CURRICULUM VITAE

Name: Edwin W. Naylor
Born: 1941 – Gloversville, NY

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EDUCATION AND TRAINING

Undergraduate:

1959 - 1964	Middlebury College Middlebury, Vermont	A.B. - 1964 Biology/Chemistry

Graduate:

1967 - 1971	Utah State University Logan, Utah	Ph.D. - 1971 Genetics/Microbiology
1971 - 1972	University of Pittsburgh Graduate School of Public Health	M.P.H. - 1974 Population/ Human Genetics

Post - Doctoral Training

1972 - 1974	Division of Human Genetics Department of Pediatrics Children's Hospital of Buffalo State University of New York Buffalo, New York	Post-doctoral Fellow Robert Guthrie, M.D., Ph.D.
1971 - 1972	Population Division Graduate School of Public Health University of Pittsburgh Pittsburgh, Pennsylvania	Post-doctoral Fellow John Cutler, M.D.

APPOINTMENTS AND POSITIONS

Academic

2005 - Present	Medical University of South Carolina Charleston, South Carolina	Adjunct Professor of Pediatrics (Genetics)
1990 - 2005	University of Pittsburgh Department of Human Genetics Graduate School of Public Health	Adjunct Associate Professor Human Genetics
1991	Duquesne University	Adjunct Faculty

1986 - 1990	University of Pittsburgh Department of Human Genetics Graduate School of Public Health	Adjunct Assistant Professor of Human Genetics
1984 - 1986	University of Pittsburgh Graduate School of Public Health	Assistant Professor of Human Genetics
1984 - 1986	University of Pittsburgh School of Medicine	Assistant Professor of Obstetrics and Gynecology
1981 - 1984	State University of New York at Buffalo	Research Associate Professor of Pediatrics
1977 - 1981	State University of New York at Buffalo	Research Assistant Professor of Pediatrics
1979	Department of Biology State University of New York at Buffalo	Visiting Lecturer
1978 - 1979	Undergraduate Health Studies College, State University of New York at Buffalo	Assistant Professor in College H
1974 - 1977	State University of New York at Buffalo	Research Instructor of Pediatrics

Non-Academic

2015 – 2017	Brainstage, Inc. Pittsburgh, PA	Member, Strategic Advisory Board
2014 – 2017	Advanced Technology Healthcare Solutions, Inc. Pittsburgh, PA	Member, Board of Directors
2014 - 2017	Myogenetics, Inc. Buffalo, NY	Member Scientific Advisory Board
2013 - Present	DiaVacs, Inc San Diego, CA	Member, Board of Directors
2012 - 2017	Parabase Genomics, Inc. Boston, MA 02210	Member, Scientific Advisory Board & Board of Directors
2012 - 2016	ConnectCare3 147 W. Airport Road Littiz, PA 17543	Senior Vice President -Research
2010 - Present	BioChem Genetics, LLC 178 Governors Drive	President

	Kiawah Island, SC 29455	
2017 - Present	Baebies, Inc. 615 Davis Drive Durham, NC 27709	Consultant (Half Time)
2017 – Present	EnMed MicroAnalytics, Inc. 178 Governors Drive Kiawah Island, SC 29455	CEO
2018 – Present	Phi Life Sciences, Inc. 645 Meeting Street Charleston, SC 29403	Lab Director
2007	5th European ISNS Regional Meeting Reykjavik, Iceland, June 2007	Co-Chairman, Session on New Screening Methods
2006 - 2010	NGS Research & Development- Europe, GmbH Frankfurt am Main, Germany	President
2006	International Neonatal Screening Symposium – Kumamoto, Japan September 2006	Co-Chairman, Workshop on New Tandem Mass Spectrometry Methods
1994 - 2005	Neo Gen Screening, Inc. 90 Emerson Lane Bridgeville, PA 15017	President & Laboratory Director Founder & Owner (Sold to Pediatrix Medical Group 2003; Resold to Perkin Elmer 2008)
1994 - 1996	Association of State & Territorial Public Health Lab Directors and CORN	Member, Joint Committee on Standardization of Newborn Screening
1993	9th International Neonatal Screening Symposium - Lille, France, September 13-17, 1993	Co-Chairman, Session on Cystic Fibrosis Screening
1993 - 1997	Magee-Womens Hospital Department of Genetics	Director, Pregnancy Screening Laboratory
1991 -1994	Magee-Womens Hospital Department of Genetics	Director, Newborn Screening Program and Laboratory
1991 - 1998	Children's Hospital of Pittsburgh-Department of Genetics	Director, Biochemical Genetics Laboratory
1991 - 1998	Mid-Atlantic Regional Genetic Network	Member, Newborn Screening Committee
1991 - 2002	Pennsylvania Department of Health	Member, Newborn Screening Technical Advisory

