

# Curriculum Vitae

## Timothy Carl Wood

### Professional Contact Information

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### Professional Appointments

2005	<b>Director</b> Biochemical Genetics Laboratory	Greenwood Genetic Center Greenwood, SC
2004	<b>Assistant Director</b> Biochemical Genetics Laboratory	Greenwood Genetic Center Greenwood, SC

### Postgraduate Training

2000 - 2004	Clinical Molecular / Biochemical Fellow Advisors: John Longshore, Ph.D. Harold Taylor, Ph.D.	Greenwood Genetic Center Greenwood, SC
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### Education

2005	<b>Board Certified in Clinical Biochemical Genetics</b> American Board of Medical Genetics	
2002	<b>Board Certified in Clinical Molecular Genetics</b> American Board of Medical Genetics (Recertified – 2012)	
1995 – 2000	<b>Ph.D. in Human Genetics</b> Advisor: Jerry Thompson, Ph.D. Title: “ <i>Mutation analysis of the heparin sulfamidase and N-acetyl-<math>\alpha</math>-D-glucosaminidase genes in Sanfilippo syndrome types A and B, respectively</i> ”	Univ. of Alabama at Birmingham Birmingham, AL
1993 – 1995	<b>M.S. in Zoology</b> Advisor: Carey Krajewski, Ph.D. Title: <i>Phylogenetic relationships among the Sarus species group (Grus Antigone)</i>	Southern Ill. Univ. at Carbondale Carbondale, IL
1989 – 1993	<b>B.S. in Biology with honors</b>	Samford Univ. Birmingham, AL

## Professional Membership

American College of Medical Genetics - Fellow  
American Society of Human Genetics  
Society of Inherited Metabolic Disease  
Society for the Study of Inherited Metabolic Disease  
Southeast Regional Genetics Group

## Professional Development

2013- present      CAP/ACMG Joint Committee  
2008 – 2013      ACMG Quality Control Committee  
2008- present      Southeast Regional Genetics and NBS Collaborative (SERC) – Steering  
                                 Committee  
2008                  American Board of Medical Genetics Item Writer  
2006 – present      Southeast Regional Genetics Group (SERGG) Board of Directors  
                                 SERGG President Elect (2012-2014)  
                                 Organized SERGG annual meeting in Charleston SC (2008)  
2004- present      Member South Carolina Newborn Screening Advisory Committee

## Invited Lectures / Teaching Experience

2014                  Annual Human genetics retreat University of Pittsburgh – “Next steps in  
                                 biochemical genetic testing”  
2013                  Grand Rounds University of Calgary, Alberta Canada “Laboratory testing  
                                 for MPS disorders: current approaches and future directions”  
2013                  ACMG Industry Supported Symposium “Laboratory Diagnosis of  
                                 Morquio Syndrome Type A”  
2012                  USC Medical School-Greenville Module 1 Instructor  
2010, 2012          MPS Laboratory Training Preceptorship  
2010                  SERC Lunch and Learn Series “Newborn Screening for VLCAD and GAI  
                                 in South Carolina”  
2001- present      Greenwood Genetic Center Graduate Course (Biology 614)  
2002 -present      USC Genetic Counseling/GGC Laboratory Experience  
2001                  Lander University Fine Arts Lecture Series - “Advances in Genetics”  
1995-2000          Jefferson State Community College – Introductory Biology Instructor

## Articles

Ellsworth KA, Pollard LM, Cathey S, **Wood T**. Measurement of Elevated Concentrations of Urine Keratan Sulfate by UPLC-MSMS in Lysosomal Storage Disorders (LSDs): Comparison of Urine Keratan Sulfate Levels in MPS IVA Versus Other LSDs. *JIMD Rep*. 2016 Jul 28.

Ng BG, Shiryaev SA, Rymen D, Eklund EA, Raymond K, Kircher M, Abdenur JE, Alehan F, Midro AT, Bamshad MJ, Barone R, Berry GT, Brumbaugh JE, Buckingham KJ, Clarkson

K, Cole FS, O'Connor S, Cooper GM, Van Coster R, Demmer LA, Diogo L, Fay AJ, Ficicioglu C, Fiumara A, Gahl WA, Ganetzky R, Goel H, Harshman LA, He M, Jaeken J, James PM, Katz D, Keldermans L, Kibaek M, Kornberg AJ, Lachlan K, Lam C, Yapliito-Lee J, Nickerson DA, Peters HL, Race V, Régal L, Rush JS, Rutledge SL, Shendure J, Souche E, Sparks SE, Trapane P, Sanchez-Valle A, Vilain E, Vøllø A, Waechter CJ, Wang RY, Wolfe LA, Wong DA, **Wood T**, Yang AC; University of Washington Center for Mendelian Genomics, Matthijs G, Freeze HH. ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. *Hum Mutat.* 2016 Jul; 37(7):653-60.

