CURRICULUM VITAE
Steven A. Skinner, M.D.

BUSINESS ADDRESS: Greenwood Genetic Center
106 Gregor Mendel Circle
Greenwood, SC 29646

EDUCATION

September 1973 - August 1975	B.S., Wofford College, Spartanburg, SC
August 1975 - May 1979	M.D., Medical University of South Carolina Charleston, SC

PROFESSIONAL TRAINING

July 1979 – April 1981	Internship – Family Practice Richland Memorial Hospital Columbia, SC
May 1981 – June 1983	Residency – Pediatrics Richland Memorial Hospital Columbia, SC
August 1987 – July 1989	Fellowship in Clinical Genetics Greenwood Genetic Center Greenwood, SC

PROFESSIONAL ACTIVITIES

July 1982 – June 1983	Chief Resident in Pediatrics Richland Memorial Hospital Columbia, SC
July 1984 – August 1987	Director of Newborn Nursery Georgetown Memorial Hospital Georgetown, SC
January 1990 – December 1990	Chairman of Department of Pediatrics Self Memorial Hospital Greenwood, SC
July 2000 – June 2005	C.A.T.C.H. S.C. Co-facilitator

July 2002 – October 2004	Medical Consultant S.C. D.H.E.C. Children With Special Health Care Needs
November 2002 – Present	S.C. EHDI (Early Hearing Detection Intervention) Chapter Champion
January 2009-Present	Foundation for Angelman Syndrome Therapeutics Scientific Advisory Board
September 2012-Present	USC Genetic Counseling Program Advisory Council

FACULTY APPOINTMENTS

December 2012 – present	Adjunct Full Professor Department of Biological Sciences Clemson University
July 2013 – June 2016	Clinical Professor of Pediatrics Department of Pediatrics University of South Carolina School of Medicine

HOSPITAL APPOINTMENTS

1987– present	Medical Staff Self Regional Healthcare
August 2006 – present	Medical Staff Shriners Hospital for Children
March 2016 - present	Medical Staff Greenville Health System

PROFESSIONAL POSITIONS

October 1983 – August 1987	Private Practice of Pediatrics and Adolescent Medicine Georgetown, SC
August 1987 – January 1997	Private Practice of Pediatrics Greenwood Children's Clinic Greenwood, SC
August 1989 – January 1997	Clinical Geneticist Greenwood Genetic Center Greenwood, SC
January 1997 – October 2000	Director Greenwood Community Children's Center Greenwood, SC
November 2000 – January 2011	Senior Clinical Geneticist Director of S.C. DDSN Genetic Services Greenwood Genetic Center Greenwood, SC
November 2003 – Present	Director of Treatment Services Greenwood Genetic Center Greenwood, SC

January 2005 – October 2012	Director of Clinical Services Greenwood Genetic Center Greenwood, SC
February 2006 – January 2011	Associate Director Greenwood Genetic Center Greenwood, SC
January 2011-Present	Director Greenwood Genetic Center Greenwood, SC
BOARD STATUS	Board Certified – American Board of Pediatrics, November 1985 Board Certified – American Board of Medical Genetics – August 1990
MEMBERSHIPS	Fellow of the American Academy of Pediatrics South Carolina Medical Association Greenwood County Medical Society The South Carolina Pediatric Society The American Society of Human Genetics American College of Medical Genetics, Founding Fellow
COMMUNITY	S.C. DHEC Commissioner's Pediatric Advisory Committee S.C. First Sound Advisory Council

RESEARCH ACTIVITIES

Current Research Support:

NIH/Rare Diseases Clinical Research Network: Rett Syndrome, MECP2 Duplications, and Rett-related Disorders Natural History (2U54HD061222-11)
September 2014 – ongoing
Role: Principal Investigator

A Randomized, Double-blind, Placebo-controlled Dose-ranging Study of the Safety and Pharmacokinetics of Oral NNZ-2566 in Pediatric Rett Syndrome (Neu-2566-RETT-002, Neuren Pharmaceuticals Ltd.)
March 2016-ongoing
Role: Principal Investigator

Completed Research Support:

Efficacy of a Therapeutic Treatment Trial in Angelman Syndrome (RDN 5204)
September 2006-August 2007.
Therapeutic treatment trial for Angelman syndrome.
Role: Principal Investigator

South Carolina Autism Project: A Search for Causes (95-219)
February 1995-January 1998
Clinical and genetic laboratory evaluation of 200 patients with Autism.
Role: Co-Investigator