		Committee & MSUD Screening Advisory Committee
1991	8th International Neonatal Screening Symposium - Sydney, Australia, November 11-15, 1991	Co-Chairman, Session on Congenital Hypothyroidism Screening
1990 - 1991	8th National Neonatal Screening Symposium - Albany, New York January 29 - February 2, 1991	Member, Organizing Committee
1987 - 1992	Quality Assurance Committee of The Council of Regional Networks For Genetic Services	Member, Subcommittee for Biochemical Genetic Laboratories
1988 - 1989	9th International Symposium of Pteridines and Folic Acid Derivatives - Zurich, Switzerland, September 3-8, 1989	Member International Scientific Committee
1987 - 1988	7th International Neonatal Screening Symposium Sao Paulo, Brazil, November 7-9, 1988	Member, International Organizing Committee Chairman, Workshop on Cystic Fibrosis Screening
1986 - 1991	Department of Medical Genetics Western Pennsylvania Hospital Pittsburgh, PA	Director, Clinical Genetic Laboratories & Newborn Screening Lab
1984 - 1986	6th International Neonatal Screening Symposium - Austin, Texas, November 16-19, 1986	Member, International Organizing Committee, Co-Chairman, Workshop on Cystic Fibrosis Screening
1980 - 1988	West Seneca Developmental Center, State of New York, Department of Mental Hygiene	Consultant in Human Genetics; Member, Human Subjects Review Board
1980 - 1984	New York State Genetics	Member, Executive Council
	Diseases Advisory Committee	Chairman, Subcommittee on Newborn Screening
1980 - 1984	Children's Hospital of Buffalo Buffalo, New York	Allied Medical Specialist Director, Biochemical Genetics Laboratory
1978 - 1986	J. N. Adams Developmental Center State of New York, Department of Mental Hygiene	Consultant in Human Genetics & Research
1976 - 1984	Planned Parenthood of Buffalo	Vasectomy Counselor

1964 - 1966	Universidad del Norte Antofagasta, Chile	U.S. Peace Corps Volunteer Professor, School of Technical Fisheries
1964 - 1966	Escuela Normal del Espiritu Santo Antofagasta, Chile	Instructor in Biology

LANGUAGE SKILLS

Fluent in Spanish.
Reading ability in French and German

NATIONAL AND INTERNALTIONAL AWARDS

2005 Robert Guthrie Award from the American Association of Mental Deficiency
2015 Robert Guthrie Award from the International Society of Neonatal Screening

CERTIFICATION AND LICENSURE

Specialty Certification

3/19/82	American Board of Medical Genetics	Ph.D. - Medical Genetics Clinical Biochemical Genetics
2/26/81	New York State Department of Health Certificate of Qualification as a Laboratory Director	Genetic Testing (Biochemical)

MEMBERSHIPS IN PROFESSIONAL AND SCIENTIFIC SOCIETIES

1971 - 1980	Society of Sigma Xi
1972 - 1985	American Society of Social Biology
1977 - 2004	American Society of Human Genetics
1998 - 2004	American Association of Clinical Chemistry
1999 - 2008	Association for Molecular Pathology
1978 - Present	Society for the Study of Inborn Errors of Metabolism
1978 - Present	Society for Inherited Metabolic Disorders
1988 - Present	International Society for Neonatal Screening

EDITING DUTIES

Journal of Chromatography (Biomedical Applications)

Clinical Chemistry
Clinica Chimica Acta
Analytical Biochemistry
American Journal of Human Genetics
Infant Screening
Journal of Neurochemistry
Screening
Exceptional Parent
American Journal of Public Health
Molecular Genetics and Metabolism

PUBLICATIONS
(Peer Reviewed Journals)

Naylor EW: Genetic screening and genetic counseling: Knowledge, attitudes and practices in two groups of family planning professionals. Soc Biol 1975; 22: 304-314.

Grushka E, Kitka EJ Jr, Naylor EW: Tryptophan and kynurenine determination in untreated urine by reverse-phase high-pressure liquid chromatography. J Chromatogr (Biomed Appl) 1977; 143: 51-56.

Naylor EW, Orfanos AP, Guthrie R: A simple screening test for arginase deficiency (hyperargininemia). J Lab Clin Med 1977; 89: 876-880.

Naylor EW, Gardner EJ: Penetrance and expressivity of the gene responsible for the Gardner Syndrome. Clin Genet 1977; 11: 381-393.

Orfanos AP, Naylor EW, Guthrie R: Micromethod for estimating adenosine deaminase activity in dried blood spots on filter paper. Clin Chem 1978;24:591-5

Naylor EW, Guthrie R: Newborn screening for Maple Syrup Urine Disease (branched-chain ketoaciduria). Pediatrics 1978; 61: 262-266.

