CURRICULUM VITAE

NAME: **Barbara K. Burton, M.D.**

ADDRESS:

Residences: 155 Harbor Drive #3207
Chicago, IL 60601

2165 Gulf of Mexico Drive, # 132
Longboat Key, FL 34228

Business: Division of Genetics, Birth Defects and Metabolism
Ann & Robert H. Lurie Children's Hospital
225 E. Chicago Avenue, Box 59
Chicago, IL 60611
Phone: (312)-227-6120
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EDUCATION:

9/66-9/69 Northwestern University
Evanston, Illinois
Honors Program in Medical Education
B.S. in Medicine, awarded 1970

9/69-6/73 Northwestern University Feinberg School of Medicine
Chicago, Illinois
M.D. with Distinction

POSTDOCTORAL TRAINING:

7/73-6/75 Children's Memorial Hospital
Chicago, Illinois

	Pediatric Residency (PL I and PL II) Chief of Service: Henry L. Nadler, M.D.
7/75-6/77	Children's Memorial Hospital Chicago, Illinois Fellowship in Medical Genetics Program Director: Henry L. Nadler, M.D.

PROFESSIONAL LICENSURE:

Illinois, #036-051197

NPI NUMBER: 1699743088

SPECIALTY CERTIFICATION:

1978	American Board of Pediatrics
1982	American Board of Medical Genetics - Clinical Genetics - Clinical Biochemical Genetics

HONORS AND AWARDS:

1972	Elected to Alpha Omega Alpha
1973	Ross Award for Pediatric Research, presented by the Midwest Society for Pediatric Research
1974	Medical Foundation Clinical Scholar Wake Forest University Medical School
2003	Archibald Hoyne Award, presented annually by the Chicago Pediatric Society to a single individual for outstanding Contributions to the clinical practice of Pediatrics in the Chicago area
1994-2020	Listed every year, Best Doctors in America
1994-2017	Chicago Magazine: Best Doctors in Chicago
2017	Lifetime Achievement Award: March of Dimes Illinois Chapter
2018	PKU Hero Award: National PKU Alliance

ACADEMIC AND ADMINISTRATIVE APPOINTMENTS:

5/99-Present	Professor of Pediatrics Northwestern University Feinberg School of Medicine Chicago, IL
5/99-Present	Member Center for Genetic Medicine Northwestern University Feinberg School of Medicine Chicago, IL
5/99-12/19	Clinical Practice Director Division of Genetics Ann & Robert H. Lurie Children's Hospital of Chicago (formerly Children's Memorial Hospital) Chicago, IL
6/15-Present	Co-Director Leukodystrophy Center of Excellence Ann & Robert H. Lurie Children's Hospital of Chicago Chicago, IL
1/05-Present	Director MPS/ML Treatment Program Ann & Robert H. Lurie Children's Hospital of Chicago Chicago, IL
2/01-Present	Consulting Geneticist Institute for Fetal Health Ann & Robert H. Lurie Children's Hospital of ChicagoChicago, IL
5/99-Present	Director PKU Clinic Ann & Robert H. Lurie Children's Hospital of Chicago Chicago, IL
8/1-9/03	Consulting Geneticist Department of Obstetrics and Gynecology Northwestern University Feinberg School of Medicine Chicago, IL
7/00-6/13	Co-Director Joint Fellowship Training Program in Clinical Genetics Northwestern University-University of Chicago Chicago, IL

5/99-12/06	Lecturer in Pediatrics Lecturer in Obstetrics and Gynecology University of Illinois College of Medicine Chicago, IL
1/89-4/99	Professor and Head Division of Genetics and Metabolism Department of Pediatrics Professor of Obstetrics and Gynecology University of Illinois College of Medicine Chicago, IL
1/91-4/99	Program Director Medical Genetics Residency Program University of Illinois College of Medicine Chicago, IL
1/88-4/99	Director Genetics Diagnostic Laboratory Michael Reese Hospital and Medical Center Chicago, IL
1/89-4/99	Director Center for Medical and Reproductive Genetics Michael Reese Hospital and Medical Center Chicago, IL
7/87-12/88	Head Section on Medical Genetics Professor of Pediatrics Wake Forest University School of Medicine Winston-Salem, NC
12/77-12/88	Director Biochemical Genetics Laboratory Wake Forest University Medical Center Winston-Salem, NC
2/78-12/88	Director Western North Carolina Screening Program For Neural Tube Defects Winston-Salem, NC
7/80-12/88	Director Fetal Alcohol Syndrome Prevention Program Wake Forest University Medical Center Winston-Salem, NC

7/82-6/87	Associate Professor of Pediatrics Wake Forest University School of Medicine Winston-Salem, NC
12/77-6/82	Assistant Professor of Pediatrics Wake Forest University School of Medicine Winston-Salem, NC

HOSPITAL APPOINTMENTS:

8/01-3/19	Consulting Physician Northwestern Memorial Hospital Chicago, IL
4/99-Present	Attending Physician Division of Genetics, Metabolism & Birth Defects Ann & Robert H. Lurie Children's Hospital of Chicago Chicago, IL
10/99-12/03	Adjunct Attending Evanston Northwestern Healthcare (Evanston and Glenbrook Hospitals) Evanston, IL
1/93-6/98	Consulting Physician Grant Hospital Chicago, IL
3/90-3/00	Attending Physician University of Illinois Hospital Chicago, IL
2/90-3/00	Consulting Physician Children's Hospital of Illinois St. Francis Medical Center Peoria, IL
1/89-4/99	Attending Physician Michael Reese Hospital and Medical Center Chicago, IL
1/82-12/88	Consulting Physician Forsyth Memorial Hospital Winston-Salem, NC

12/77-12/88 Attending Physician
North Carolina Baptist Hospital
Winston-Salem, NC

GRANTS AND CONTRACTS:

1. Denali Therapeutics
   April 5, 2023- ongoing
   Budget dependent on number of subjects enrolled
   DNLI-E-007: A Phase 2/3, Multicenter, Double-Blind, Randomized Study to Determine The Efficacy and Safety of DNL310 vs. Idursulfase in Pediatric Participants with Neuronopathic and Non-Neuronopathic Mucopolysaccharidosis Type II.

2. Ultragenyx
   Feb 1, 2022- ongoing
   Budget dependent on number of subjects enrolled
   "Long Chain Fatty Acid Oxidation Defects In-Clinic Disease Monitoring Program"

3. Denali Therapeutics
   April 5, 2023-ongoing
   Budget dependent on number of subjects recruited
   "A Phase III Double-blind Randomized Controlled Study of DNL 310 vs. Idursulfase in Patients with Mucopolysaccharidosis Type II (Hunter Syndrome)"

4. JCR Pharmaceuticals
   January 1, 2022- ongoing
   Budget dependent on number of subjects recruited
   "A Phase III Study of JR-141 in Mucopolysaccharidosis Type II (Hunter Syndrome) Patients"

5. Denali Therapeutics
   November, 2020- open ended
   Budget dependent on number of subjects recruited
   "A Phase 1/2 Multicenter, Open-Label Study to Determine the Safety, Pharmacokinetics and Pharmacodynamics of DNL310 in Pediatric Patients with Hunter Syndrome.

6. Biomarin Pharmaceutical
   January 1, 2020- Open ended
   Budget dependent on number of subjects recruited
   "BMRN 307-902: A Prospective Clinical Study of Phenylketonuria"

5. Biomarin Pharmaceutical
   May 1, 2020- open ended
   Budget dependent on number of subjects recruited

" BMRN 307-201: A Phase ½ Open-Label Dose Escalation Study to Determine the Safety and Efficacy of BMRN 307 in Adeno-Associated Viral Vector-Mediated Gene Transfer of Human Phenylalanine Hydroxylase in Subjects with Phenylketonuria and Phe Levels >600 umol/L".

6 .Shire HGT (a Takeda company)
February 1, 2019- open ended
Budget dependent on number of subjects recruited
"SHP611-201: A Global, Multicenter, Open-Label, Matched Historical Control Study of Intrathecal SHP611 in Subjects with Late Infantile Metachromatic Leukodystrophy"

7. Reneo Pharma Ltd.
March 1, 2019- December 31, 2020
Budget dependent on number of patients recruited
"REN001-102: An Open-Label Study to Determine the Safety and Tolerability of 12 Weeks Treatment with Oral REN001 in Subjects with Fatty Acid Oxidation Disorders (FAOD)."

8. Moderna Therapeutics
January 1, 2019- open ended
Budget dependent on number of patients recruited
"MaP: Mapping the Patient Journey in Methylmalonic Acidemia and Propionic Acidemia: A Longitudinal, Exploratory, Natural History Study to Further Characterize and Describe The Signs and Symptoms of Patients with Organic Acidemias (mRNA-3704-P001)"

9. Homology Medicines
March 1, 2019- open ended
Budget dependent on number of subjects recruited
"HMI-102-101: A Phase ½ Open-Label, Randomized, Concurrently-Controlled, Dose Escalation Study to Evaluate the Safety and Efficacy of HMI-102 in Adult PKU Subjects With PAH Deficiency"

10. Shire HGT (a Takeda company)
July 1, 2018- June 30, 2019
Budget $101,786
"Followup of Newborn Screening for Mucopolysaccharidosis Type II"

11. Sangamo Biosciences, Inc.
January 1, 2017 – Open ended
Budget dependent on number of subjects recruited
"SB-913-1602: A Phase 1, Multicenter, Open- label Single – dose, Dose Ranging Study to Assess the Safety and Tolerability in Subjects with Mucopolysaccharidosis II. (MPS II)

12. Armagen

March 4, 2015 – May 31, 2017.
Budget dependent on numbers of subjects recruited
"AGT-182-102: A Phase 1 Safety and Dose – finding Study of a Human Insulin Receptor Monoclonal Antibody – Human Iduronate 2-Sulfatase (IDS) Fusion Protein, AGT-182 in Adult patients with Mucopolysaccharidosis II (MPS II)"

13. Armagen
September 30, 2015 – March 30, 2017
Budget dependent on number of subjects recruited
"AGT – 181-101: A Phase I Safety and Dose Finding Study of a Human Insulin Receptor Monoconal Antibody Human Alpha-L-Iduronidase (HIRMAb-IDUA) Fusion Protein, AGT-181 in Adult Patients with Mucopolysaccharidosis I (MPS I)

14. Shire HGT
March 1, 2014 – March 30, 2017
Budget dependent on number of subjects recruited
"HGT-HIT094: A Controlled, Randomized Two-Arm Open-Label, Assessor-Blinded, Multicenter Study of Intrathecal Elaprase-IT Administered in Conjunction with Elaprase in Pediatric Patients with Hunter Syndrome and Early Cognitive Impairment"

15. Ultragenyx Pharmaceutical
November 1, 2013 – June 30, 2020
Budget dependent on number of subjects recruited
"UX007-CL201: An Open-Label Phase 2 Study to Assess Safety and Clinical Effects of UX007 in Subject with Long-Chain Fatty Acid Oxidation Disorders (LC-FAOD)"

16. Shire HGT
April 1, 2013 – February, 2016
Budget dependent on number of subjects recruited
"HGT-MLD-092: A Natural History Study of Children with Metachromatic Leukodystrophy"

17. Biomarin Pharmaceutical
April 1, 2013 – Open ended
Budget dependent on number of subjects recruited
"BMRN 009-901: Collection and Storage of Human Biospecimens for Research into Rare Diseases and Medical Conditions"