NIH/Rare Diseases Clinical Research Network: Angelman Syndrome Natural History (RDN 5203)
September 2003-July 2008; Renewed: August 2009-July 2014
Natural history study of Angelman Syndrome.
Role: Principal Investigator

NIH/Rare Diseases Clinical Research Network (NIH RR019478)
Sleep Abnormalities in Rare Genetic Disorders: Angelman Syndrome, Rett Syndrome, and Prader-Willi Syndrome (RDN 5207)
August 2009-July 2014
Role: Principal Investigator

A Phase II Randomized Placebo-controlled Trial of Levodopa in Angelman Syndrome (R01FD003523)
July 2010-June 2014
Therapeutic treatment trial for Angelman syndrome
Role: Principal Investigator

A Randomized, Double-blind, Placebo-controlled, Parallel Group Study to Evaluate AFQ056 in Adult Patients with Fragile X Syndrome. (Clinical Trial Protocol CAFQ056A2212, Novartis Pharmaceutical Corporation)
August 2011-December 2015
A therapeutic trial for Fragile X Syndrome.
Role: Principal Investigator

Novartis Pharmaceuticals: An open-label study to evaluate the long-term safety, tolerability and efficacy of AFQ056 in adult patients with Fragile X Syndrome (CAFQ056B2279)
August 2011-December 2015
Role: Principal Investigator

Novartis Pharmaceuticals: A randomized, double-blind, placebo-controlled, parallel group study to evaluate the efficacy and safety of AFQ056 in adolescent patients with Fragile X Syndrome (CAFQ056B2214)-active
May 2011-December 2013
Role: Principal Investigator

Whole Blood Acquisition and Analysis for the Determination of the Degree of Methylation of the FMR1 Gene in Support of Developing a Diagnostic Test in Fragile X Syndrome (Novartis Pharmaceutical Corporation) –
June 2011-June 2014
Assist with acquisition of blood samples from patients with Fragile X syndrome to support development of a companion diagnostic test in Fragile X syndrome
Role: Co-Principal Investigator

Reduced NADH Production in the Presence of Tryptophan as a Biomarker of Autism Spectrum Disorders
NIH/NICHD (1R21HD072473-01)
April 2012-March 2014
Role: Clinical Geneticist

NIH/Rare Diseases Clinical Research Network: Rett Syndrome Natural History (RDN 5201)
September 2003-July 2008; Renewed: August 2009-July 2014
Natural history study of Rett Syndrome.
Role: Principal Investigator

NIH/Rare Disease Clinical Research Network (NIH RR019478)
September 2003-July 2008; Renewed: August 2009-July 2015
Title of Project: Angelman, Rett & Prader-Willi Syndromes Consortium
This study will perform natural history studies as well as develop meaningful therapies.
Role: Co-Investigator

ABSTRACTS

1. Phelan MC, Skinner SA, Schroer RJ, and Stevenson RE: Radial aplasia in trisomy 18 and triploidy. 1990 Southern Genetics Group Summer Meeting, July 19-21, 1990, Destin, Florida.

2. Skinner SA, Blackburn WR, and Stevenson RE: Hazards of Breech Birth – 14 year Follow-Up. 1995 David W. Smith Workshop, July 30 – August 30, 1995, Big Sky, Montana.

3. Skinner SA, Wilcox WR, and Spranger J: Geleophysic dysplasia – A lysosomal storage disorder with distinctive facies. 1996 David W. Smith Workshop, September 26 – October 1, 1996, Lake Arrowhead, California.

4. Skinner SA, Collins JS, Wood TC, Seaver LH, and Stevenson RE: Neural Tube Defects and Holoprosencephaly: Further Evidence of a Nonrandom Association. 2002 David W. Smith Workshop, August 7-11, 2002, Greenville, S.C.

5. S.U. Peters, L.M. Bird, R. Barbieri-Welge, W.H. Tan, R.J. Hundley, S. Skinner, A. Bauer-Carlin, T. Sahoo and C.A. Bacino. The relationship between molecular subtype and autism symptom severity in Angelman Syndrome. 2008 International Meeting for Autism Research, May 15 – 17, 2008, London.