Naylor EW, Murphey WH, Domoszlai EI, Guthrie R: Erythropoietic protoporphyria, heterozygous cystinuria, and reduced peptidase A activity in a parent with 46,XX/46,XX,18q- mosaicism. J Med Genet 1978; 15: 157-160.

Naylor EW, Orfanos AP, Guthrie R: An improved screening test for adenosine deaminase deficiency. J Pediatr 1978; 93: 473-476.

Naylor EW, Lebenthal E: Early detection of adenomatous polyposis coli in Gardner's Syndrome. Pediatrics 1979; 63: 222-227.

Evans JE, Tieckelmann H, Naylor EW, Guthrie R: Measurement of urinary pyrimidine bases and nucleosides by high-performance liquid chromatography. J Chromatogr (Biomed Appl) 1979; 163: 29-36.

Naylor EW, Gardner EJ, Richards RC: Desmoid Tumors and mesenteric fibromatosis in Gardner's Syndrome - Report of kindred 109. Arch Surg 1979; 114: 1181-1185.

Paul TD, Naylor EW, Guthrie R: Urine screening for metabolic disease in newborn infants. J Pediatr 1980; 96: 653-656.

Orfanos AP, Naylor EW, Guthrie R: Fluorometric micromethod for determination of arginase activity in dried blood spots on filter paper. Clin Chem 1980; 26: 1198-1200.

Naylor EW, Mosovich LL, Guthrie R, Evans JE, Tieckelmann H: Intermittent non-ketotic dicarboxylic aciduria in two siblings with hypoglycemia: An apparent defect in beta-oxidation of fatty acids. J Inher Metab Dis 1980; 3: 19-24.

Orfanos AP, Naylor EW, Guthrie R: Ultramicromethod for estimation of total glutathione in dried blood spots on filter paper. Anal Biochem 1980; 104: 70-4

Lee GJL, Evans JE, Tieckelmann H, Dulaney JT, Naylor EW: Enzymatic Studies of urinary isomeric chondroitin sulfates from patients with mucopolysaccharidoses. The application of high performance liquid chromatography. Clin Chim Acta 1980; 104: 65-75.

Naylor EW, Lebenthal E: Gardner's Syndrome - Recent developments in research and management. Dig Dis Sci 1980; 25: 945-959.

Garrick MD, Orfanos AP, Rogers L, Naylor EW, Guthrie R: A simple screening test for reduced glutathione in filter paper spots of blood. J Pediatr 1981; 98: 265-267.

Naylor EW: Newborn screening for urea cycle disorders. Pediatrics 1981; 68: 453-457.

Naylor EW, Gardner EJ: Adrenal adenomas in a patient with Gardner's Syndrome. Clin Genet 1981; 20: 67-73.

Naylor EW, Cederdam SD: Urinary pyrimidine excretion in arginase deficiency. J Inher Metab Dis 1981; 4: 207-210.

Orfanos AP, Naylor EW, Guthrie R: Screening test for alpha-1-antitrypsin in dried-blood specimens. Clin Chem 1982; 28: 615-617.

Talbot HW, Jr, Naylor EW, Guthrie R: Neonatal urine screening for metabolic disease with auxotrophic strains of Bacillus subtilis. Clin Chim Acta 1982; 119: 345-349.

Cederbaum SD, Moedjono SJ, Shaw KNF, Carter M, Naylor EW, Walser M: Treatment of hyperargininemia due to arginase deficiency with a chemically defined diet. J Inher Metab Dis 1982; 5: 95-99.

Naylor EW: Newborn screening for urea cycle disorders. Adv Exp Med Biol 1982; 153: 9-18.

Talbot HW, Sumlin AB, Naylor EW, Guthrie R: A neonatal screening test for argininosuccinic acid lyase deficiency and other urea cycle disorders. Pediatrics 1982; 70: 526-531.

Orfanos AP, Naylor EW: A rapid screening test for Duchenne muscular dystrophy using dried blood specimens. Clin Chim Acta 1984; 138: 267-274.

Hoganson G, Berlow S, Kaufman S, Milstien S, Schuett V, Matalon R, Naylor EW, Seifert W: Biopterin synthesis
defects: Problems in diagnosis. Pediatrics 1984; 74: 1004-1011.

Jinks DC, Guthrie R, Naylor EW: Simplified procedure for producing Bacillus subtilis spores for the Guthrie phenylketonuria and other microbiological screening tests. J Clin Microbiol 1985; 21: 826-829.

Szymanski HV, Orfanos A, Narisawa K, Grosz R, Naylor EW: Dihydropteridine reductase in schizophrenic patients. Psychiatry Res 1985; 15: 115-119.