18. Synageva Pharma
March 1, 2013 – February, 2015
Budget dependent on number of subjects recruited
"ARISE: A Phase 3 Multicenter, "A Double-Blind, Placebo-Controlled Trial of SBC-100 in Patients with Cholesterol Ester Storage Disease"

19. Shire HGT
February 1, 2013 – June 30, 2017
Budget dependent on number of subjects recruited

"HGT-HIT-090: A Study of the Natural History of Cognitive Decline in Patients with Hunter Syndrome (MPS II) Associated with Cognitive Impairment"

20. Biomarin Pharmaceutical
June 1, 2013 – August, 2016
Budget dependent on number of subjects recruited
"PRISM 301: A Phase 3 Study of Pegylated Phenylalanine Ammonia Lyase (PEG- PAL) in Patients with PKU Naïve to the Study Drug"

21. Biomarin Pharmaceutical
July 1, 2013 – September 30, 2018
Budget dependent on number of subjects recruited
"PRISM 302: A Phase 3 Randomized Discontinuation and Long Term Extension Study of Pegylated Phenylalanine Ammonia Lyase (PEG-PAL) in Subjects with PKU"

22. Shire Human Genetic Therapies
August 4, 2011 – December 31, 2016
$156,178
"Follow-up Assessment of a Pilot Newborn Screening Program for Lysosomal Storage Disorders"

23. Biomarin Pharmaceuticals
June 1, 2011 – July, 2014
Budget dependent on number of subjects recruited
"PKU-016: A Double-Blind, Placebo-Controlled Randomized Study to Evaluate the Safety and Therapeutic Effects of Sapropterin Dihydrochloride on Neuropsychiatric Symptoms in Subjects with Phenylketonuria (PKU ASCEND)"

24. Shire Human Genetic Therapies
September 24, 2009 – March 31, 2011
$126,238
"Does Enzyme Replacement Therapy Reduce the Incidence and/or Severity of Infectious Illnesses in Patients with Mucopolysaccharidosis?"

25. BioMarin Pharmaceuticals
April 1, 2011 – December 31, 2013
Budget dependent on number of subjects recruited
"MOR-004: A Multicenter, Multinational Double-Blind, Placebo-Controlled Study of the Safety and Efficacy of 2mg/kg/week and 2mg/kg/every other week BMN 110 in patients with Mucopolysaccharidosis IVA (Morquio IVA)"

26. BioMarin Pharmaceuticals
March 1, 2012 – April 30, 2014
Budget dependent on number of subjects recruited

"MOR-005: A Multicenter, Multinational Extension Study to Evaluate the Long-Term Efficacy and Safety of BMN 110 in patients with Mucopolysaccharidosis IVA (Morquio A Syndrome)"

27. BioMarin Pharmaceuticals
March 1, 2012 – November 30, 2014
Budget dependent on number of subjects recruited
"MOR-008: A Randomized Double-Blind Pilot Study of the Safety and Physiological Effects of Two Doses of BMN 110 in patients with Mucopolysaccharidosis IVA (Morquio A Syndrome)"

28. NIH – Subcontract with Michigan Public Health Institute
March 1, 2011 – February 28, 2016
$40,547 for year 1
"Inborn Errors of Metabolism Information System (IBEM-IS)"

29. Cytonet GMBH & Co
February 1, 2011 – Open ended
Budget dependent on number of subjects recruited
"An Open Prospective, Historic-Controlled, Multicenter Study to Evaluate the Safety and Efficacy of Infusion of Liver Cell Suspension (hhlivc) in Children with Urea Cycle Disorders"

30. BioMarin Pharmaceuticals
July 1, 2009 – December 30, 2012
Budget dependent on number of subjects recruited
"PAL-002: A Phase 2, Open-Label Dose-Finding Study to Evaluate the Safety, Efficacy, and Tolerability of Multiple Subcutaneous Doses of rAvPAL-PEG in Subjects with PKU"

31. BioMarin Pharmaceuticals
July 1, 2009 – Open ended
Budget dependent on number of subjects recruited
"PAL-003: Long Term Extension of a Phase 2, Open-Label Dose-Finding Study to Evaluate the Safety, Efficacy, and Tolerability of Multiple Subcutaneous Doses of rAvPAL-PEG in Subjects with PKU"

32. BioMarin Pharmaceuticals
March 1, 2009 – Open ended
Budget dependent on number of subjects recruited
"PKU-015: A Phase 3b Open-Label Study to Evaluate the Effect of Kuvan on Neurocognitive Function, Maintenance of Blood Phenylalanine Concentrations, Safety and Population Pharmacokinetics in Young Children with Phenylketonuria"

33. BioMarin Pharmaceuticals
March 1, 2009 – February 28, 2010
Budget dependent on number of subjects recruited (subcontract with Washington University)

"Behavioral Effects of Kuvan® on Children with Mild PKU"

34. BioMarin Pharmaceuticals
September 1, 2008 – May 30, 2014
Budget dependent on number of subjects recruited
"MOR-001: A Multicenter, Multinational Cross-Sectional Clinical Assessment Study of Subjects with Mucopolysaccharidosis IVA (Morquio Syndrome)"

35. BioMarin Pharmaceuticals
October 1, 2008 – Open ended
Budget dependent on number of subjects recruited
"PKUDOS: A Long Term Observational Registry of Patients with PKU Exposed to Kuvan"

36. BioMarin Pharmaceuticals
July 1, 2008 – June 30, 2009
$120, 238
"Adult PKU Outreach Program"

37. BioMarin Pharmaceuticals
March 1, 2008 – March 31, 2009
Budget dependent on number of subjects recruited
"rAvPAL-PEG-001: A Phase 1, Multicenter, Open-Label Study to Evaluate the Safety and Tolerability of rAvPAL-PEG in Patients with PKU"

38. Shire Human Genetic Therapies
January 16, 2008 – July 15, 2009
$100,743
"Screening for Mucopolysaccharidosis in Patients with Inguinal or Umbilical Hernias"

39. Shire Human Genetic Therapies
January 16, 2007 – July 15, 2008
$109,281
"Screening for Mucopolysaccharidosis in Patients with Kyphosis"

40. Shire Human Genetic Therapies
November 1, 2005 – January 15, 2007
$75,000
"Mucopolysaccharidosis (MPS) Treatment Center"

41. BioMarin Pharmaceuticals
March 17, 2006 – July, 2008
Budget dependent on number of subjects recruited
"PKU-008: A Phase 3b, Multicenter, Open-Label Extension Study of Phenoptin in Subjects with Phenylketonuria Who Participated in Studies PKU-004 or PKU-006

42. BioMarin Pharmaceuticals

March 20, 2006 – April, 2009
Budget dependent on number of subjects recruited
"PKU-007: A Phase 2, Multicenter, Open-Label Study to Evaluate the Safety and Efficacy of Phenoptin in Subjects with Hyperphenylalaninemia Due to Primary BH4 Deficiency"

43. BioMarin Pharmaceuticals
October 13, 2005 – March, 2007
Budget dependent on number of subjects recruited
"PKU-006: A Phase 3, Multicenter, Randomized, Double-Blind, Placebo-Controlled Study to Evaluate the Safety and Efficacy of Phenoptin 20mg/kg/day to Increase Phenylalanine Tolerance in Phenylketonuric Children on a Phenylalanine-restricted Diet"

44. BioMarin Pharmaceuticals
June 6, 2005 – June, 2007
Budget dependent on number of subjects recruited
"PKU-004: A Phase 3, Multicenter, Open-Label Extension Study of Phenoptin in Subjects with Phenylketonuria Who Have Elevated Phenylalanine Levels"

45. BioMarin Pharmaceuticals
March 3, 2005 – February, 2006
Budget dependent on number of subjects recruited
"PKU-003: A Phase 3, Randomized, Double-Blind, Placebo-Controlled Study to Evaluate the Safety and Efficacy of Phenoptin in Subjects with Phenylketonuria"

46. BioMarin Pharmaceuticals
October 1, 2004 – February, 2006
Budget dependent on number of subjects recruited
"PKU-001: A Phase 2, Multicenter, Open-Label Study to Evaluate the Response to and Safety of a 7-day Course of Phenoptin in Subjects with Phenylketonuria Who Have Elevated Phenylalanine Levels"

47. The Mid-Atlantic Connection for PKU and Allied Disorders, Inc.
September, 2002-Sept. 2003
$10,500
"Evaluation of Tetrahydrobiopterin Responsiveness in Patients with Phenylalanine Hydroxylase Deficiency"

48. PKU Organization of Illinois
July, 2002-Nov., 2003
$8,900
"Evaluation of Tetrahydrobiopterin (BH4) Responsiveness in Patients with Phenylalanine Hydroxylase Deficiency"

49. National Institutes of Health
(Subcontract with Children's Hospital of Los Angeles)

January, 2000-Dec 31, 2000
$12,500
"Phenylketonuria in Adulthood: A collaborative study"

50. Welch's Corporation
July 1, 1994 - June 30, 1995
$20,000
Pediatric Birth Defects Clinic

51. Columbia Michael Reese Hospital Intramural Research Grant Program
September 1, 1994 - August 31, 1995
$35,000
Preimplantation Diagnosis of Cytogenetic Disorders

52. State of Illinois Department of Health
July 1, 1992 - June 30, 1999; renewed annually
$50,000/year
Genetic Counseling Outreach Program

53. Women's Board
Michael Reese Hospital and Medical Center
January 1, 1991 - December 31, 1991
$50,000 Total Support
Research in Medical Genetics

54. Junior Medical Research Institute Council
Michael Reese Hospital and Medical Center
July 1, 1990 - June 30, 1992
$50,000 Total Support
Prospective Evaluation of a New Prenatal Screening Test
for Down Syndrome"

55. March of Dimes Birth Defects Foundation
July 1, 1986 - June 30, 1988
$30,000 Total Support
Fetal Alcohol Syndrome Information Service

56. State of North Carolina Developmental Disabilities Council
October 1, 1983 - September 30, 1986
$295,679 Total Support
"Comprehensive Fetal Alcohol Syndrome Project"

57. National Center for Health Services Research
July 1, 1981 - June 30, 1984
$296,469 Total Support
"Emotional and Developmental Impact of MSAFP Screening"

58. March of Dimes Birth Defects Foundation
Basil O'Connor Starter Research Grant
September 1, 1978 - June 30, 1981
$55,401 Total Support
"Lysosomal Acid Lipase and Its Relationship to Inborn Errors of Metabolism"

59. Intramural Research Support Program
Bowman Gray School of Medicine
July 1, 1978 - June 30, 1979
$3,000 Total Support
Lysosomal Acid Lipase and Its Relationship to
Inborn Errors of Metabolism"

60. State of North Carolina Department of Human Resources
July 1, 1981 - December 31, 1988
$11,082 annually
"Genetic Satellite Clinics"

61. State of North Carolina Department of Human Resources
July 1, 1986 - December 31, 1988
$175,824 annually
Genetic Counseling Program

62. State of North Carolina Department of Human Resources
July 1, 1986 - June 30, 1989
$46,000 annually
"Fetal Substance Abuse Prevention Project"

63. State of North Carolina Department of Human Resources
July 1, 1979 - December 31, 1988
$236,581 annually
"Regional Screening Program for Neural Tube Defects"

PROFESSIONAL MEMBERSHIPS, past and present:

American Medical Association
American Society of Human Genetics
Illinois Chapter, American Academy of Pediatrics
Executive Committee 2000-2004
Chicago Pediatric Society
Executive Committee 1994-2002
President 1999-2000
American College of Medical Genetics
Society for Pediatric Research

Society for Inherited Metabolic Disease (Board of Directors 1996-2011;
Treasurer 1999-2002; Secretary 2002; Program Director 2003-2005;
President-Elect 2005-2007, President 2007-2009)
Genetics Task Force of Illinois (President 1992-1993)

JOURNAL REVIEWER:

American Journal of Medical Genetics
American Journal of Obstetrics and Gynecology
Clinical Genetics
Journal of Pediatrics
Molecular Genetics and Metabolism
MGM Reports
Obstetrics and Gynecology
Orphanet Journal of Rare Disease
Pediatric Research
Pediatrics
Prenatal Diagnosis
Journal of Inherited Metabolic Disease
JIMD Reports

EDITORIAL BOARDS

International Journal of Newborn Screening

NIH STUDY SECTIONS:

2015-2021	Permanent member Therapeutic Approaches to Genetic Disease (TAG) study section.
2013-2014	Ad Hoc reviewer. Therapeutic Approaches to Genetic Diseases study section.