Champaigne NL, Leroy JG, Kishnani PS, Decaestecker J, Steenkiste E, Chaubey A, Li J, Verslype C, Van Dorpe J, Pollard L, Goldstein JL, Libbrecht L, Basehore M, Chen N, Hu H, **Wood T**, Friez MJ, Huizing M, Stevenson RE. New observation of sialuria prompts detection of liver tumor in previously reported patient. *Mol Genet Metab.* 2016 Jun; 118(2):92-9. doi: 10.1016/j.ymgme.2016.04.004. Epub 2016 Apr 16.

Zhang W, James PM, Ng BG, Li X, Xia B, Rong J, Asif G, Raymond K, Jones MA, Hegde M, Ju T, Cummings RD, Clarkson K, **Wood T**, Boerkoel CF, Freeze HH, He M. A Novel N-Tetrasaccharide in Patients with Congenital Disorders of Glycosylation, Including Asparagine-Linked Glycosylation Protein 1, Phosphomannomutase 2, and Mannose Phosphate Isomerase Deficiencies. *Clin Chem.* 2016 Jan; 62(1):208-17.

Ng BG, Raymond K, Kircher M, Buckingham KJ, **Wood T**, Shendure J, Nickerson DA, Bamshad MJ; University of Washington Center for Mendelian Genomics, Wong JT, Monteiro FP, Graham BH, Jackson S, Sparkes R, Scheuerle AE, Cathey S, Kok F, Gibson JB, Freeze HH. Expanding the Molecular and Clinical Phenotype of SSR4-CDG. 2015. *Hum Mutat.* Nov;36 (11):1048-51

Albert JS, Bhattacharyya N, Wolfe LA, Bone WP, Maduro V, Accardi J, Adams DR, Schwartz CE, Norris J, **Wood T**, Gafni RI, Collins MT, Tosi LL, Markello TC, Gahl WA, Boerkoel CF. Impaired osteoblast and osteoclast function characterize the osteoporosis of Snyder - Robinson syndrome. 2015. *Orphanet J Rare Dis.* Mar 7; 10:27.

Zhang H, **Wood T**, Young SP, Millington DS. A straightforward, quantitative ultra-performance liquid chromatography-tandem mass spectrometric method for heparan sulfate, dermatan sulfate and chondroitin sulfate in urine: An improved clinical screening test for the mucopolysaccharidoses. 2015. *Mol Genet Metab.* Feb; 114(2):123-8.

Bodamer OA, Giugliani R, **Wood T**. The laboratory diagnosis of mucopolysaccharidosis III (Sanfilippo syndrome): A changing landscape. 2014. *Mol Genet Metab.* Sep-Oct; 113(1-2):34-41.

Leroy JG, Sillence D, **Wood T**, Barnes J, Lebel RR, Friez MJ, Stevenson RE, Steet R, Cathey SS. A novel intermediate mucopolipidosis II/III $\alpha\beta$  caused by GNPTAB mutation in the cytosolic N-terminal domain. 2014. *Eur J Hum Genet.* 22: 594-601.

Peron A, Spaccini L, Norris J, Bova SM, Selicorni A, Weber G, **Wood T**, Schwartz

CE, Mastrangelo M. Snyder-Robinson syndrome: a novel nonsense mutation in spermine synthase and expansion of the phenotype. *Am J Med Genet A*. 2013 Sep; 161(9):2316-20.

Lieu MT, Ng B, Rush J, **Wood T**, Basehore M, Hedge M, Chang R, Abdenur J, Freeze H, Wang R. Severe, fatal multisystem manifestations in a patient with dolichol kinase-congenital disorder of Glycosylation. 2013. *Molecular Genetics and Metabolism*. 110(4):484-9.