Naylor EW: Recent developments in neonatal screening. Sem Perinatol 1985; 9: 232-249.

Naylor EW, Ennis D, Davidson AGF, Wong LTK, Applegarth DA, Niederwieser A: Guanosine triphosphate cyclohydrolase I deficiency - Early diagnosis by routine urine pteridine screening. Pediatrics 1987; 79: 374-378.

Szymanski HV, Naylor EW, Karoum F: Plasma phenylethylamine and phenylalanine in chronic schizophrenic patients. Biol Psychiatry 1987; 22: 194-198.

Ferre J, Naylor EW: Sepiapterin reductase in cultured human cells. Biochem Biophys Res Comm 1987; 148: 1475-1481.

Ferre J, Naylor EW: Sepiapterin reductase in human amniotic and skin fibroblasts, chorionic villi, and various blood fractions. Clin Chim Acta 1988; 174: 271-282.

Ferre J, Naylor EW, Jacobson KB: Repetitive recycling for guanosine triphosphate cyclohydrolase I for synthesis of dihydroneopterine triphospate. Anal Biochem 1989; 176: 15-18.

Batshaw ML, Naylor EW, Thomas GH: False positive alanine tolerance test results in heterozygote detection of urea cycle disorders. J Pediatr 1989; 115: 595-598.

Baruchin A, Weisberg EP, Miner LL, Ennis D, Nisenbaum LK, Naylor EW, Stricker EM, Zigmond MJ, Kaplan BB: Effects of cold exposure on rat adrenal tyrosine hydroxylase: An analysis of RNA, protein, enzyme activity, and cofactor levels. J Neurochem 1990; 54: 1769-1775.

Hommes FA, Blitzer MG, Brewster MA, Burton BK, Buist NRM, Colombo JP, Elsas LJ, Goldsmith BM, Hammond J, Kruckeberg WC, Leichtman LG, Lin KT, Markin RS, Matalon R, Naylor EW, O'Brien WE, Roe CR, Ruesel A, Shapira E, Taylor H, Tedesco TA, Thompson JN, Tocci PM, Ward JC, Wilson WG: Proficiency testing for biochemical genetics laboratories: The first ten rounds of testing. Amer J Hum Genet 1990; 46: 1001-1004.

Allanson J, Mclnnes R, Bradley L, Tarby T, Naylor EW, Nardella M: Combined transient and peripheral defects in tetrahydrobiopterin synthesis. J Pediatr 1991; 118: 261-263.

Naylor EW: New technologies in newborn screening. Yale J Biol Med 1991; 64: 21-24.

Naylor EW, Hoffman EP, Paulus-Thomas J, Wessel HB, Reid KS, Mitchell B, Schmidt BJ: Neonatal screening for Duchenne muscular dystrophy: Reconsideration based on molecular diagnosis and potential therapeutics. Screening 1992; 1: 99-113.

Spence WC, Paulus-Thomas J, Orenstein DM, Naylor EW: Neonatal screening for cystic fibrosis: Addition of molecular diagnostics to increase specificity. Biochem Med Metab Biol 1993; 49: 200-211.

Citron BA, Kaufman S, Milstien S, Naylor EW, Greene CL, Davis MD: Mutation in the 4a-carbinolamine dehydratase gene leads to mild hyperphenylalaninemia with defective cofactor metabolism. Amer J Hum Genet 1993; 53: 768-774.

Moss G, Naylor EW: Postoperative enteral hyperalimentation results in earlier evaluation of serum branched-chain amino acid levels. Amer J Surg 1994; 168: 33-35.

Magness ST, Tugores A, Christensen SR, Wagner-McPherson C, Evans GA, Naylor EW, Brenner DA: Deletion of the
ferrochetetase gene in a patient with protoporphyria. Hum Mol Genet 1994; 3: 1695-7

Singer-Granick C, Seigel SF, Hill I, Unger A, Finegold DN, Naylor EW: Normal newborn 17-hydroxyprogesterone level in an infant with 11b-hydroxylase deficiency. Screening 1994; 3: 85-89.

Chace DH, Hillman SL, Millington DS, Kahler SG, Roe CR, Naylor EW: Rapid diagnosis of maple syrup urine disease from newborn blood spots by tandem mass spectrometry. Clin Chem 1995; 41: 62-68.

Ziadeh R, Hoffman EP, Finegold DN, Hoop RC, Brackett JC, Strauss AW, Naylor EW. Medium chain acyl-CoA
dehydrogenase deficiency in Pennsylvania: neonatal screening shows high incidence and unexpected mutation
frequencies. Pediatr Res 1995; 37: 675- 678.

Nassif AC, Naylor EW: Immediately elevated postoperative serum branched-chain amino acids (BCAA) following effective GI decompression and enteral feeding. Nutrition 1996; 12: 159-163.