6. Jennifer K. Gentile, Wen-Hann Tan, Carlos A. Bacino, Steven A. Skinner, Rene Barbieri-Welge Astrid Baeur-Carlin, Arthur L. Beaudet, Terry Jo Bichell, Lucia T. Horowitz, Hye-Seung Lee, Trilochan Sahoo, Susan E. Waisbren, Lynne M. Bird, Sarika U. Peters: A neurodevelopmental survey of Angelman syndrome with genotype-phenotype correlations. ACMG Meeting, March 2009

7. W.H. Tan, C.A. Bacino, S.A. Skinner, I. Anselm, R. Barbieri-Welge, A. Baeur-Carlin, A.L. Beaudet, T.J. Bichell, J.K. Gentile, D.G. Glaze, L.T. Horowitz, H.S. Lee, M.P. Nespeca, S.U. Peters, T. Sahoo, D. Sarco, S.E. Waisbren, L.M. Bird: Clinical features in 102 patients with Angelman syndrome. ACMG Meeting, March 2009

8. M. Shinawi, T. Sahoo, P.B. Santos-Celestino, R. Zascavage, J.R. German, A. Porter, P. Fang, D. Treadwell-Deering, C. Skinner, S.A. Skinner, R.E. Stevenson and A.L. Beaudet: Exon-Focused Microarray Analysis of Candidate Genes in Autism. 2009 International Meeting for Autism Research, May 7 – 9, 2009, Chicago, Illinois.

PUBLICATIONS

1. Saul RA, Stevenson RE, Rogers RC, **Skinner SA**, Prouty LA, and Flannery DB: Growth References from Conception to Adulthood, Jacobs Press, 1988.

2. **Skinner SA**, Stevenson RE, Flannery DB: Catel-Manzke Syndrome, Proc Greenwood Genetic Center 8:60-63, 1989.

3. **Skinner SA** and Stevenson RE: Obstruction as a cause of failure of Testicular Descent, Proc Greenwood Genetic Center 8:28-30, 1989.

4. Macpherson RI, **Skinner SA**, Donnenfeld AE: Acampomelic Campomelic dysplasia, Pediatric Radiology 20:90-93, 1989.

5. Stevenson RE, Phelan MC, and **Skinner SA**: Pregnancy and triple X, Proc Greenwood Genetic Center 8:44-49, 1989.

6. Allen WP, Stevenson RE, Saul RA, and **Skinner SA**: Macrocephaly-mesodermal hamartoma spectrum, Proc Greenwood Genetic Center 9:16-18, 1990.

7. **Skinner SA**: Osteodysplastic primordial dwarfism, Proc Greenwood Genetic Center 10:31-33, 1991.

8. Phelan MC, **Skinner SA**, Cooley L, and Richter B: Familial translocation (8; 9) resulting in trisomy 8q24.1®qter in three generations, Proc Greenwood Genetic Center 11:22-25, 1992.

9. Taylor HA, **Skinner SA**: Beta Galactosidase Deficiency, Proc Greenwood Genetic Center 13:20-22, 1994.

10. **Skinner SA**: Neurenteric Malformations, Proc Greenwood Genetic Center 13:20-22, 1994.

11. **Skinner SA**, Cooley L, Phelan MC, et al: Fetal Autopsies: A ten year experience, Proc Greenwood Genetic Center 13:26-28, 1994.

12. Phelan MC, Saul RA, Gailey Jr TA, **Skinner SA**: Prenatal diagnosis of mosaic 4p- in a fetus with trisomy 21, Prenat Diagn. 15:274-277, 1995.

13. Michaelis RC, **Skinner SA**, Lethco BA, Simensen RJ, Donlon TA, Tarleton J, Phelan MC: Deletion Involving D15S113 in a Mother and Son Without Angelman Syndrome: Refinement of the Angelman Syndrome Critical Deletion Region. Am J Med Genet. 55:120-126, 1995.

14. **Skinner SA**: Mental Retardation in South Carolina VI. Recognizable Syndromes of Presumed Genetic Cause, Proc Greenwood Genetic Center 15:71-75, 1996.

15. Michaelis RC, **Skinner SA**, Deason R, Skinner C, Moore CL, Phelan MC: Intersitial deletion of 20p: new candidate region for Hirschsprung disease and autism?, Am J Med Genet. 71:298-304, 1997.

16. Schroer RJ, Phelan MC, Michaelis RC, Crawford EC, **Skinner SA**, Cuccaro M, Simensen RJ, Bishop J, Skinner C, Fender D, Stevenson RE: Autism and maternally derived aberrations of chromosome 15q, Am J Med Genet. 76:327-336, 1998.

