation* 4:679-687.
15. Elsas, L.J., P.P. Dembure, S. Langley, E.M. Paulk, L.N. Hjelm, and J.L. Fridovich-Keil.(1994). A Common Mutation Associated with the Duarte Galactosemia Allele. *Am. J. Hum. Genet.* 54:1030-1036.
16. Bradley, D.W., J.L. Fridovich-Keil, J.M. Gudas, and A.B. Pardee. (1994). Serum-responsive expression from the murine thymidine kinase promoter is specifically disrupted in a transformed cell line. *Cell Growth and Differentiation* 5:1137-1143.
17. Fridovich-Keil, J.L., S.D. Langley, L.A. Mazur, J.C. Lennon, P.P. Dembure, and L.J. Elsas. (1995) Identification and functional analysis of three distinct mutations in the human gal-1-P uridyltransferase gene associated with galactosemia in a single family. *Am. J. Hum. Gen.* 56:640-646.
18. Elsas, L.J., S. Langley, E. Steele, J. Evinger, J.L. Fridovich-Keil, A. Brown, R. Singh, P. Fernhoff, L.N. Hjelm, and P.P. Dembure.(1995) Galactosemia: A Strategy to Identify New Biochemical Phenotypes and Molecular Genotypes. *Am. J. Hum. Gen.* 56:630-639.
19. Fridovich-Keil, J.L., B.B. Quimby, L. Wells, L.A. Mazur, and J.P. Elsevier (1995). Characterization of the N314D Allele of Human Galactose-1-Phosphate

- Uridyltransferase Using a Yeast Expression System. *Biochemical and Molecular Medicine* 56:121-130.
20. Elsevier, J.P., L. Wells, B.B. Quimby, and J.L. Fridovich-Keil. (1996) Heterodimer formation and activity in the human enzyme galactose-1-phosphate uridylyltransferase. *Proc. Natl. Acad. Sci. USA.* 93:7166-7171.
 21. Quimby, B.B., L. Wells, K.D. Wilkinson, and J.L. Fridovich-Keil. (1996) Functional requirements of the active site position 185 in the human enzyme galactose-1-phosphate uridylyltransferase. *J. Biol. Chem.* 271:26835-26842.
 22. Elsevier, J.P. and J.L. Fridovich-Keil. (1996) The Q188R mutation in human galactose-1-phosphate uridylyltransferase acts as a partial dominant negative. *J. Biol. Chem.* 271:32002-32007.
 23. Wells, L., and J.L. Fridovich-Keil. (1997) Biochemical characterization of the S135L allele of galactose-1-phosphate uridylyltransferase associated with galactosemia. *J. Inher. Metab. Dis.* 20:633-642.
 24. Quimby, B.B., A. Alano, S. Almashanu, A.M., DeSandro, T.M. Cowan, and J.L. Fridovich-Keil. (1997) Characterization of two mutations associated with epimerase-deficiency galactosemia using a yeast expression system for human UDP-galactose-4-epimerase. *Am. J. Hum. Gen.* 61:590-598.
 25. Wohlers, T.M., N.C. Christacos, M. T. Harreman, and J.L. Fridovich-Keil. (1999) Identification and characterization of a mutation in the human UDP-galactose-4-epimerase (GALE) gene associated with generalized epimerase-deficiency galactosemia. *Am. J. Hum. Gen.* 64:462-470.
 26. Lang, B.D and J.L. Fridovich-Keil. (2000) Scp160p, a multiple KH-domain protein, is a component of mRNP complexes in yeast. *Nucleic Acids Res.* 28:1576-1584.
 27. Thoden, J.B., T.M. Wohlers, J.L. Fridovich-Keil, and H.M. Holden. (2000) Crystallographic evidence for tyr157 functioning as the active site base in human UDP-galactose 4-epimerase. *Biochemistry* 39(19):5691-5701.
 28. Crews, C., K.D. Wilkinson, L. Wells, C. Perkins, and J.L. Fridovich-Keil. (2000) Functional consequence of substitutions at position 171 in human galactose-1-P uridylyltransferase. *J. Biol. Chem.* 275:22847-53.
 29. Christacos, N.C., M.J. Marson, L. Wells, K. Riehman, and J.L. Fridovich-Keil. (2000) Subcellular Localization of galactose-1-phosphate uridylyltransferase in the yeast *Saccharomyces cerevisiae*. *Mol. Gen. and Metabolism* 70:272-280.
 30. Henderson, J.M., L. Wells, and J.L. Fridovich-Keil. (2000) Covalent heterogeneity of the human enzyme galactose-1-phosphate uridylyltransferase. *J Biol Chem.* 275:30088-91.
 31. Wohlers, T.M. and J.L. Fridovich-Keil. (2000) Studies of the V94M-substituted human UDP-galactose-4-epimerase enzyme associated with generalized epimerase-deficiency galactosemia. *J. Inher. Metab. Dis* 23:713-729.
 32. Riehman, K., C. Crews, and J.L. Fridovich-Keil. (2001) Relationship between genotype, activity, and galactose sensitivity in yeast expressing patient alleles of human galactose-1-phosphate uridylyltransferase. *J. Biol. Chem.* 276(14):10634-40.
 33. Thoden, J.B., T.M. Wohlers, J.L. Fridovich-Keil, and H.M. Holden (2001) Human UDP-Galactose 4-epimerase: Accommodation of UDP-N-Acetylglucosamine Within the Active Site. *J. Biol. Chem.* 276(18):15131-15136.
 34. Thoden, J.B., T.M. Wohlers, J.L. Fridovich-Keil, and H.M. Holden (2001) Molecular basis for severe epimerase-deficiency galactosemia: X-ray structure of the human V94M-substituted UDP-galactose-4-epimerase. *J. Biol. Chem.* 276(23):20617-20623.
 35. Lang, B.D., A-M. Li, H.D. Black-Brewster, and J.L. Fridovich-Keil. (2001) The brefeldin A resistance protein Bfr1p is a component of polyribosome-associated mRNP complexes in yeast. *Nucleic Acids Research* 29(12):2567-2574.
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