NATIONAL COMMITTEES:

2022-present	Member, Scientific Advisory Board, National MPS Society
2021-present	ACMG Committee to Develop Revised Clinical Practice Guidelines for PKU
2011-2012	NIH Workgroup on Phenylketonuria Long Term Outcomes
2011-2012	ACMG Ad Hoc Committee to Develop Clinical Practice Guidelines for Phenylketonuria (PKU)

2007-2011	Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children U.S. Department of Health and Human Services (SIMD Liaison Representative)
2006-2010	Executive Committee Public Health Special Interest Group American College of Medical Genetics
2003-2005	Program Committee American College of Medical Genetics
2002-2006	Clinical Practices Committee Section of Genetics American Academy of Pediatrics
2002-Present	Medical Advisory Board Propionic Acidemia Foundation
1996-2000	Medical Advisory Board KURE (Kids with Urea Cycle Defects Research Enterprise)
1993-2000	Medical Advisory Board National Urea Cycle Defects Foundation
1990-1993	Subcommittee on Alpha-Fetoprotein Genetic Services Committee American Society of Human Genetics
1988-1992	Subcommittee on MSAFP Screening Council on Regional Genetic Networks (CORN)
1979-1982	Ethics Committee Birth Defects and Clinical Genetics Society

STATE AND REGIONAL COMMITTEES:

2017-Present	Member, Rare Disease Commission State of Illinois Department of Public Health
2015-2021	Member, Genetic and Metabolic Screening Advisory Committee. State of Illinois Department of Public Health Chairman, 2018-2021
2009–2020	Chairman, Subcommittee on LSD Screening

State of Illinois Department of Public Health

2006-2008	Steering Committee Illinois State Genetic Services State Plan
2006-2008	Chairman, Genetic and Metabolic Screening Advisory Committee State of Illinois Department of Public Health
2002-2008 and 1993-1994	Member, Genetic and Metabolic Screening Advisory Committee State of Illinois Department of Public Health
2004–2008	Chairman, Subcommittee on Cystic Fibrosis Newborn Screening Genetic and Metabolic Screening Advisory Committee State of Illinois Department of Public Health
2004–Present	Laboratory Subcommittee Genetic and Metabolic Screening Advisory Committee State of Illinois Department of Public Health
2004–2016	Region 4 Genetics Collaborative (A HRSA-Funded Public Health Initiative Involving 7 States) Long Term Newborn Screening Follow-up and Evaluation of Clinical Outcomes Working Group Illinois Representative
2001-2002	Task Force for Implementation of Expanded Newborn Screening State of Illinois Department of Health
1990-1992	Committee on Access to Genetic Services Genetics Task Force of Illinois
1986-1988	Task Force on Prevention of Fetal Alcohol Syndrome State of North Carolina Department of Human Resources
1983-1988	Screening Committee North Carolina Medical Genetics Association (Chairman, 1985-1986)
1983-1988	Advisory Committee on Newborn Screening State of North Carolina Division of Health Services
1982-1983	Work Group on Child Health North Carolina 2000 Project of the State Goals and Policy Board State of North Carolina

MEDICAL SCHOOL COMMITTEES:

2000-2019	Co-Chairman Committee on Appointments, Promotions and Tenure Department of Pediatrics Northwestern University Feinberg School of Medicine
2004	Search Committee Palliative Care Director Children's Memorial Hospital
2001-2006	Clinical Board Department of Pediatrics Northwestern University Feinberg School of Medicine
2001-2002	Search Committee Endocrine Division Head Department of Pediatrics Northwestern University Medical School
2000-2002	Advocacy Council Department of Pediatrics Northwestern University Feinberg School of Medicine
1995-1998 & 1991-1994	Faculty Senate University of Illinois at Chicago
1995-1997	Department of Pediatrics Advisory Committee University of Illinois College of Medicine
1992-1994	Woman's and Children's Task Force Strategic Planning Program University of Illinois College of Medicine
1990-1991	Dean's Advisory Council University of Illinois College of Medicine
1988	Adopt-a-School Program Wake Forest University Medical Center
1987-1988	Faculty Forum Committee Wake Forest University Medical Center
1987-1988	Associate Dean's Advisory Council

Wake Forest Medical Center

1986-1988 Admissions Committee
Wake Forest University Medical Center

1985-1986 Ad Hoc Promotions Committee
Wake Forest University Medical Center

1982-1987 Chairman
Security Advisory Committee
Wake Forest University Medical Center

1982 Chairman
Ad Hoc Committee on Special Research Requirements
Wake Forest University Medical Center

1978-1981 Intramural Research Support Committee
Wake Forest University Medical Center

HOSPITAL COMMITTEES:

2011-2012 Test Utilization Committee
Children's Memorial Hospital

2003 Service Excellence Committee
Children's Memorial Hospital

2001 Ambulatory Access Task Force
Children's Memorial Hospital

1996-1998 Summer Student Research Program Subcommittee
Michael Reese Hospital and Medical Center (Chairman)

1990-1992 Patient Care Committee
Michael Reese Hospital and Medical Center

1988-1999 Research Committee
Michael Reese Hospital and Medical Center
(Vice-Chairman 1998-1999)

COMMUNITY SERVICE:

2001-2008 Medical Advisory Council
Make a Wish Foundation

	Northern Illinois Chapter
2006-2008	Co-Chairman Illinois Folic Acid Council Sponsored by March of Dimes Birth Defects Foundation Greater Chicago Chapter
2008-Present	Member, Emeritus Board of Directors March of Dimes Birth Defects Foundation Greater Chicago Chapter
2003-2008	Member, Board of Directors March of Dimes Birth Defects Foundation Greater Chicago Chapter
1998-2008	Member, Community Service Grants Committee March of Dimes Birth Defects Foundation Greater Illinois Chapter
1998-2001	Chairperson Genetics and Your Practice Education Program for Primary Care March of Dimes Birth Defects Foundation Greater Illinois Chapter
1998-2000	Member, Health Leadership Awards Committee March of Dimes Birth Defects Foundation Greater Illinois Chapter

BIBLIOGRAPHY

Books

Kumar P, Burton BK, eds. Congenital Malformations: Evidence-Based Evaluation and Management. New York: McGraw-Hill, 2008.

Blair N, Burton BK, Thony B, Van Spronsen FJ, Waisbren S. Phenylketonuria and BH4 Deficiencies. Bremen: UNI-MED, 2010; Revised edition 2021

Chapters in Books:

Nadler HL, Burton BK. Enzymes in the amniotic fluid and the prenatal diagnosis of inborn errors of metabolism. In: Fairweather DVI, Eskes TKAB, eds. Amniotic fluid. Amsterdam: Excerpta Medica, 1973:223-261.

Burton BK, Gerbie AB, Nadler HL. Enzymes in the amniotic fluid and the prenatal diagnosis of inborn errors of metabolism. In: Fairweather DVI, Eskes TKAB, eds. Amniotic fluid: research and clinical application, 2nd ed Amsterdam: Excerpta Medica, 1978:225-245.

Burton, BK. Empiric recurrence risks for congenital hydrocephalus. Birth Defects: Original Articles Series 1979:15(5c):107-115.

Burton, BK, Nadler, HL. Screening the newborn for genetic and metabolic disorders. In: Moss A, ed. Pediatric update: review for physicians. New York: Elsevier, 1979:131-142.

Burton BK, Gerbie AB, Nadler HL. Biochemical and biologic pitfalls in the use of cell culture for prenatal diagnosis. In: Milunsky A, ed. Genetic disorders and the fetus: diagnosis, prevention and treatment. New York: Plenum, 1979:369-377.

Nadler HL, Burton, BK. Genetics. In: Kretchmer N, Quilligan EJ, eds. Fetal and maternal medicine. New York: Wiley & Sons, 1980:59-107.

Nadler HL, Burton BK. Genetics in surgery. In: Raffensperger J, ed. Swenson's pediatric surgery. New York: Appleton-Century-Crofts, 1980:85-103.

Sowers SG, Burton BK. The clinical significance of low maternal serum alpha-fetoprotein in obstetric practice. In: Nyhan WL, Jones K, etc. Annual review of birth defects, 1981 New York: Alan R. Liss, Inc., 1982:181-184.

Burton BK, Nadler HL. Biological and biochemical pitfalls in the prenatal diagnosis of inborn errors of metabolism by amniocentesis. In: Milunsky A, ed. Genetic disorders and the fetus: diagnosis, prevention and treatment, 2nd ed. New York: Plenum, 1986:437-451.

Nelson LH, Burton BK, Sowers SG. Maternal serum alpha-fetoprotein. In: Sabbagha RE, ed. Diagnostic ultrasound applied to obstetrics and gynecology, 2nd ed. Philadelphia:Lippincott, 1987:252-263.

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Giugliani R, Bodmer O, Burton B, DeMeirleir L, Harmatz P, Jones S, Lampe C, Malm G, Parini R, Steiner R, Mendelsohn N, on behalf of the HOS Investigators. Investigating numbers of surgical interventions performed in patients with mucopolysaccharidosis type II (MPS II) using data from the Hunter Outcome Survey (HOS). Society for the Study of Inborn Errors of Metabolism (SSIEM) Annual Meeting 2010; Poster presentation.

Harmatz P, Chang M, Decker C, Lee S, Martell L, Burton B, Guffon N, Hendriksz C, Hollak C, Jones S, Lin S, Mengel E, Mitchell J, Parini R, Valayannopoulos V, Vellodi A, Cheng S. A multicenter, multinational, longitudinal clinical assessment study of subjects with mucopolysaccharidosis IVA (Morquio Syndrome). SSIEM Annual Meeting 2010; Platform presentation.

Kim KH, Burton BK, Parent care-giver administration of home enzyme replacement therapy in the mucopolysaccharidoses (MPS disorders). Annual Meeting of the Amer Soc Hum Genet 2010; Poster presentation.