Zhang Z, Norris J, Kalscheuer V, **Wood T**, Wang L, Schwartz C, Alexov E, and Van Esch H. A Y328C missense mutation in spermine synthase causes a mild form of Snyder-Robinson syndrome. 2013. *Hum Mol Genet*. 1-9.

Marshall LS, Simon J, **Wood T**, Peng M, Owen R, Feldman GS, Zaragoza MV. Deletion Xq27.3q28 in female patient with global developmental delays and skewed X-inactivation. 2013. *BMC Medical Genetics*, 14(1):49.

Hendriksz CJ, Harmatz P, Beck M, **Wood T**, Lachman R, Gravance C, Orii T, Tomatsu S. Review of clinical presentation and diagnosis of mucopolysaccharidosis IVA. 2013. *Mol Genet Met*. April 10.

Mendelsohn N, **Wood T**, Olson R, Temme R, Hale S, Zhang H, Read L, White K. Spondyloepiphyseal dysplasias and bilateral Legg-Calve-Perthes Disease: Diagnostic considerations for Mucopolysaccharidoses. 2013. *JIMD Reports*. May 9.

**Wood TC**, Bainbridge K, Beck M, Graeff Burin M, Chien Y, Church H, D'Almeida V, van Diggelen O, Fietz M, Giugliani R, Harmatz P, Hawley S, Hwu W, Ketteridge D, Lukacs Z, Miller N, Pasquali M, Schenone A, Thompson J, Tylee K, Yu C, Hendriksz C. Diagnosing MPSIVA. *JIMD*. 2013 Epub:Feb 1.

Pollard LM, Jones JR, **Wood TC**. Molecular characterization of 355 mucopolysaccharidosis patients reveals 104 novel mutations. *JIMD*. 2012 Epub:Sep 14.

**Wood TC**, Bodamer OA, Burin MG, D'Almeida V, Fietz M, Giugliani R, Hawley S, Hendriksz CJ, Hwu WL, Ketteridge D, Lukacs Z, Mendelsohn NJ, Miller N, Pasquali M, Schenone A, Schoonderwoerd K, Winchester B, Harmatz P. MPS VI: Illuminating the Path to Diagnosis. *Mol Genet Met*. 2012 106(1): 73-82.

De Biase I, Champaigne N, Schroer R, Pollard L, Longo N, **Wood TC**. Primary carnitine deficiency presents atypically with long QT syndrome: a case report. *JIMD reports*. 2012 2: 87-90.

Mudd H, Wagner C, Luka Z, Stabler S, Allen R, Schroer R, **Wood TC**, Wang J, Wong L-J. Two patients with hepatic mtDNA depletion syndromes and marked elevations of S-adenosylmethionine and methionine. *Mol Gene Met*. 2012 105(2): 228-36.

Burruss D, **Wood TC**, Espinoza L, Dwivedi A, Holden K, Severe Hunter syndrome (Mucopolysaccharidosis II) phenotype secondary to large deletion in the X chromosome encompassing IDS, FMR1 and AFF2 (FMR2). *J Child Neurol.* 2012 27(6): 776-780.

Sowell J, Fuqua M, **Wood TC**. Quantification of total and free carnitine in human plasma by hydrophilic interaction liquid chromatography tandem mass spectrometry. *J Chromatogr Sci.* 2011 49(6): 463-8.

Sowell J, Pollard L, **Wood TC**. Quantification of branched-chain amino acids in blood spots and plasma by liquid chromatography tandem mass spectrometry for the diagnosis of maple syrup urine disease. 2011 *J. Sep. Sci.* 34, 1-9.

Sowell J, Norris J, Jones K, Schwartz C, **Wood TC**. Diagnostic screening for spermine synthase deficiency by liquid chromatography tandem mass spectrometry. *Clinica Chimica Acta.* 2011 412(7-8): 655-60.

Sowell J, **Wood TC**. Towards a selected reaction monitoring mass spectrometry fingerprint approach for the screening of oligosaccharidoses. *Analytica Chimica Acta.* 2011 686: 102-106.

Champion KJ, Basehore MJ, **Wood TC**, Destrée A, Vannuffel P, Maystadt I. Identification and characterization of a novel homozygous deletion in the alpha-N-acetylglucosaminidase gene in a patient with Sanfilippo type B syndrome (mucopolysaccharidosis IIIB). *Mol Genet Met.* 2010 100(1): 51-6.