17. Erickson RP, **Skinner SA**, Jacquet H, Campion D, Buckley PG, Mantripragada KK, Dumanski JP: Does Chromosome 22 Have Anything to Do With Sex Determination: Further Studies on a 46,XX,22q11.2 Del Male, Am J Med Genet. 123A:64-67, 2003.

18. Percy AK, Lane JB, Childers J, **Skinner S**, Annese F, Barrish J, Caeg E, Glaze DG, MacLeod P: Rett Syndrome: North American Database, J Child Neurol. 22:1338-1341, 2007.

19. Ben-Shachar S, Lanpher B, German JR, Qasaymeh M, Potocki L, Nagamani SC, Franco LM, Malphrus A, Bottenfield GW, Spence JE, Amato S, Rousseau JA, Moghaddam B, Skinner C, **Skinner SA**, Bernes S, Armstrong N, Shinawi M, Stankiewicz P, Patel A, Cheung SW, Lupski JR, Beaudet AL, Sahoo T: Microdeletion 15q13.3: a locus with incomplete penetrance for autism, mental retardation, and psychiatric disorders, J Med Genet. 46:382-8, 2009.

20. Percy AK, Lee HS, Neul JL, Lane JB, **Skinner SA**, Geerts SP, Annese F, Graham J, McNair L, Motil KJ, Barrish JO, Glaze DG: Profiling scoliosis in Rett syndrome, Pediatr Res. 67:435-439, 2010.

21. Kirby RS, Lane JB, Childers J, **Skinner SA**, Annese F, Barrish JO, Glaze DG, Macleod P, Percy AK: Longevity in Rett syndrome: analysis of the North American Database, J Pediatr. 156:135-138, 2010.

22. Glaze DG, Percy AK, **Skinner S**, Motil KJ, Neul JL, Barrish JO, Lane JB, Geerts SP, Annese F, Graham J, McNair L, Lee HS: Epilepsy and the natural history of Rett syndrome, Neurology 74:909-12, 2010.

23. Geer JS, **Skinner SA**, Goldin E, Holden KR: Mucolipidosis type IV: a subtle pediatric neurodegenerative disorder, Pediatr Neurol. 42:223-6, 2010.

24. Gentile JK, Tan WH, Horowitz LT, Bacino CA, **Skinner SA**, Barbieri-Welge R, Bauer-Carlin A, Beaudet AL, Bichell TJ, Lee HS, Sahoo T, Waisbren SE, Bird LM, Peters SU: A neurodevelopmental survey of Angelman syndrome with genotype-phenotype correlations, J Dev Behav Pediatr. 31(7):592-601, 2010.

25. Percy AK, Neul JL, Glaze DG, Motil KJ, **Skinner SA**, Khwaja O, Lee HS, Lane JB, Barrish JO, Annese F, McNair L, Graham J, Barnes K: Rett syndrome diagnostic criteria: lessons from the natural history study, Ann Neurol 68:951-955, 2010.

26. Tan WH, Bacino CA, **Skinner SA**, Anselm I, Barbieri-Welge R, Bauer-Carlin A, Beaudet AL, Bichell TJ, Gentile JK, Glaze DG, Horowitz LT, Kothare SV, Lee HS, Nespeca MP, Peters SU, Sahoo T, Sarco D, Waisbren SE, Bird LM: Angelman syndrome: mutations influence features in early childhood, Am J Med Genet 155(1):81-90, 2011.

27. Bird LM, Tan WH, Bacino CA, Peters SU, **Skinner SA**, Anselm I, Barbieri-Welge R, Bauer-Carlin A, Gentile JK, Glaze DG, Horowitz LT, Mohan KN, Nespeca MP, Sahoo T, Sarco D, Waisbren SE, Beaudet AL. A therapeutic trial of pro-methylation dietary supplements in Angelman syndrome. Am J Med Genet A. 155(20):2956-63, 2011.

28. Motil KJ, Barrish JO, Lane J, Geerts SP, Annese F, McNair L, Percy AK, **Skinner SA**, Neul JL, Glaze, DG: Vitamin D deficiency is prevalent in girls and women with Rett syndrome, J Pediatr Gastroenterol Nutr. 53(5):569-74, 2011.

29. Lane JB, Lee HS, Smith LW, Cheng P, Percy AK, Glaze DG, Neul JL, Motil KJ, Barrish JO, **Skinner SA**, Annese F, McNair L, Graham J, Khwaja O, Barnes K, Krischer JP: Clinical severity and quality of life in children and adolescents with Rett syndrome, Neurol 77(20):1812-18, 2011.