Bandholz AM, Rosenfeld JA, Torchia BS, Ravnan JB, Schulz RA, Alliman S, Bamforth JS, Babu D, Weaver DD, Miller A, Burton BK, Hartung D, Peterson E, Lafayette D, Smith R, Ellingwood SA, Lamb AN. Molecular and phenotypic characterization of 2p15p16.1 microdeletions and microduplicatons. Annual meeting of the Amer Coll Med Genet 2011; Poster presentation.

Leviton L, Vespa H, Burton BK, Mental health screening in the Phenylketonuria (PKU) Clinic. Annual meeting of the Amer Coll Med Genet 2011; Poster presentation.

Burton BK, Charrow J, Angle B, Widera S, Waggoner D. A pilot newborn screening program for Lysosomal Storage disorders (LSDs) in Illinois. Lysosomal Disease Network WORLD Symposium 2012; Platform presentation.

Kim K, Dodsworth C, Paras A, Burton B. High dose genistein treatment in severe MPS II and III patients. Lysosomal Disease Network WORLD Symposium 2012; Poster presentation.

Kim K, Dodsworth C, Paras A, Burton BK. Assessing high dose genistein treatment in MPS patients with neurologic involvement. Annual meeting of the Amer Coll Med Genet 2012; Poster presentation.

Prasad S, Burton BK, Feigenbaum A, Grant M, Hendren R, Mardach R, Phillips J, Sanchez-Valle A, Singh R, Siriwardena K, Thomas J, Stahl S, Lang W, Kim S, Jurecki E. Baseline findings in the first 60 subjects in PKU ASCEND (016): A double-blind placebo-controlled, randomized study to evaluate the safety and therapeutic effects of sapropterin dihydrochloride on neuropsychiatric symptoms in subjects with phenylketonuria (PKU). Annual meeting of the Amer Coll Med Genet and Soc for Inher Metab Dis 2012; Poster presentation

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Hillman R, Peck D, Arnold GL, Burton BK, Enns GM, Longo N, Mofidi S, Pridjian G, Waisbren S, White D, Gravance C. Sapropterin use in pregnant phenylketonuria patients:The PKU MOMS subregistry. Annual meetings of the Amer Coll Med Genet and Soc for Inher Metab Dis 2012; Poster presentation.

Kim K, Bausell H, Sale T, Zhou Y, Burton BK. Comparison of phenylketonuria (PKU) patients' height, weight and body mass index (BMI) to the general population. Annual meeting of the Soc for Inher Metab Dis 2012; Poster presentation.
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Widera S, Charrow J, Burton B, Helgeson M. Newborn screening for Fabry disease leads to a diagnosis in a symptomatic maternal relative. Presented at the Lysosomal Network's WORLD Symposium 2013, Feb 16-18, 2013, Orlando, FL.

Jiang R, Paras A, He M, Valencia CA, Burton B, Hegde M. New preclinical treatment paradigms for ALG12-CDG (CDG-1g). Presented at the 2013 Meeting of the American College of Medical Genetics, March 19-23, Phoenix, AZ.

Longo N, Siriwardena K, Feigenbaum A, Dimmock D, Burton B, Stockler S, Waisbren S, Lang W, Jurecki E, Prasad S. Long term developmental progression in young children taking sapropterin for phenylketonuria: a two-year analysis of safety and efficacy. Presented at the 2013 meeting of the American College of Medical Genetics, March 19-23, Phoenix, AZ.

Hendriksz CJ, Muenzer J, Vandever A, Davis JM, Burton BK, Mendelsohn NJ, Wang N, Pan L, Pano A, Cammarata S, Barbier A. Levels of glycosaminoglycans in the cerebrospinal fluid of healthy young adults, surrogate-normal children, and Hunter

syndrome patients with and without cognitive impairment. Platform presentation at the WORLD Symposium of the Lysosomal Disease Network, Feb 12-14, 2014.

Kim KH, Widera S, Messinger Y, Burton BK. Immune modulation therapy in severe MPS II patient: follow up after one year of treatment. Poster presentation at the WORLD Symposium of the Lysosomal Disease Network, Feb 12-14, 2014

Jones SA, Burton BK, Morin I, on behalf of the HOS Investigators. Survival and causes of death in patients with Hunter syndrome: data from the Hunter Outcome Survey (HOS). Poster presentation at the Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Sept 2-5, 2014.

Edano C, Malick M, Burton BK. Infusion management of elosulfase alfa for patients with Morquio A syndrome. (MPS IVA). Poster presentation at the Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Sept 2-5, 2014.

Balwani, M, Burton B, Burrow TA, Quinn AG and the ARISE Investigators. Results of a Global Phase 3, Randomised, Double-Blind, Placebo-Controlled Trial Evaluating the Efficacy and Safety of Sebelipase Alfa as an Enzyme Replacement Therapy in Children and Adults with Lysosomal Acid Lipase Deficiency. Presented at the Annual Meeting of the American Association for the Study of Liver Diseases (AASLD), Nov 7-11, 2014.

Burton B, on behalf of the ARISE Investigators. Results of a global phase 3, randomized of a global phase 3, randomized, double-blind placebo controlled trial evaluating the efficacy and safety of sebelipase alfa as an enzyme replacement therapy in children and adults with lysosomal acid lipase deficiency. Presented at the Lysosomal Disease Network's WORLD symposium, Feb 10-12, 2015.

Burton B, Harmatz P, Mitchell J, Muschol N, Jones S; Pastores G, Lau H, Sparkes R, Sutton VR, Berger K, Lewis G, Tarnopolsky M, Genter F, Haller C, Shaywitz A. Impact of elosulfase alfa on exercise capacity and muscle strength and safety in patients with Morquio syndrome type A. Presented at the Lysosomal Disease Network's WORLD Symposuim, Feb 10-12, 2015.

Morin I, Mendelsohn N, Burton B, Jones SA, Lampe C, Molter D. Ear, nose, and throat and hernia surgeries in children with Hunter syndrome: Data from the Hunter outcome survey (HOS). Presented at the Lysosomal Disease Network's World Symposium, Feb 10-12, 2015.

Thomas JA, Longo N, Zori R, Burton BK, et al. Evaluation of multiple dosing regimens in Phase 2 studies of rAvPAL-PEG (BMN 165, Pegvaliase) in control of blood phenylalanine levels in adults with phenylketonuria. Poster presentation Annual Meeting of the Society for Inherited Metabolic Disorders Mar 28-31, 2015.

Longo N, Thomas JA, Wasserstein M, Burton BK, et al. Evaluation of long-term safety and efficacy with RAVPAL-PEG (BMN.165) for control of blood phenylalanine levels in

adults with phenylketonuria (PKU). Poster presentation, Annual Meeting of the society for Inherited Metabolic Disorders, March 28-31, 2015.

Harding C, Longo N, Thomas JA, Burton BK, et al. Phase 2 studies contribute to RAVPAL-PEG phase 3 trial design. Poster presentation, Annual Meeting of the Society for Inherited Metabolic Disorders, Mar 28-31, 2015.

Khan A, Barshop BA, Burton BK, Vas M, Emre SH. Long term outcome in children with urea cycle disorder after hepatocyte transplantation. Poster presentation. Annual Meeting of the Society for Inherited Metabolic Disorders. Mar 28-31, 2015.

Leeth E, Kirschmann D, Burton B, Kim K, Bucher J, Young L, Jennings L. Identification of a novel SMAD3 variant leads to diagnosis of Loeys-Dietz syndrome in a three generation family previously suspected as probable Marfan syndrome. Poster presentation, Annual Meeting of the American College of Medical Genetics and Genomics, Mar 25-28, 2015.

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Burton BK, Harding C, Longo N, Thomas JA, et al. Phase 2 studies contribute to rAvPAL-PEG phase 3 trial design. Poster presentation Annual meeting of the SSIEM, Sept 1-4, 2015.

Burton B, Ficicioglu C, Bosch J, Morin I; Jurecka A, Tylki-Szymanska A. Characteristics of patients with mucopolysaccharidosis type II identified at a very young age: data from the Hunter outcome survey (HOS). Poster presentation, Annual Meeting of the SSIEM, Sept 1-4, 2015.

Amartino H, Burton B, Giugliani R, et al. Development and reliability assessment of the MPS II disease severity score. Poster presentation, Annual Meeting of the SSIEM, Sept 1-4, 2015.

Longo N, Thomas JA, Wasserstein M, Burton BK, et al. Evaluation of long-term safety and efficacy with rAv-PAL-PEG for control of blood phenylalanine levels in adults with phenylketonuria (PKU). Platform presentation, Annual Meeting of the SSIEM, Sept 1-4, 2015.

Burton B, Berger KL, Lewis GD, et al. Impact of elosulfase alfa on exercise capacity in patients with Morquio A syndrome in a randomized double-blind pilot study. Poster presentation, Annual Meeting of the SSIEM, Sept 1-4, 2015

Stewart F Harmatz P, Braulin E, Bentley A, Burton B, et al. Management of fertility and pregnancy in individuals with mucopolysaccharidosis (MPS). Poster presentation, Annual Meeting of the SSIEM, Sept 1-4, 2015.

Burton B, Balwani M, Feillet F, et al. Efficacy and safety of sebelipase alfa in children and adults with lysosomal acid lipase deficiency: results of a phase 3 trial. Platform presentation. Annual Meeting of the SSIEM, Sept 1-4, 2015.

Giugliani R, Burton BK, Harmatz P, et al. Impact of long-term elosulfase alfa treatment on three-minute stair climb test, pulmonary function tests and normalized urine keratin sulfate in patients with Morquio A syndrome. Poster presentation, Annual meeting of the SSIEM, Sept 1-4, 2015.

Harmatz P, Burton BK, Giugliani R, et al. Impact of long-term elosulfase alfa treatment on six-minute walk test distance in patients with Morquio A. syndrome. Poster presentation, Annual meeting of the SSIEM. Sept 1-4, 2015.

Atherton AM, Burton BK, Day-Salvatore DL, et al. Guidelines for the diagnosis and management of infants with MPS I identified through newborn screening. Presented at the American Academy of Pediatrics National Conference and Exhibition, October 24-27, 2015.