Zimmerman PA, Hercules DM, Naylor EW: Direct analysis of filter paper blood specimens for identification of Smith-Lemli-Opitz syndrome using time-of-flight secondary-ion mass spectrometry. Amer J Med Genet 1997; 68: 300-304.

Chace DH, Hillman SL, Van Hove JL, Naylor EW. Rapid diagnosis of MCAD deficiency: quantitative analysis of
octanoylcarnitine and other acylcarnitines in newborn blood spots by tandem mass spectrometry. Clin Chem
1997; 43: 2106- 2113.

Pastor CM, Williams D, Yoneyama T, Hatekeyama K, Singleton S, Naylor E, Billiar TR: Competition for Tetrahydrobiopterin Between Phenylalanine Hydroxylase and Nitric Oxide Synthase in Rat Livers. J Biol Chem 1996; 271: 24534-24538.

Gibson KM, Bennett MJ, Naylor EW, Morton DH: 3-Methylcrotonyl conenzyme A carboxylase deficiency in Amish/Mennonite adults identified by detection of increased acylcarnitines in blood spots of their children. J
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Rovera GM, Sigurdsson L, Reyes J, Bouch LD, Naylor EW, Todo S, Kocoshis SA: Elevated immunoreactive trypsinogen levels in pediatric patients with short bound syndrome. Transplant Proc 1998; 30: 2549-2550.

Chace DH, Naylor EW: Expansion of newborn screening programs using automated tandem mass spectrometry. MRDD
Res Rev 1999; 5: 150-154.

Chace DH, DiPerna JC, Naylor EW: Laboratory integration and utilization of tandem mass spectrometry in neonatal screening: a model for clinical mass spectrometry in the next millennium. Acta Pediatr Suppl 1999; 88: 45-47.

Dobrowolski SF, Banas RA, Naylor EW, Powdrill T, Thakkar D: DNA microassay technology for neonatal screening. Acta Pediatr Suppl 1999. 88: 61-64.

Heath EM, O'Brien DP, Banas R, Naylor EW, Dobrowolski S: Optimization of an automated DNA purification protocol for neonatal screening. Arch Pathol Lab Med 1999; 123: 1154-1160.

Naylor EW, Chace DH: Automated tandem mass spectrometry for mass newborn screening for disorders in fatty acid, organic acid, and amino acid metabolism. J Child Neurol 1999; 14 (Suppl 1): S4-S8.

Rovera GM, Sigurdsson L, Reyes J, Bouch LD, Naylor EW, Kocoshis SA. Immunoreactive trypsinogen levels in pediatric patients with intestinal failure awaiting intestinal transplantation. Clin Transplant 1999; 13: 395-399.

Wasant P, Naylor EW, Liammongkolkul S. Detection of inherited metabolic disorders via tandem mass spectrometry in Thai infants. Southeastern Asian J Trop Med Public Health 1999; 30 (Suppl 2): 154-159.

Wasant P, Svasti J, Srisomsap C, Liammongkolkul S, Naylor EW, Matsumoto I. Inherited metabolic disorders in Thailand - Siriraj experience. Southeastern Asian J Trop Med Public Health 1999; 30 (Suppl 2): 124-137.

Velazquez A, Vela-Amieva M, Naylor EW, Chace DH. Resultados del tamiz neonatal ampliado, como nueva estrategia para la prevencion de los defectos al nacimiento. Rev Mex Pediatria 2000: 67; 206-213.

Chace DH, DiPerna JC, Mitchell BL, Sgroi B, Hofman LF, Naylor EW. Electrospray tandem massspectrometry for analysis of acylcarnitines in dried postmortem blood specimens collected at autopsy from infants with unexplained cause of death. Clin Chem 2001: 47: 1166-1182

Andresen BS, Dobrowolski SF, O'Reilly L, Munzer J, McCandless SE, Frazier DM, Udvari S, Bross P, Knudsen I, Banas R, Chace DH, Engel P, Naylor EW, Gregersen N. Medium-chain acyl-CoA dehydrogenase (MCAD) mutations identified by MS/MS-based prospective screening of newborns differ from those observed in patients with clinical symptoms: Identification and characterization of a new, prevalent mutation that results in mild MCAD deficiency. Am J Hum Genet 2001: 68: 1408

Chace DH, DiPerna JC, Kalas TA, Johnson RW, Naylor EW: Rapid diagnosis of methylmalonic and propionic acidemias: Quantitative tandem mass spectrometry analysis of propionylcarnitine in filter-paper blood specimens obtained from newborns. Clin Chem 2001: 47: 2040-2044.

Chace DH, Kalas TA, Naylor EW: The application of tandem mass spectrometry to neonatal screening for inherited disorders of intermediate metabolism. Ann Rev Genomics Hum Genet 2002; 3: 17-45.