30. McCauley MD, Wang T, Mike E, Herrera J, Beavers DL, Huang TW, Ward CS, **Skinner S**, Percy AK, Glaze DG, Wehrens XH, Neul JL. Pathogenesis of lethal cardiac arrhythmias in mecp2 mutant mice: implication for therapy in Rett syndrome, Sci Transl Med. 14;3(113):113-125, 2011.

31. Shinawi M, Sahoo T, Maramda B, **Skinner SA**, Skinner C, Chinault C, Zascacage R, Pters SU, Patel A Stevenson RE, Beaudet AL. 11p14.1 Microdeletions Associated with ADHD, Autism, Developmental Delay, and Obesity. Am J Med Genet A. 2011 June;155A(6):1272-80. Epub 2011 May 12.

32. Motil KJ, Caeg E, Barrish JO, Geerts S, Lane JB, Percy AK, Annese F, McNair L, **Skinner SA**, Lee HS, Neul JL, Glaze DG. Gastrointestinal and nutritional problems occur frequently throughout life in girls and women with Rett Syndrome. J Pediatr Gastroenterol Nutr 55(3):292-298, 2012.

33. Tarquinio, DC, Motil KJ, Hou W, Lee HS, Glaze DG, **Skinner SA**, Neul JL, Annese F, McNair L, Barrish JO, Geerts SP, Lane JB, Percy AK. Growth failure and outcome in Rett syndrome: Specific growth references. Neurology 79(16):1653-61, 2012. Epub 2012 Nov 21.

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35. Zarate YA, Dwivedi A, Bartel FO, Bellomo MA, Cathey SS, Champaigne NL, Clarkson LK, Dupont BR, Everman DB, Geer JS, Gordon BC, Lichty AW, Lyons MJ, Rogers RC, Saul RA, Schroer RJ, **Skinner SA**, Stevenson RE. Clinical utility of the X-chromosome array, Am J Med Genet A, 161A(1):120-30, 2013.

36. JC Hodge, E Mitchell, V Pillalamarri, TL Toler, F Bartel, HM Kearney, YS Zou, WH Tan, C Hanscom, S Kirmani, RR Hanson, **SA Skinner**, RC Rogers, DB Everman, E Boyd, C Tapp, SV Mullegama, D Keelean-Fuller, CM Powell, SH Elsea, CC Morton, JF Gusella, B DuPont, A Chaubey, AE Lin and ME Talkowski. Disruption of MBD5 contributes to a spectrum of psychopathology and neurodevelomental abnormalities. Molecular Psychiatry (2013), 1-12.

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38. Chapleau CA, Lane J, Kirwin SM, Schanen C, Vinette KM, Stubbolo D, MacLeod P, Glaze DG, Motil KJ, Neul JL, **Skinner SA**, Kaufmann WE, Percy AK. Detection of rarely identified multiple mutations in MECP2 gene do not contribute to enhanced severity in Rett syndrome. Am J Genet A. 2013Jul: 161A(7): 1638-46.

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40. Neul JL, Lane JB, Lee HS, Geerts S, Barrish JO, Annese F, Baggett LM, Barnes K, **Skinner SA**, Motil KJ, Glaze DG, Kaufmann WE, Percy AK. Developmental delay in Rett syndrome: data from the natural history study. J Neurodev Disord. 2014; 6(1):20.

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44. Tarquinio DC, Hou W, Neul JL, Kaufmann WE, Glaze DG, Motil KJ, **Skinner SA**, Lee HS, Percy AK. The Changing Face of Survival in Rett Syndrome and MECP2-Related Disorders Pediatr Neurol. 2015 Nov;53(5):402-11. doi: 10.1016/j.pediatrneurol.2015.06.003. Epub 2015 Jun 26.

45. Killian JT Jr, Lane JB, Lee HS, Pelham JH, **Skinner SA**, Kaufmann WE, Glaze DG, Neul JL, Percy AK. Caretaker Quality of Life in Rett Syndrome: Disorder Features and Psychological Predictors. Pediatr Neurol. 2016 Jan 11. pii: S0887-8994(15)30314-3. doi: 10.1016/j.pediatrneurol.2015.12.021. [Epub ahead of print]

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49. Molinero I, Broman-Fulks J, Lyons MJ, Matheus MG, Chaubey A, DuPont BR, Friez MJ, **Skinner SA**, Holden KR. Importance of Genetic Testing in Global Health During the Evaluation of Familial Microcephaly. Clin Case Rep. 2016 Aug;4(10):968-971.