Amartino H, Burton BK, Giugliani R, et al. New measure to assess severity of MPS II: the disease severity score. Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Burton BK, Jego V, Jones SA. Survival in idursulfase-treated and untreated patients with MPS II: data from the Hunter Outcome Survey (HOS). Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Burton BK, Hoganson GE, Charrow J, et al. Newborn screening for lysosomal disorders in Illinois. Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Friedman M, Valayannopoulos V, Camarene Grande C, Consuelo Sanchez A, Kane J, Kostyleva M, Tylki-Szymanska A, Sokal E, Sharma R, Rojas-Caro S, Wolfendale N, Burton BK. Safety findings from three trials of treatment with sebelipase alfa in children and adults with lysosomal acid lipase deficiency. Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Harmatz PR, Jester A, Mengel E, Treadwell M, Burton BK, Berger KI, Hendriksz CJ, Geberhiwot T, Sisic Z, Decker C. Impact of elosulfase alfa in patients with Morquio syndrome type A who have limited ambulation: an open-label phase 2 study. Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Hendriksz CJ, Burton BK, Alsayed MD, Giugliani R, Guelbert N, Hughes D, Mealiffe M, Mitchell JJ, Parini R, Raiman J, Shaywitz HJ, Slasor P, Solano Villarreal M, Stewart F, Berger KI, Itarmatz PR. Impact of long-term elosulfase alfa treatment on pulmonary function in patients with Morquio syndrome type A. Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Keating KG, Whiteaker L, Corkery J, Charrow J, Burton B. Pseudodeficiency of alpha-iduronidase is a common finding identified from newborn screening in the State of Illinois. Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Muenzer J, Burton BK, Harmatz P, Solano Villarreal ML, Amartino HM, Ruiz-Garcia M, Gonzalez Gutierrez-Solana L, Sciarappa K, Alexanderian D, Jones SA. A phase II/III intrathecal enzyme replacement therapy clinical trial for MPS II patients with cognitive impairment. Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Stewart F, Harmatz P, Braulin E, Bentley A, Burton B, Guffon N, Hale S, Johnston T, Kircher S, Kochhar P, Mitchell J, Plockinger U, Sisic Z. Pregnancy in individuals with mucopolysaccharidosis (MPS): a case series. Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Stewart F, Harmatz P, Braulin E, Bentley A, Burton B, Guffon N, Hale S, Johnston T, Kircher S, Kochhar P, Mitchell J, Plockinger U, Sisic Z. Management of fertility and pregnancy in individuals with mucopolysaccharidosis (MPS). Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Tylki-Szymanska A, Ficicioglu C, Morin I, Jurecka A, Burton B. Characteristics of patients with MPS II diagnosed at a very young age: data from the Hunter Outcome Survey (HOS). Presented at the Lysosomal Disease Network's WORLD Symposium, Feb 29-Mar 3, 2016.

Burton B, Jego V, Jones S. Survival in idursulfase-treated and untreated patients with MPS II: data from the Hunter Outcome Survey (HOS). Presented at the Annual Meeting of the American College of Genetics and Genomics, March 8-12, 2016.

Burton BK. An observational study to evaluate neurodevelopmental status in pediatric patients with Hunter syndrome: Study design and methodology. Presented at the Annual Meeting of the American College of Genetics and Genomics, March 8-12, 2016.
Wilson DP, Marulkar S, Tripuraneni R, Burton BK. Sebelipase alfa improves atherogenic measures in adults and children with lysosomal acid lipase deficiency. Presented at the Annual Meeting of the National Lipid Association, May 19-22, 2016.

Vockley J, Burton B, Berry G, et al. Interim results from an open-label phase 2 study to assess safety and clinical effects of investigational UX007 long chain fatty acid oxidation disorders (LC-FAOD). Poster presentation, Annual Meeting of the Society for Inherited Metabolic Disorders, Apr 3-6, 2016.

Longo N, Amato S, Vockley J, Wierenga K, Li H, Bilder D, Burton B, Dimmock D, Hardine C, Posner J, Thomas JA, Zori R, Greblikas F, Zhonghua G, Merilainen M, Weng HH, Levy H. Prism 301: An open-label, randomized phase 3 clinical trial evaluating efficacy and safety of pegvaliase for the treatment of adults with phenylketonuria. Poster

presentation, Annual Meeting of the Society for Inherited Metabolic Disorders, Apr 3-6, 2016.

Opladin T, Lindner M, Das A, Marquardt T, Khan A, Emre SH, Burton BK, et al. In vivo monitoring of urea cycle activity with 13C-acetate as a tracer of ureagenesis. Poster presentation, Annual Meeting of the Society for Inherited Metabolic Disorders, Apr 3-6, 2016.

Harmatz P Lin SP, Muenzer J, Giugliani R, Guffon N, Jego V, Burton B. Characteristics of patients aged 5 years and older at first signs and symptoms of mucopolysaccharidosis type II: data from the Hunter Outcome Survey (HOS). Poster presentation, Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Sep 6-9, 2016.

Reynders J, Burton B, DelAngel G. Novel LIPA mutations resulting in lysosomal acid lipase deficiency. Poster presentation, Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Sep 6-9, 2016.

Levy H, Harding C. Longo N, Bilder D, Burton B, Zori R, Posner J, Thomas J, Olbertz J, Rosen O, Bu Z, Merilainen M, Weng HH, Dimmock D. Phase 3 PRISM-2 long-term extension evaluating efficacy and safety of pegvaliase for treatment of adults with phenylketonuria. Poster presentation, Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Sep 6-9, 2016.

Harding C, Amato S, Vockley J, Wierenga K, Li H, Bilder D, Burton B, Dimmock D, Longo N, Posner J, Thomas J, Zori R, Rosen O, Greblikas F, Gu, K, Li M, Merilainen M, Weng HH, Levy H. Phase 3 PRISM-1 and PRISM-2 clinical trial results: to evaluate the efficacy and safety of pegvaliase for the treatment of adults with phenylketonuria (PKU). Poster presentation, Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Sep 6-9, 2016.

Longo N, Thomas J, Wasserstein M, Burton B, Vockley J, Grange D, Decker C, Weng HH, Li M, Schweighardt B, Zori R. Evaluation of long-term safety and efficacy of pegvaliase treatment for adults with phenylketonuria: updated 4-year results. Platform presentation, Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Sep 6-9, 2016.

Vockley J, Burton B, Berry GT, Longo N, Phillips J, Sanchez-Valle A, Tanpaiboon P, Grunewald S, Murphy E, Bowden A, Zhang L, Catalda J, Marsden D, Kakkis E. Interim results from open-label phase 2 study assessing the safety and clinical effects of investigational UX007 (triheptanoin) in subjects with long-chain fatty acid oxidation disorders (LC-FAOD). Poster presentation, Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Sep 6-9, 2016.

Burton BK, Marulkar S, Friedman M, Tripuraneni R, Furuya KN. Long-term benefit a sebelipase alfa over 76 weeks in children and adults with lysosomal acid lipase deficiency (LAL-D) (ARISE). Platform presentation, 13th Annual World Symposium. Feb 13-17, 2017.

Harmatz P, Lin S-P, Muenzer J, Giugliani R, Guffon N, Jego V, Burton BK, Characteristics of patients with mucopolysaccharidosis type II (MPS II) diagnosed aged < 5 years: data from the Hunter Outcome Survey (HOS). Poster presentation, 13th Annual WORLD Symposium, San Diego, CA, Feb 13-17, 2017.

Aleck KA, Tylki-Szymanska A, Ficicioglu C, Burton BK, Jego V, Guffon N. Urinary glycosaminoglycan levels in a mucopolysaccharidosis type II pediatric population receiving idursulfase therapy: data from the Hunter Outcome Survey (HOS) for patients aged < 18 months. Poster presentation. 13th Annual WORLD Symposium, San Diego, CA, Feb 13-17, 2017.

Hendriksz CJ, Parini R, Alsayed MD, Raiman J, Giugliani R, Mitchell JJ, Burton BK, et al. Elosulfase alfa treatment and changes in physical functioning and disability in Morquio syndrome type A. Poster presentation. 13th Annual WORLD Symposium, San Diego, CA, Feb 13-17, 2017.

Reynders J, Burton BK, del Angel G. Novel LIPA mutations resulting in lysosomal acid lipase deficiency. Poster presentation. 13th Annual WORLD Symposium, San Diego, CA, Feb 13-17, 2017.

Thomas J, Vockley J, Longo N, Wasserstein M, Burton B, et al. Long term safety and efficacy of pegvaliase treatment for adults with phenylketonuria: updated year 4 results from phase 2 PAL-003 extension. Presented at the 2017 Meeting of the American College of Medical Genetics, Phoenix, AZ, Mar 21-25, 2017.

Harding C, Thomas J, Levy H, Longo N, Bilder D, Burton B, et al. Phase 3 PRISM-2 long term extension evaluating efficacy and safety of pegvaliase for treatment of adults with phenylketonuria. Poster presentation. 2017 Meeting of the American College of Medical Genetics, Phoenix, AZ, Mar 21-25,2017.

Vockley J, Burton B, Berry G, et al. Results from a 78 week single-arm, open label phase 2 study to evaluate UX007 (triheptanoin) in pediatric and adult long chain fatty acid oxidation disorders (LC-FAOD). Presented at International Congress of Inborn Errors of Metabolism, Rio de Janeiro, Brazil, Sept 5-8, 2017.

Mayhew J, Vockley J, Burton B, et al. Assessments of exercise tolerance and muscle function in long chain fatty acid oxidation disorders (LC- FAOD): results from a phase 2 open label study of UX007. Presented at International Congress of Inborn Errors of Metabolism, Rio de Janeiro, Brazil, Sept.5-8, 2017.

Harmatz P, Lin S-P, Muenzer J, Giugliani R, Guffon N, Jego V, Burton B. Characteristics of patients with mucopolysaccaridosis type II (MPS II) diagnosed aged ≥ 5 years: data from the Hunter Outcome Survey (HOS). Presented at the International Congress of Inborn Errors of Metabolism, Rio de Janeiro, Brazil. Sept. 5-8, 2017.

Aleck K, Tylki-Szymanska A, Ficicioglu C, Burton B, Jego V, Guffon N. Urinary glycosaminoglycan levels in a mucopolysaccharidosis type II pediatric population aged ≤18 months receiving idursulfase therapy: data from the Hunter Outcome Survey (HOS). Presented at the International Congress of Inborn Errors of Metabolism, Rio de Janeiro, Brazil. Sept. 5-8, 2017.

Vockley J, Levy H, Amato S, Zori R, Thomas J, Burton B, et al,. Phase 3 PRISM-2 long term extension study evaluating efficacy and safety of pegvaliase for treatment of adults with phenylketonuria. Presented at the International Congress of Inborn Errors of Metabolism, Rio de Janeiro, Brazil.Sept. 5-8, 2017.

Lilienstein J, Burton B, Grant M, et al. Interim analysis of the phenylketonuria (PKU) patients enrolled in the PKUDOS registry. Poster presentation. International Congress of Inborn Errors of Metabolism, Rio de Janeiro, Brazil. Sept 5-8, 2017.

Goodman Z, Friedman M, Burton B. Change in liver histopathology in a diverse population of children and adults with lysosomal acid, lipase deficiency after 48 and 96 weeks of sebelipase alfa therapy. Presented at the Annual Meeting of the American Association for the Study of Liver Diseases, Oct 20-24, 2017.

Burton B, Feilet F, Furuya K, Friedman M, Marulkar S, Balwani M. Long-term benefit of sebelipase alfa over 100 weeks in children and adults with lysosomal acid lipase deficiency (ARISE STUDY). Presented at the Annual Meeting of the American Association for the Study of Liver Diseases, Oct 20-24, 2017.

Burton B, Sanchez AC, Kostyleva M, Allen K, Friedman M, Martins AM. Long-term benefit of sebelipase alfa over 96 weeks in a diverse population of children and adults with lysosomal acid lipase deficiency. Presented at the Annual Meeting of the American Association for the Study of Liver Diseases, Oct 20-24, 2017.

Goodman Z, Friedman M, Burton B. Change in liver histopathology in a diverse population of children and adults with lysosomal acid lipase deficiency after 48 and 96 weeks of sebelipase alfa therapy. Presented at the Annual Meeting of the American Association for the Study of Liver Diseases, Oct 20-24, 2017.

Harmatz P, Muenzer J, Burton BK, Ficicoglu C, Lau HA, Leslie ND, Conner E, Wong Po Foo C, Vaidya S, Wechsler T, Whitley CB. Update on phase 1/2 clinical trials for MPSI and MPS II using ZFN-mediated in vivo genome editing. Presented at 14th Annual WORLD Symposium, San Diego, Feb 6-8, 2018.