Hathaway SC, Friez M, Limbo K, Parker C, Salomons GS, Vockley J, **Wood TC**, Abdul-Rahman OA. X-linked creatine transporter deficiency presenting as a mitochondrial disorder. *J Child Neurol.* 2010 25(8): 1009-12.

Cathey SS, Leroy JG, **Wood TC**, Eaves K, Simensen RJ, Kudo M, Stevenson RE, Friez MJ, Phenotype and genotype in mucopolipidoses II and III alpha/beta: a study of 61 probands. *J Med Genet.* 2010 47(1): 38-48.

Pollard LM, Williams NR, Espinoza L, **Wood TC**, Spector EB, Schroer RJ, Holden KR. Diagnosis, Treatment, and Long-Term Outcomes of Late-Onset (Type III) Multiple Acyl-CoA Dehydrogenase Deficiency. *J Child Neurol.* 2010 25(8): 954-60.

Carling RS, Hogg SL, **Wood TC**, Calvin J. Simultaneous determination of guanidinoacetate, creatine and creatinine in urine and plasma by un-derivatized liquid chromatography-tandem mass spectrometry. *Ann Clin Biochem.* 2008 45(Pt 6): 575-84.

Giurgiutiu DV, Espinoza LM, **Wood TC**, Dupont BR, Holden KR. Persistent Growth Failure in Prader-Willi Syndrome Associated With Short-Chain Acyl-CoA Dehydrogenase Gene Variant. *J Child Neurol.* 2008 23(1): 112-7.

Wang D, **Wood TC**, Sadilek M, Scott CR, Turecek F, Gelb MH. Tandem mass spectrometry for the direct assay of enzymes in dried blood spots: application to newborn screening for mucopolysaccharidosis II (Hunter disease). 2007 *Clin Chem*. 53(1): 137-40.

Lyons MJ, **Wood TC**, Espinoza L, Stensland HM, Holden KR. Early onset alpha-mannosidosis with slow progression in three Hispanic males. *Dev Med Child Neurol*. 2007 49(11): 854-7.

Schimmenti LA, Crombez EA, Schwahn BC, Heese BA, **Wood TC**, Schroer RJ, Bentler K, Cederbaum S, Sarafoglou K, McCann M, Rinaldo P, Matern D, Amat di San Filippo C, Pasquali M, Berry SA, Longo N. Expanded newborn screening identifies maternal primary carnitine deficiency. *Mol Genet Met*. 2007 90(4): 441-445.

Clark AJ, Rosenburg EH, Almeida LS, **Wood TC**, Jakobs C, Stevenson RE, Schwartz CE, Salomons GS. X-linked creatine transporter (SLC6A8) mutations in about 1% of males with mental retardation of unknown etiology. *Hum Genet*. 2006 119(6): 604-10.

Cason AL, Ikeguchi Y, Skinner C, **Wood TC**, Holder KR, Lubs HA, Martinez F, Simensen RJ, Stevenson RE, Pegg AE, Schwartz CE: X-linked spermine synthase gene (SMS) defect: the first polyamine deficiency syndrome. *Eur J Hum Genet*. 2003 11(12): 937-944,

Li P, **Wood TC**, Thompson JN. Diversity of mutations and distribution of single nucleotide polymorphic alleles in the human  $\alpha$ -L-iduronidase (IDUA) gene. *Gene Med*. 2002 4(6): 420-426.

Hahn KA, Salomons GS, Tackels-Horne D, **Wood TC**, Taylor HA, Schroer RJ, Lubs HA, Jakobs C, Olson RL, Holden KR, Stevenson RE, Schwartz CE. X-linked mental retardation with seizures and carrier manifestations is caused by a mutation in the creatine-transporter gene (SLC6A8) located in Xq28. *Am J Hum Genet*. 2002 70(5): 1349-56.

**Wood TC**, Krajewski C, Mitochondrial DNA sequence variation among the subspecies of Sarus Crane (*Grus Antigone*). *The Auk*. 1996 113(3): 655-663.

Krajewski C, **Wood TC**. Phylogenetics relationships within the Sarus Crane species group (Gruiformes: Gruidae) based on mitochondrial DNA sequences. *Emu*. 1995 95: 99-105.

## Book Chapters

Disorders of creatine metabolism (2007) Young, S.P., Struys, E., **Wood, TC**, in *Current Protocols in Human Genetics*. 17.3 Supplement 54.