Wasant P, Matsumoto I, Naylor E, Liammongkolkul S: Mitochondrial fatty acid oxidation disorders in Thai infants: a report of 3 cases. J Med Assoc Thai 2002; 85: Suppl 2: S710-719.

Dobrowolski SF, Banas RA, Suzow JG, Berkley M, Naylor EW: Analysis of common mutations in the galactose-1-phosphate uridyl transferase gene: new assays to increase the sensitivity and specificity of newborn screening for galactosemia. J Mol Diag 2003; 5: 42-47.

Dobrowolski SF, Angeletti J, Banas RA, Naylor EW: Real time PCR assays to detect common mutations in the

biotinidase gene and amplification of mutational analysis to newborn screening for biotinidase deficiency. Mol
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Concentrations: Normative Data by Electrospray Tandem Mass Spectrometry. Pediatr Res 2003; 53: 823-829.

Hofman LF, Foley TP Jr, Henry JJ, Naylor EW: Assays for thyroid-stimulating hormone using dried blood spotted on
filter paper specimens to screen for hypothyroidism in older children and adults. J Med Screen 2003; 10: 5-10.

Chace DH, Kalas TA, Naylor EW: Use of tandem mass spectrometry for multianalyte screening of dried blood
specimens from newborns. Clin Chem 2003; 49: 1797-1817.

Waisbren SE, Albers S, Amato S, Ampola M, Brewster TG, Demmer L, Greenstein R, Korson M, Larson C, Marsden, Msall M, Naylor EW, Pueschel S, Seashore M, Shih VE, Levy HL: Effect of expanded newborn screening for
biochemical genetic disorders on child outcomes and parental stress. JAMA 2003; 290: 2564-2572.

Wasibren SE, Albers S, Amato S, Ampola M, Brewster TG, Demmer L, Eton RB, Greenstein R. Korson M, Larson C, Marsden D, Msall M, Naylor EW, Pueschel S, Seashore M, Shih VE, Levy HL: Effect of expanded newborn screening
for biochemical genetic disorders on child outcome and parental stress. Obstet Gynecol Surv 2004; 59: 415-
417.

Lin Z, Suzow JG, Fontaine JM, Naylor EW: A high throughput beta-globin genotyping method by multiplexed melting
temperature analysis. Mol Genet Metab 2004; 81: 237-243.

Hofman LF, Foley TP, Henry JJ, Naylor EW: The use of filter paper-dried blood spots for thyroid-antibody screening
in adults. J Lab Clin Med 2004; 144: 307-312.

Lin Z, Fontaine JM, Freer DE, Naylor EW: Alternative DNA-based newborn screening for glucose-6-phosphate
dehydrogenase deficiency. Mol Genet Metab 2005; 86: 212-219.

Lin Z, Suzow JG, Fontaine JM, Naylor EW: A simple automated DNA extraction method for dried blood specimens
collected on filter paper. J Assoc Lab Autom 2005; 10: 310-314.

Dobrowolski SF, Ellingson C, Coyne T, Grey J, Martin R, Naylor EW, Koch R, Levy HL. Mutations in the *phenylalanine*
hydroxylase gene identified in 95 patients with phenylketonuria using novel systems of mutation scanning and
specific genotyping based upon thermal melt profiling. Mol Genet Metab. 2007; 91:218-227.

Dobrowolski SF, Pey AL, Koch R, Levy H, Ellingson CC, Naylor EW, Martinez A. Biochemical characterization of

mutant phenylalanine hydroxylase enzymes and correlate to clinical presentation in hyperphenylalaninemic
patients, J Inher Metab Dis 2009;32:10-21.

Dobrowolski SF, Borski K, Ellingson C, Koch R, Levy H, Naylor EW. A limited spectrum of phenylalanine hydroxylase
mutations is observed in phenylketonuria patients in western Poland and implications for treatment with 6R
tetrahydrobiopterin. J Hum Genet 2009; 54: 335-339.

Yusupov R, Finegold DN, Naylor EW, Sahai I, Waisbren S, Levy HL. Sudden death in medium chain acyl-coenzyme
A dehydrogenase deficiency (MCADD) despite newborn screening. Mol Genet Metab 2010; 101: 33-39.

Dobrowolski SF, Pham HT, Downes FP, Prior TW, Naylor EW, Swoboda KJ. Newborn screening for spinal muscular
atrophy by calibrated short-amplicon melt profiling. Clin Chem 2012; 58: 1033-1039.

Foley TP Jr, Henry JJ, Hofman LF, Thomas RD, Sanfilippo JS, Naylor EW. Maternal screening for hypothyroidism
and thyroiditis using filter paper specimens. J Womens Health 2013; 22: 991-996.