Jones SA, Burton BK, Botha J, Whiteman DAH. Profile of natural history in 104 patients with mucopolysaccharidosis type II. Insights from the Hunter Outcome Survey (HOS) Presented at the 14th Annual WORLD Symposium, San Diego,
Feb 6-8, 2018.

Burton BK, Lampe C, Lagler FB, Botha J, Whiteman DAH. Infusion-related reactions in patients with mucopolysaccharidosis type II on Idursulfase. Presented at the 14th Annual WORLD Symposium, San Diego, Feb 6-8, 2018.

Gillis J, Burton B, Inbar-Fergenberg M, Mackrell M, Phornphutkul C, Stockton DW, Khan A. North American experience with laronidase enzyme replacement therapy for mucopolysaccharidosis type I in a home infusion setting. Presented at the 14th Annual WORLD Symposium, San Diego, Feb 6-8, 2018.

Lilienstein J, Burton B, Grant M, et al. Interim analysis of the phenylketonuria (PKU) patients enrolled in the PKUDOS registry. Poster presentation. 40th Annual Meeting of the Society for Inherited Metabolic Disorders, San Diego, March 11-14, 2018.

Vockley J, Burton B, Berry GT, et al. Results from a 78-week single-arm, open-label phase 2 study to evaluate UX007 in pediatric and adult patients with moderate to severe long-chain fatty acid oxidation disorders (LC-FAOD). Poster presentation. 40th Annual Meeting of the Society for Inherited Metabolic Disorders, San Diego, March 11-14, 2018.

Burton B, Cederbaum S, Jurecki E, Lilienstein J, Alverez I, Cohen-Pfeffer J, Irwin D, Levy H, Rohr F. Prevalence of comorbid conditions among phyenylketonuria patients: a retrospective study of US health insurance claims data. Poster presentation. Annual Meeting of the American College of Medical Genetics and Genomics, Charlotte, April 10-14, 2018.

Jones SA, Burton B, Botha J, Whiteman D. Profile of natural history in patients with mucopolysaccharidosis type II: insights from the Hunter Outcome Survey (HOS). Platform presentation, 15th International Symposium on MPS and Related Diseases, San Diego, Aug 2-4, 2018.

Aleck K, Tylki-Szymanska A, Ficicioglu C, Burton B, Jego V, Guffon N. Urinary Glycosaminoglycan levels in a mucopolysaccharidosis type II pediatric population' Aged <18 months rece3iving idursulfase therapy: data from the Hunter Outcome Survey (HOS). Poster presentation, 15th International Symposium on MPS and Related Diseases, San Diego, Aug 2-4, 2018.

Burton B, Lampe C, Lagler F, Botha J, Whiteman D. Infusion-related reactions in patients with mucopolysaccharidosis type II on idursulfase enrolled in the Hunter Outcome Survey (HOS). Poster presentation, 15th International Symposium on MPS and Related Diseases, San Diego, Aug 2-4, 2018.

Muenzer, J, Burton BK, Harmatz P, Gutierrez-Solana LG, Ruiz-Garcia M, Jones SA, Guffon N, Inbar-Feigenberg M, Bratkoviv D, Wu Y, Alexanderian D. Efficacy and safety of intrathecal idursulfase in pediatric patients with Hunter syndrome and early cognitive impairment. Poster presentation, Annual Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM), Athens, Greece, Sept 3-7, 2018.

Burton BK, Harding CO, Thomas JA, Longo N, Posner J, Dimmock D, Zori R, Weng

HH, Olbertz J, Gershman A, Rosen O, Gupta S, Jones S, Gu Z, Vockley J. Long-term safety of induction, titration and maintenance dosing of pegvaliase treatment in adults with phenylketonuria. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM), Athens, Greece, Sept 3-7, 2018.
Harding CO, Thomas JA, Burton BK, Zori R, Dimmock D, Vockley J, Weng HH, Olbertz J, Gershman A, Rosen O, Jones S, Li M, Longo N. Phase 3 PRISM clinical trials: characterization of hypophenylalaninemia in pegvaliase treated adults with PKU. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM), Athens, Greece, Sept. 3-7, 2018.

Burton B. Longo N, Singh R, Stuy M, Vockley J, van Bockle J, Lane P, Alvarez I, Lilienstein J, Jurecki E. The burden of illness in adults with phenylketonuria (PKU): an interim analysis of a cross-sectional study. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM), Athens, Greece, Sept. 3-7, 2018.

Muntau AC, Feillet F, Burton B, MacDonald A, Wessel A, Alvarez I, Lilienstein J, Lane P, Jurecki E, Longo N. A meta-analysis of growth outcomes in phenylketonuria patients treated with a phenylalanine-restricted diet and sapropterin. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM), Athens, Greece, Sept. 3-7, 2018.

Feillet F, Ficicioglu C, Lagler FB, Longo N, Alm J, Muntau AC, Burlina A, Belanger-Quintana A, Trefz FK, Kittus R, Jurecki E, Alvarez I, Lilienstein J, Burton B on behalf of the KAMPER and PKUDOS investigators. An interim analysis of the KAMPER and PKUDOS registries: efficacy and safety of sapropterin before and during pregnancy. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM), Athens, Greece, Sept. 3-7, 2018.

Muenzer J, Burton BK, Harmatz P, Amartino H, Jones SA, Gutierrez-Solana LG, Ruiz-Garcia M, Wu Y, Alexanderian D. Neurodevelopmental status and adaptive behavior of pediatric patients with hunter syndrome in a 2-year observational study. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Athens, Greece, Sept. 3-7, 2018.

Muenzer J, Burton BK, Harmatz P, Botha J, Kampmann C. Evaluation of the long-term Treatment effects of idursulfase using statistical modeling: data from the Hunter Outcome Survey (HOS). Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Athens, Greece, Sept. 3-7, 2018.

Burton BK, Lampe C, Lagler FB, Botha J, Whiteman DAH. Infusion-related reactions In patients with mucopolysaccharidosis type II on idursulfase. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Athens, Greece, Sept. 3-7, 2018.

Burton B, Sanchez AC, Kostyleva M, Allen K, Abel F. Effect of sebelipase alfa on liver parameters over 96 weeks in a diverse population of children and adults with lysosomal

acid lipase deficiency. Poster presentation. EASL-NAFLD 2018 Summit, Geneva, Switzerland, Sept. 20-22.

Muenzer J, Prado C, Lau HA, Burton B, Ficicioglu C, Wong PFC, Vaidya SA, Whitley CB, Harmatz P. Novel treatment of MPS II (Hunter syndrome) with SB-913 ZFN-mediated in vivo human genome editing: Update from a Phase 1/2 clinical trial. Oral presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Athens, Greece, Sept. 3-7, 2018.

Burton, B, Cederbaum S, Jurecki E, Lilienstein J, Alvarez I, Cohen-Pfeffer J, Irwin D, Levy H, Rohr F, Jones KB. Prevalence of comorbidities among phenylketonuria patients-A retrospective study of US health insurance claims data. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Athens, Greece, Sept. 3-7, 2018.

Burton BK, Hoganson GE, Grange DK, Braddock SR, Christensen KM, Hitchins L, Hickey R, Shao R, Basheeruddin K. Newborn screening for mucopolysaccharidosis type II (MPS II) in Illinois: The first year's experience. Platform presentation, 15th Annual WORLS Symposium, Orlando, Feb 4-8, 2019

Muenzer J, Burton BK, Harmatz P, Amartino H, Jones SA, Gutierrez-Solana LG, Ruiz-Garcia M, Wu Y, Alexanderian D. Neurodevelopmental status and adaptive behavior of pediatric patients with Hunter syndrome: A longitudinal observational Study. Poster presentation. 15th Annual WORLD Symposium, Orlando, Feb 4-8, 2019.

Muenzer J, Burton BK, Harmatz P, Botha J, Kampmann C. Evaluation of the long-term treatment effects of idursulfase using statistical modelling: Data from the Hunter Outcome Survey (HOS). Poster presentation. The 15th Annual WORLD Symposium, Orlando, Feb 4-8, 2019

Muenzer J, Burton BK, Kampmann C, Botha J, Jones SA. Characteristics of patients with mucopolysaccharidosis type II who have received a bone marrow transplant: Data from the Hunter Outcome Survey. Poster presentation. The 15th Annual WORLD Symposium, Orlando, Feb 4-8, 2019.

Muenzer J, Prada CE, Burton B, Lau HA, Ficicioglu C, Po Foo CW, Vaidya SA, Whitley CB, Harmatz P. CHAMPIONS: A phase 1/2 clinical trial with dose escalation of SB-913 ZFN-mediated in vivo human genome editing for treatment of MPS II (Hunter Syndrome). Platform presentation. The 15th Annual WORLD Symposium, Orlando, Feb 4-8, 2019.

Harding CO, Thomas JA, Burton, BK, Zori R, Dimmock D, Vockley J, Weng HH, Olbertz J, Gershman A, Rosen O, Jones S, Li M, Longo N. Characterization of hypophenylalaninemia in pegvaliase-treated adults with phenylketonuria. Poster presentation. Annual Meeting of the American College of Medical Genetics and Genomics, Seattle, WA, April 3-6, 2019

Burton BK, Harding CO, Thomas JA, Longo N, Posner J, Dimmock D, Zori R, Weng HH, Olbertz J, Gershman A, Rosen O, Jones S, Gu Z, Vockley J. Long-term safety of induction, titration and maintenance dosing of pegvaliase treatment in adults with phenylketonuria. Poster presentation. Annual Meeting of the American College of Medical Genetics and Genomics, Seattle, WA, April 3-6, 2019.

Paras A, Vucko E, Bausell H, Havens K, Philipp T, Arduini K, Johnson A, Kalb F, Katz R, Shim S, Adams J, Shively V, Widera S, Burton, BK. Pegvaliase treatment protocol and patient outcomes: experience of a single PKU treatment center. Poster presentation. Annual Meeting of the American College of Medical Genetics and Genomics, Seattle WA, April 3-6, 2019.

Burton B, Longo N, Maillot F, Rahman Y, Singh R, Sivri HS, Stuy M, Vockley J, Van Backle J, Jha A, Lane P, Lilienstein J, Jurecki E. The burden of illness in adults with phenylketonuria (PKU): interim analysis of a cross-sectional study. Poster presentation. Annual Meeting of the Society for Inherited Metabolic Disorders, Seattle WA, April 6-9, 2019.

Vockley J, Zori R, Thomas J, Stuy M, Burton BK, Longo N, Rosen O, Gu Z, Olbertz J, Weng HH. Subgroup analysis of pegvaliase in adults with phenylketonuria in Phase 3 PRISM studies: evaluation efficacy and safety based on previous sapropterin response or dietary phenylalanine intake. Poster presentation. Annual Meeting of the Society for Inherited Metabolic Disorders, Seattle WA, April 6-9, 2019.

Rohr F, Burton BK, Longo N, Thomas J, Harding C, Rosen O, Gu Z, Olbertz J, Weng HH. Phase 3 PRISM clinical trials: evaluation change in diet with pegvaliase treatment in adults with phenylketonuria. Poster presentation. Annual Meeting of the Society for Inherited Metabolic Disorders, Seattle WA, April 6-9, 2019.

Muntau A, Feillet F, Burton B, MacDonald A, Wessel A, Alvarez I, Lilienstein J, Lane P, Jurecki E, Longo N. A meta-analysis of growth outcomes in phenylketonuria Patients treated with phenylalanine-restricted diet + sapropterin. Poster presentation, Annual Meeting of the Society for Inherited Metabolic Disorders, Seattle WA, April 6-9, 2019.

Feillet F, Ficicioglu C, Lagler FB, Longo N, Alm J, Muntau A, Burlina A, Belanger-Quintana A, Trefz FK, Jurecki E, Alvarez I, Lilienstein J, Burton B, on behalf of the KAMPER and PKUDOS investigators. An interim analysis of the KAMPER and PKUDOS registries: efficacy and safety of sapropterin before and during pregnancy. Poster presentation. Annual Meeting of the Society for Inherited Metabolic Disorders, Seattle WA, April 6-9, 2019.

Vucko E, Paras A, Bausell H, Havens K, Phillip T, Arduini K, Johnson A, Kalb F, Katz R, Shim S, Adams J, Shively V, Widera S, Burton BK. Pegvaliase treatment protocol and patient outcomes: experience of a single PKU treatment center. Annual Meeting of the Society for Inherited Metabolic Disorders, Seattle WA, April 6-9, 2019.

Burton BK. Newborn Screening for Lysosomal Storage Disorders. Platform presentation. 5th International Forum for Lysosomal Storage Disorders. Tokyo, Japan. July 11-13, 2019

Vockley J, Zori R, Stuy M, Burton BK, Longo N, Rosen O, Gu Z, Olbertz J, Weng HH. Efficacy and safety of pegvaliase by sapropterin response or dietary phenylalanine intake in adults with phenylketonuria. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Rotterdam, Netherlands, Sept. 2-6, 2019

Burton BK, Cederbaun S, Lane P, Jurecki E, Lilienstein J, Ali I, Irwin D, Levy H, Rohr F, Jones KB. 50 most prevalent comorbidities among phenylketonuria patients- A retrospective study of US health insurance claims data. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Rotterdam, Netherlands, Sept 2-6, 2019.

Burton BK, Northrup H, Zori R, Posner J, Olbertz J, Lounsbury D, Weng HH, Vockley J. Phase 3 PRISM studies: efficacy and safety of pegvaliase 60 mg dose in adult patients with phenylketonuria. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Rotterdam, Netherlands, Sept. 2-6, 2019

Longo N, Thomas J, Jurecki E, Lane P, Olbertz J, Gershon A, Wang B, Harding CO, Burton BK, Rohr F, van Calcar S. Dietary intakes and adverse events in pegvaliase-treated phenylketonuric adults who had low blood phenylalanine concentrations. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Rotterdam, Netherlands, Sept 2-6, 2019.

Longo N, Rohr F, Burton BK, Thomas J, Harding CO, Rosen O, Gu Z, Olbertz J, Weng HH. Phase 3 PRISM clinical trials: evaluating change in diet with pegvaliase treatment in adults with phenylketonuria. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Rotterdam, Netherlands, Sept. 2-6, 2019

Muenzer J, Burton BK, Kampmann C, Botha J, Jones SH. Characteristics of patients With MPS II who have received a bone marrow transplant. Data from the Hunter Outcome Survey (HOS). Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Rotterdam, Netherlands, Sept. 2-6, 2019

Burton BK. Evaluating short and long-term management goals for lysosomal storage Diseases. Symposium platform presentation. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism, Rotterdam, Netherlands, Sept. 2-6

Burton B, Sanchez AC, Kostyleva M, Martins AM, Marulkar S, Abel F, Baric I. Efficacy and safety of sebelipase alfa over 144 weeks in a diverse population of children and adults with lysosomal acid lipase deficiency .Poster presentation. 70th Annual Meeting of the American Association for the Study of Liver Diseases, Boston, MA, Nov 8-12, 2019.

Burton B, Feillet F, Furuya K, Marulkar S, Balwani M. Benefits of sebelipase alfa in Children and adults with lysosomal acid lipase deficiency are sustained for up to 5 years (ARISE Study). Poster presentation. 70th Annual Meeting of the American Association of for the Study of Liver Diseases, Boston, MA, Nov 8-12, 2019.

Burton B, Guffon N, Ramaswami U, Chien Y-H, Hwu W-L, Lauw M, Longo N, Ngu LH, Pan W, Mitchell J. Long term treatment with elosulfase alfa has an acceptable safety Profile for patients with Morquio syndrome type A: Real-world results from the Morquio A Registry Study (MARS). Poster presentation. WORLD Symposium 2020, Orlando, FL Feb 10-13, 2020.

Harmatz P, Jayakar P, Pena LDM, Cattaneo F, Ceravolo F, Paleari Y, Sutton VR, Burton BK, Longo N. A new randomized placebo-controlled study to establish the safety and efficacy of velmanase alfa (human recombinant alpha-mannosidase) enzyme replacement therapy for the treatment of alpha-mannosidosis. Poster presentation. WORLD Symposium 2020. Orlando, Florida, Feb 10-13, 2020.

Kim KK, Vucko E, Desai AK, Kishnani P, Burton BK. Development of high sustained IgG antibody titers and corresponding clinical decline in an adolescent with atypical infantile Pompe disease after 11+ years on enzyme replacement therapy with alglucosidase alfa. Poster presentation. WORLD Symposium 2020. Orlando, Florida Feb 10-13, 2020.

Mitchell J, Ramaswami U, Longo N, Chien Y-H, Guffon N, Hwu W-L, Lauw M, Ngu, LH, Pan W, Burton B. Long term clinical outcomes of patients treated with elosulfase alfa: five-year real-world results from the Morquio A Registry Study (MARS). Poster presentation. WORLD Symposium 2020. Orlando, Florida, Feb 10-13, 2020.

Burton BK, Northrup H, Zori RT, Posner J, Olbertz J, Lounsbury D, Weng HH, Vockley J. Phase 3 PRISM studies: efficacy ad safety of pegvaliase 60 mg dose in adult patients with phenylketonuria. American College of Medical Genetics and Genomics Annual Meeting 2020 (virtual).

Burton BK, Rohr F, Longo N, Thomas JT, Harding CO, Rosen O, Gu Z, Olbertz J, Weng HH. Evaluating change in diet with pegvaliase treatment in adults with phenylketonuria; Results from phase 2 and phase 3 clinical trials. American College of Medical Genetics and Genomics Annual Meeting 2020 (virtual).

Thomas JA, Jurecki E, Lane P, Olbertz J, Wang B, Longo N, Harding CO, Burton BK, Rohr F, van Calcar S. Dietary intakes and adverse events in pegvaliase treated phenylketonuria adults who has low blood phenylalanine concentrations. American College of Medical Genetics and Genomics Annual Meeting 2020 (virtual).

Burton BK, Ilan AB, Delaney K, Madden DT, Jurecki E, Longo N, Harding CO, Thomas J. Concept elicitation and outcomes assessment tool mapping with an international cohort of adult phenylketonuria patients. American College of Medical Genetics and Genomics

Annual Meeting 2020 (virtual).

Burton B, Longo N, Maillot F, Rahman Y, Singh R, Sivri HS, Stuy M, Vockley J, Van Backle J, Jha A, Lane P, Lilienstein J, Jurecki E. The burden of illness in adults with phenylketonuria (PKU); interim analysis of a cross-sectional study conducted in North America and Europe. Mol Genet Metab 2020; 127: 259 (abstract accepted for presentation at meeting of Society for Inherited Metabolic Disorders).

Feillet F, Ficicioglu C, Lagler F, Longo N, Alm J, Muntau A, Burlina A, Berlanger-Quintana A, Trefz F, Jurecki E, Alvarez I, Lilienstein J, Burton B. An interim analysis of The Kamper and PKUDOS registries: efficacy and safety of sapropterin before and during pregnancy. Mol Genet Metab 2020; 127: 267 (abstract accepted for presentation at annual meeting of the Society for Inherited Metabolic Disorders).

Hitchins L, Charrow J, Burton B. A single-center experience with newborn screening for Pompe disease. Mol Genet Metab 2020; 127: 275 (abstract accepted for presentation at the annual meeting of the Society for Inherited Metabolic Disorders).

Rosenfeld JA, Kim KH, Burrage LC, Lu S, Wangler MF, Yamamoto S, Kanca O, Hicks MJ, Kayani S, Stetler M, Undiagnosed Diseases Network, Lee B, Bacino CA, Bellen HJ, Burton BK, Craigen WJ. HIP1R may be associated with a recessive, progressive neurologic disorder. American Society for Human Genetics Annual Meeting 2020.

Quadri A, Kim KH, Hickey R, Paras A, Baker J, Charrow J, Burton B. Population based Newborn screening for mucopolysaccharidosis type II: a single center's experience. 2021 WORLD Symposium on Lysosomal Disorders. Feb 8-12.

Muenzer J, Burton BK, Scarpa M, Botha J, Giugliani R. Genotype-phenotype findings in patients with mucopolysaccharidosis II (MPS II): data from the Hunter Outcome Survey (HOS). Presented at the 2021 WORLD Symposium on Lysosomal Disorders, Feb 8-12.

Muenzer J, Burton BK, Harmatz P, et al. Long-term safety and efficacy of intrathecal idursulfase-IT in patients with neuronopathic mucopolysaccharidosis II: 2-year results from a Phase 2/3 extension study. Presented at the 2021 WORLD Symposium on Lysosomal Disorders. Feb 8-12.

Muenzer J, Burton, BK, Harmatz, et al. Comparison of cognitive function in sibs with neuronopathic mucopolysaccharidosis II: evaluation of early treatment with IV idursulfase and intrathecal idursulfase-IT. Presented at the 2021 WORLD Symposium on Lysosomal Disorders. Feb 8-12.

Muenzer J, Burton BK, Harmatz P, et al. Single-arm open-label, phase 2/3 substudy and extension evaluating safety and efficacy of intrathecal idursulfase-IT in patients younger than 3 years old with neuronopathic mucopolysaccharidosis II. Presented at the 2021 WORLD Symposium on Lysosomal Disorders. Feb 8-12.

Harmatz P, Bakardijiev AI, Escolar M, Burton BK, et al. Design and preliminary results of a first in-human 24 week study of intravenous DNL310 (brain penetrant IDS fusion Protein) in MPS II. Presented at the 2021 WORLS Symposium on Lysosomal Disorders, Feb 8-12.

Quadri A, Kim KH, Hickey R, Paras A, Baker J, Charrow J, Burton B. Newborn screening for mucopolysaccharidosis type II: a single center's experience. Presented at the Annual Meeting of the American College of Medical Genetics and Genomics. April 13-16, 2021.

Muenzer J, Burton BK, Scarpa M, Botha J, Giugliani R. Genotype-phenotype findings in patients with mucopolysaccharidosis II (MPS II): data from the Hunter Outcome Survey (HOS). Presented at the Annual Meeting of the American College of Medical Genetics and Genomics, April 13-16, 2021.

Muenzer J, Burton BK, Harmatz P, et al. Long-term safety and efficacy of intrathecal idursulfase-IT in patients with neuronopathic mucopolysaccharidosis II: 2-year results from a Phase 2/3 extension study. Presented at the Annual Meeting of the American College of Medical Genetics and Genomics, April 13-16, 2021.