Wood MF, Hughes SC, Hache LP, Naylor EW, Abdel-Hamid HZ, Barmada MM, Dobrowolski SF, Stickler DE, Clemens PR. Paternal attitudes toward newborn screening for Duchenne/Becker muscular dystrophy and spinal muscular
atrophy. Muscle Nerve 2014; 49: 822-828.

Bhattacharjee A, Sokolsky T, Wyman SK, Reese MG, Puffenberger, Strauss K, Morton H, Parad RB, Naylor EW.
Development of DNA confirmatory and high-risk diagnostic testing for newborns using targeted next generation
sequencing. Genetics in Medicine 2014; 17: 337-347.

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of newborn screening patients with muscular dystrophy. Muscle Nerve 2015: 53; 570-578.

Naylor EW, Dobrowolski SF, Pai GS, Stickler DE, Abdel-Hamid H, Hoffman EP. Pediatricians' attitudes toward
newborn screening for muscular dystrophy and spinal muscular atrophy. 2018 (In Preparation)

ARTICLES, LETTERS, AND CHAPTERS

Naylor EW, Paul TD, Guthrie R: Comparison of early and late treatment among 90 PKU sibling pairs. Summary of the proceedings of the PKU collaborative study. 1977; 29-35.

Naylor EW: Screening For Maple Syrup Urine Disease. In: Koch R, et al, eds. Maple Syrup Urine Disease Symposium - Issues and Perspectives. Department of Health, Education Education and Welfare Publication No. (HSA) 79-5294 1979: 2-18.

Paul TD, Naylor EW, Guthrie R: Urine screening for metabolic disease in newborn infants. (letter) J Pediatr 1980; 97: 872.

Naylor EW: Newborn screening for Maple Syrup Urine Disease (branched-chain ketoaciduria). In: Bickel H, et al, eds. Neonatal screening for Inborn Errors of Metabolism. Springer-Verlag, Heidelberg, 1980; 213-224.

Naylor EW: New directions in newborn screening. Public Health Laboratory 1980; 38: 264-271.

Naylor EW: Optimal screening and recall procedures. In: Burrow and Dussault, eds. Neonatal Thyroid Screening. Raven Press, New York, 1980; 213-224.

Naylor EW: Future directions in newborn screening. Lab Management 1982; 28: 29-38.

Tocci PM, Cheng MH, Chen RG, Naylor EW, Orfanos AP, Thomas GH: Phenylalanine, fluorometric method, In: Faulkner and Meites, eds. Selected Methods for the Small Clinical Chemistry Laboratory, Vol. 9. 1982; 305-311.

Guthrie R, Naylor EW: EIA and neonatal screening. In: Naruse, Irie, and Tsuji, eds. Neonatal Hypothyroid Screening by Enzyme Immunoassay. Okada Publishing Co, Tokyo, 1982; 29-32.

Naylor EW: Neonatal screening for urea cycle disorders and hyperornithinemia. In: Naruse H and Irie M, eds. Neonatal
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Naylor EW: Future trends in newborn screening. In: Therrel B, ed. Proceedings of a Neonatal Symposium on Laboratory Aspects of Newborn Screening. Texas Department of Health Publications, Austin, 1982; 91-100.

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Ferre J, Naylor EW: Pteridine biosynthesis in human amniocytes and chorionic villi. In: Cooper BA and Whitehead VM, eds. Chemistry and Biology of Pteridines. Walter de Gruyter,New York,1986;309-13

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Hammond K, Naylor EW, Wilcken B: Screening for Cystic Fibrosis. In: Therrel B.L. Jr, ed. Advances In Neonatal Screening. Elsevier, Amsterdam, 1987; 377-382.

Unger R, Naylor EW: Parental attitudes towards neonatal screening for Cystic Fibrosis In: Therrel B.L. Jr. ed. Advances In Neonatal Screening. Elsevier, Amsterdam, 1987; 383-384.

Ferre J, Naylor EW: Sepiapterin reductase in human amniocytes, skin fibroblasts, chorionic villi and stimulated mononuclear blood cells. In: Curtius HC and Blau N, eds. Unconjugated Pterins and Related Biogenic Amines. Walter de Gruytor, Berlin, 1987; 247-256.

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Latin America. Latin American Society for Newborn Screening meeting Santiago, Chile November 2015.