Muenzer J, Burton, BK, Harmatz, et al. Comparison of cognitive function in sibs with neuronopathic mucopolysaccharidosis II: evaluation of early treatment with IV idursulfase and intrathecal idursulfase-IT. Presented at the Annual Meeting of the American College of Medical Genetics and Genomics, April 13-16, 2021.

Muenzer J, Burton BK, Harmatz P, et al. Single-arm open-label, phase 2/3 substudy and extension evaluating safety and efficacy of intrathecal idursulfase-IT in patients younger than 3 years old with neuronopathic mucopolysaccharidosis II. Presented at the Annual Meeting of the American College of Medical Genetics and Genomics, April 13-16, 2021.

Bodamer O, Burton B, Ile J, Cohn G, White Y, Kane E, Diaz G. The pheNIX trial: first-in-human gene therapy trial for PKU due to phenylalanine hydroxylase (PAH) deficiency. Presented at the Annual Meeting of the American College of Medical Genetics and Genomics, April 13-16, 2021.

Blok LS, Verseput J, Kit D, et al (including BBurton). A clustering of missense variants in the crucial chromatin modifier WDR5 defines a new neurodevelopmental disorder. Presented at the Annual Meeting of the European Society of Human Genetics, June 12-15, 2021.

Burton BK. Arginase-1 deficiency: report of a patient followed for 15 years. Poster Presentation. International Congress of Inborn Errors of Metabolism (ICIEM), Sydney, Australia, Nov 20-23, 2022.

Muenzer J, Scarpa M, Tylki-Szymanska A, Amartino H, Harmatz P, Lin S-P, Link B,

Molter D, Raiman J, Whiteman DH, Botha J, Jakukeviviene D, Fertek D, Audi J, Burton BK. Fifteen years of the Hunter Outcome Survey (HOS): real-world insights into the patient population living with mucopolysaccharidosis type II (MPS II). Platform presentation, WORLD Symposium on Lysosomal Disorders 2022, San Diego, Feb 7-11, 2022.

Quadri A, Shively V, Burton BK. Outcome in infants treated with very early ERT supports newborn screening for mucopolysaccharidosis type II. Poster presentation. WORLD Symposium on Lysosomal Disorders 2022, San Diego, Feb 7-11, 2022.

Walsh C, Hickey RA, Charrow JC, Burton BK, Quadri A, Rubin JA, Kim K, Hitchins L, Baker J. Krabbe disease newborn screening: a single center's experience.. Poster presentation. WORLD Symposium on Lysosomal Disorders 2022, San Diego, Feb 7-11, 2022.

Mitchell J, Burton BK, Bober MB, et al. Long-term outcomes of MPS IVA patients treated with elosulfase alfa: findings from the Morquio A Registry Study (MARS) after 6 years. Platform presentation. WORLD Symposium on Lysosomal Disorders 2022, San Diego, Feb 7-11, 2022.

Sacharow S, Northrup H, Whitehall KB, Rowell R, Lindstrom K, Burton B, Thomas J. Efficacy and safety of the recommended pegvaliase dosing regimen in adults with Phenylketonuria in the phase 3 PRISM studies. Platform presentation. Annual Meeting of the American College of Medical Genetics and Genomics (ACMG), Nashville, TN, March 22-26, 2022

Burton BK, Andersson HC, Clague GC, et al. Long-term safety and efficacy of sapropterin: final results of the phenylketonuria demonnstration, outcomes and safety (PKUDOS) registry. Poster presentation. Annual Meeting of the American College of Medical Genetics and Genomics (ACMG), Nashville TN, March 22-26, 2022.

Hickey RA, Rubin JP, Whitehead J, Burton BK. Newborn screening for X-linked adrenoleukodystrophy: identification of isodisomy of X in an affected female infant. Poster presentation. Annual Meeting of the American College of Medical Genetics and Genomics (ACMG), Nashville, TN, March 22-26, 2022.

Vockley J, Burton B, Berry G, et al. Triheptanoin for the treatment of long-chain fatty Acid oxidation disorders (LC-FAOD): final results of an open-label long-term extension study. Platform presentation. Annual Meeting of the American College of Medical Genetics and Genomics (ACMG), Nashville, TN, March 22-26, 2022.

Sacharow S, Northrup H, Whitehall KB, Rowell R, Lindstrom K, Burton B, Thomas J. Efficacy and safety of the recommended pegvaliase dosing regimen in adults with Phenylketonuria in the phase 3 PRISM studies. Poster presentation, Annual Meeting of Society for Inherited Metabolic Disorders 2022, April 10-13, Orlando, Florida.

Muenzer J, Amartino H, Burton BK, Giugliani R, Harmatz P, Lin S-P, Link B, Molter D, Ramaswami U, Scarpa M, Botha J, Audi J. Exploring the unmet needs of adults living with mucopolysaccharidosis II (MPS II): data from the Hunter Outcome Survey (HOS). Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM), Freiburg, Germany, Aug 29-Sept 1, 2022

Feillet F, Ficicioglu C, Lagler FB, Longo N, Alm J, Muntau AC, Burlina A, Belanger-Quintana A, Trefz FK, Lillienstein J, Clague GE, Rowell R, Burton BK. Efficacy and safety of sapropterin before and during pregnancy: an interim analysis of the Kuvan Advanced Maternal Paediatric European Registry (KAMPER) and Phenylketonuria Developmental Outcomes and Safety (PKUDOS) PKU-MOMS sub-registry. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM), Freiburg, Germany, Aug 29-Sept 1, 2022.

Burton B, Sacharow S, Northrup H, Whitehall K, Rowell R, Lindstrom K, Thomas J. Efficacy and safety of the recommended pegvaliase dosing regimen in adults with phenylketonuria in the phase 3 PRISM studies. Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM), Freiburg, Germany, Aug 29-Sept 1, 2022

Stepien KM, Burton B, Bober MB, et al. Long-term outcomes in adults with MPS IVA treated with elosulfase alfa: findings from the Morquio A Registry Study (MARS). Poster presentation. Annual Meeting of the Society for the Study of Inborn Errors of Metabolism (SSIEM), Freiburg, Germany, Aug 29-Sept 1, 2022

Muenzer J, Harmatz P, Burton BK, Rajan D, Jones S, Chen C, et al. Interim 73-week cohorts A,B and C results of a phase 1/2 study of intravenous DNL310 (brain-penetrant enzyme replacement therapy) in MPS II. Presented at the Annual Meeting of the Society For the Study of Inborn Errors of Metabolism (SSIEM), Freiburg, Germany, Aug 29-Sept. 1, 2022

Stepien KM, Burton B, Bober M, et al. Long-term outcomes in adults with MPS IVA t treated with elosulfase alfa: findings from the Morquio A Registry Studay (MARS). Poster presentation. Brazilian Congress of Medical Genetics, Sept. 28-Oct.1, 2022.

Burton B, Sacharow S, Northrup H, Whitehall K, Rowell R, Lindstrom K, Thomas J. Efficacy and safety of the recommended pegvaliase dosing regimen in adults with phenylketonuria in the phase 3 PRISM studies. Poster presentation. Brazilian Congress of Medical Genetics, Sept. 28-Oct. 1, 2022

Burton BK, Shively V, Quadri A, et al. Newborn screening for mucopolysaccharidosis type II (MPS II): lessons learned. Platform presentation. WORLD Symposium 2023, Orlando, FL, Feb 21-6, 2023.

Burton BK, Guffon N, Roberts J, et al. Safety profile of idursulfase administered at home In patients with mucopolysaccharidosis II (MPS II) enrolled in the Hunter Outcome

Survey. Poster presentation. WORLD Symposium 2023, Orlando, FL, Feb 21-26, 2023.

Reisewitz P, Hinds D, Dosenovic S, Ma Y, Jha A, Hunt A, Burton B, Guffon N. Real-world impact of enzyme replacement therapy on endurance in patients with MPS IVA. Poster presentation. WORLD Symposium 2023, Orlando, FL, Feb 21-26, 2023.

Burton BK, Ficicioglu C, Pedro H, Pathak R, Robinson I, Wright E, Muenzer J. Clinical characteristics and management of patients with mucopolysaccharidosis II (MPS II) in The United States from the Hunter Outcome Survey. Poster presentation. WORLD Symposium 2023, Orlando, FL, Feb 21-26, 2023.

Burton BK, Ficicioglu C, Pedro H, Pathak R, Robinson I, Wright E, Muenzer J. Clinical characteristics and management of patients with mucopolysaccharidosis II (MPS II) in The United States from the Hunter Outcome Survey. Poster presentation. Annual Meeting of the American College of Medical Genetics and Genomics (ACMG), Salt Lake City, Utah, March 14-18, 2023.

Carey M, Edick MJ, Justice K, Burton B. Neurological and psychological comorbidities In 36 individuals with MSUD enrolled in the IBEM-IS database. Poster presentation. Annual Meeting of the Society for Inherited Metabolic Disorders, Salt Lake City, Utah, March 18-21, 2023.

Schiff M, Schwahn BC, Chabrol B, Merritt JL, Vockley J, Vernon H, Berry GT, Santra S Lee C, Koeberll D, Li H, Burton B, De las Heras J, Diaz G, Faria-Urbina M, Luo J, Attarwala H, Sikirica V, Liang M, Guey LT, Lukacs C, Martini PGV, Glaser R, Carrillo N. MaP natural history study: clinical and biomarker findings in methylmalonic acidemia due to MUT deficiency. Poster presentation. Annual Meeting of the Society for Inherited Metabolic Disorders, Salt Lake City, Utah, March 18-21, 2023.

Schwahn B, Berry GT, Santra S, Vernbon H, Li H, Merritt JL, Schiff M, Chabrol B, De las Heras J, Vockley J, Lee C, Koeberl D, Burton B, Grunewald S, Morgan T, Diaz G, Ficicioglu C, Luo J, Attarwala H, Sikirica V, Liang M, Guey LT, Lukacs C, Martini PGV, Glaser R, Carrillo N. MaP natural history study: clinical and biomarker findings in propionic acidemia. Poster presentation. Annual Meeting of the Society for Inherited Metabolic Disorders, Salt Lake City, Utah, March 18-21, 2023.

Schwahn B, Berry GT, Santra S, Vernbon H, Li H, Merritt JL, Schiff M, Chabrol B, De las Heras J, Vockley J, Lee C, Koeberl D, Burton B, Grunewald S, Morgan T, Diaz G, Ficicioglu C, Luo J, Attarwala H, Sikirica V, Liang M, Guey LT, Lukacs C, Martini PGV, Glaser R, Carrillo N. MaP natural history study: clinical and biomarker findings in propionic acidemia. Poster presentation. Pediatric Academic Societies (PAS) Annual , Meeting, Washington, DC, Apr 27-May 1, 2023.

Schiff M, Schwahn BC, Chabrol B, Merritt JL, Vockley J, Vernon H, Berry GT, Santra S Lee C, Koeberll D, Li H, Burton B, De las Heras J, Diaz G, Faria-Urbina M, Luo J, Attarwala H, Sikirica V, Liang M, Guey LT, Lukacs C, Martini PGV, Glaser R, Carrillo

N. MaP natural history study: clinical and biomarker findings in methylmalonic acidemia due to MUT deficiency. Poster presentation. Pediatric Academic Societies (PAS) Annual Meeting, Washington DC, Apr 27-May 1, 2023.

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