PROFESSIONAL ACTIVITIES

Research Grants Received	Years Inclusive	Grant Title and Number	Source
$ 199,638	06/01/13 – 11/30/13	Development and Validation of a High Risk and Potential Newborn Screening for Inherited Metabolic Disorders and Rare Genetic Syndromes Using Focused Next Generation Sequencing (Parabase Genomics)	NIH-NICHD-SBIR Phase I #R43-HD76544-01
$ 200,527	01/01/13 – 06/30/13	Confirmatory Screening for Congenital Non-syndromic Hearing Loss Using Targeted Next Generation Sequencing (Parabase Genomics)	NIH-NIDCD-SBIR Phase I #R43-DC13012-01
$ 782,108	09/30/10 - 09/29/13	Newborn Screening for Spinal Muscular Atrophy and Duchenne/ Becker Muscular Dystrophy.	NIH-NICHD-CDBPM 10-15-PS (Sub-Contract)
$ 97,856	09/30/01 – 02/31/02	Comprehensive CFTR Genotyping by Peptide Mass Signature Genotyping	NIH – SBIR – Phase I #R43 – HL66833 - 01
$1,362,909	04/01/01 – 03/31/04	Newborn Screening for Hearing Impairment	NIH – SBIR – Phase II #R44 – DC04902 -02
$ 99,172	07/01/01 – 12/31/01	Newborn Screening for Hearing Impairment	NIH – SBIR – Phase I #R44 – DC04902 – 01
$1,316,226	04/01/01 – 03/31/03	Newborn Screening by Multiplex Molecular Analysis	NIH – SBIR – Phase II #R44 – HD37757-02
$1,347,000	09/15/00 – 03/15/03	Comprehensive CYP21 Genotyping	NIH – SBIR – Phase I/II # R44 - DK 58522-01
$ 86,059	03/01/99 - 08/31/99	Newborn screening by multiplex molecular analysis	NIH - SBIR - Phase I # R43 - HD 37757 - 01

$ 734,454	01/01/99 - 12/31/00	Simplified population screening for adult hypothyroidism.	NIH - SBIR - Phase II # R44 - DK 53666 - 02
$ 750,000	03/23/98 - 03/22/00	A simple method for population screening for IDDM genetic risk using a unique fluorescent assay for HLA genotyping.	NIH - SBIR Contract Phase II # N44 - DK8 - 2272
$ 84,670	10/01/97 - 03/31/98	Simplified population screening for adult hypothyroidism.	NIH - SBIR - Phase I # R3DK5366A
$ 90,038	09/96 - 03/97	Simple Method for Screening for IDDM Genetic Risk Using a unique Fluorescent Assay for HLA Genotyping	NIH - SBIR Contract #N43-DK-6-2221
$ 27,500	07/96 - 12/96	Development of Free Carnitine Assay on Filter Paper Blood Spots using Tandem Mass Spectrometry.	Sigma Tau Pharmaceuticals
$ 284,392	07/92 - 12/95	Newborn Screening for Duchenne and Becker	Muscular Dystrophy Association
$ 56,975	05/91 - 03/93	Newborn Screening/ Education Program for Maple Syrup Urine Disease	Pennsylvania Dept. of Health (Contract)
$ 72,000	11/89 - 10/90	Pilot Study of Intimal Hyperplasia in Renal Access Grafts	West Penn Hospital Foundation
$ 25,725	11/88 - 10/89	Intimal Hyperplasia in Renal Biosynthesis	West Penn Hospital Foundation #8012
$101,450	11/87 - 10/89	Inherited Disorder of Tetrahydrobiopterin Biosynthesis	West Penn Hospital Foundation
$ 339,304	07/88 - 06/93	National Screening Center for PKU and Molybdenum Cofactor Variants	Dept. of Health and Human Services Maternal Child Health #MCJ-9086
$1,000	1987	Travel Grant to attend 4th	Society for Inherited

		International Congress of Inborn Errors of Metabolism in Sendai Japan, May 26-30, 1987	Metabolic Disorders
$176,390	07/84 - 06/88	National Screening Center for PKU Cofactor Variants	Department of Health & Human Services Maternal Child Health #MCJ-9049
$146,350	07/84 - 06/86	PKU Cofactor Variant and Biopterin Biosynthesis Research Program	Magee-Womens Hospital Board of Trustees #2611.712
$ 14,340	04/84 - 09/84	Regional GC-MS and Newborn Screening Confirmation Program HRI Contract	New York State Genetic Diseases Testing & Counseling Project #33067-10/13
$ 14,110	04/83 - 09/83	" "	" "
$ 12,700	01/83 - 03/84	“ “	“ “
$ 9,645	10/82 - 03/83	" "	" "
$ 17,535	01/81- 09/82	" "	" "
$ 32,585	07/81- 09/82	Screening Program for Cystic Fibrosis	Cystic Fibrosis Foundation (CFF-G037-2-01)
$ 2,085	06/80-03/81	A Biochemical Study of Inherited Metabolic Defects Beta-Oxidation of Fatty Acid	Biomedical Research Support Grant in the Children’s Hospital Of Buffalo BRS #2835