BIOGRAPHICAL SKETCH

Provide the following information for the Senior/key personnel and other significant contributors.
Follow this format for each person. DO NOT EXCEED FIVE PAGES.

NAME: Stevenson, Roger

eRA COMMONS USER NAME (credential, e.g., agency login): RESTEVENSON

POSITION TITLE: Senior Clinical Geneticist

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable.)*

INSTITUTION AND LOCATION	DEGREE (if applicable)	Completion Date MM/YYYY	FIELD OF STUDY
Furman University, Greenville, SC	BS	1962	
Bowman Gray School of Medicine, Winston-Salem, NC	MD	1966	
Johns Hopkins Hospital, Baltimore, MD	Fellow	1972	

B. Positions and Honors

Positions and Employment

1969 - 1971	Chief of Pediatrics, US Air Force, Eilson Air Force Base Hospital, Fairbanks, AK
1971 - 1972	Research Fellow, Metabolism Division; Fellow, Division of Genetics, Johns Hopkins University School of Medicine, Balitmore, MD
1972 - 1974	Asst. Professor of Pediatrics, University of Texas School of Medicine, Houston, TX
1974 - 1989	Director, Greenwood Genetic Center, Greenwood, SC
1975 - 1984	State Health Care Advisory Board, South Carolina Department of Health and Environmental Control, Columbia, SC
1979 -	Task Force on Biomedical Research, South Carolina Commission on Higher Education, Columbia, SC
1982 -	Senior Clinical Geneticist, Greenwood Genetic Center, Greenwood, SC
1989 - 2004	Director of Research, Greenwood Genetic Center, Greenwood, SC
1997 - 2010	Director, Greenwood Genetic Center, Greenwood, SC
2006 -	Advisory Council, Birth Defects Program of South Carolina, Columbia, SC

Other Experience and Professional Memberships

- Member, American Society of Human Genetics
- Member, South Carolina Pediatric Society
- Member, American Academy of Pediatrics
- Member, South Carolina Medical Association
- Member, American Medical Association
- Organizing Group, David W. Smith Workshop
- Member, American College of Medical Genetics
- Member, South Carolina Commission on Higher Education
- Board Member, American Board of Medical Genetics

Honors

1966	Proctor Award of Excellence in Clinical Pediatrics, Bowman Gray School of Medicine
1966	Mosby Scholastic Achievement Award, Bowman Gray School of Medicine
1974	Best Teacher of Faculty, University of Texas Medical School in Houston
1985	Order of the Palmetto, South Carolina

1989	Distinguished Alumnus Award, Furman University
1993	Honorary Doctorate (Humanities), Lander University
2006	Honorary Doctorate (Science), Clemson University
2010	Honorary Doctorate (Science), Furman University
2010	Weston Award, USC Department of Pediatrics
2011	Society of Scholars , Johns Hopkins University

C. Contribution to Science

1. Birth defects. Study of the embryological anarchy produced by various genetic and environmental influences that lead to birth defects has been a career-long focus of my study and scholarly works. This pursuit began during medical school with my delineation of the effects of maternal phenylketonuria on the developing fetus. Later investigations explored the mechanisms by which sirenomelia, gastroschisis, limb-body wall defects, neural tube defects, and other malformations occur. These studies resulted in numerous peer-reviewed publications and two books (The Fetus and Newly Born Infant, Influences of the Prenatal Environment, WB Saunders 1971, 1977; Human Malformations and Related Anomalies, Oxford, 1985, 2006, 2015). I am one of the 5 organizing members of the David W. Smith Workshop on Malformations and Morphogenesis, an annual international meeting devoted to understanding the mechanisms by which birth defects occur. In 1992, I established the first statewide neural tube defect surveillance program. This model program continues today and has resulted in a greater than a 50% reduction in the number of neural tube defects in South Carolina, a high risk geographic region for these defects.

    a. Bupp CP, Sarasua SM, Dean JH, Stevenson RE. When folic acid fails: Insights from 20 years of neural tube defect surveillance in South Carolina. Am J Med Genet A. 2015 Oct;167A(10):2244-50. PubMed PMID: 26108864.
    b. Hunter AG, Seaver LH, Stevenson RE. Limb-body wall defect. Is there a defensible hypothesis and can it explain all the associated anomalies?. Am J Med Genet A. 2011 Sep;155A(9):2045-59. PubMed PMID: 21815262.
    c. Stevenson RE, Rogers RC, Chandler JC, Gauderer MW, Hunter AG. Escape of the yolk sac: a hypothesis to explain the embryogenesis of gastroschisis. Clin Genet. 2009 Apr;75(4):326-33. PubMed PMID: 19419415.
    d. Stevenson RE, Jones KL, Phelan MC, Jones MC, Barr M Jr, Clericuzio C, Harley RA, Benirschke K. Vascular steal: the pathogenetic mechanism producing sirenomelia and associated defects of the viscera and soft tissues. Pediatrics. 1986 Sep;78(3):451-7. PubMed PMID: 3748679.

2. Intellectual Disability. Delineation of the causes of intellectual disability, the associated environmental and genetic syndromes, and strategies for prevention or curative therapies comprise the second area of my research endeavors. Early in my career, my focus was on metabolic disturbances that have intellectual disability as a component manifestation; beginning in the late 1980s, the focus shifted to X-linked intellectual disability. The latter studies were conducted in collaboration with Dr. Charles Schwartz and other investigators in the United States, Europe, Australia, South Africa and South America with whom 28 XLID syndromes were clinically defined/refined, 19 XLID syndromes were mapped and 24 causative genes were identified, resulting in 102 peer-reviewed publications and one book (Atlas of X-Linked Disability Syndromes, Oxford, 2000, 2012). Together we hosted the 12th International Conference on Fragile X and X-linked intellectual disability (Williamsburg, VA, 2005).

a. Lubs HA, Stevenson RE, Schwartz CE. Fragile X and X-linked intellectual disability: four decades of discovery. Am J Hum Genet. 2012 Apr 6;90(4):579-90. PubMed PMID: 22482801; PubMed Central PMCID: PMC3322227.
b. Stevenson RE, Arena JF, Ouzts E, Gibson A, Shokeir MH, Vnencak-Jones C, Lubs HA, May M, Schwartz CE. Renpenning syndrome maps to Xp11. Am J Hum Genet. 1998 May;62(5):1092-101. PubMed PMID: 9545405; PubMed Central PMCID: PMC1377092.
c. Stevenson RE, Goodman HO, Schwartz CE, Simensen RJ, McLean WT Jr, Herndon CN. Allan-Herndon syndrome. I. Clinical studies. Am J Hum Genet. 1990 Sep;47(3):446-53. PubMed PMID: 2393019; PubMed Central PMCID: PMC1683863.

3. Autism. The failure of traditional genetic approaches to solve the enigma of autism prompted my research into this frequent childhood disability. Beginning in 1995, we began organizing well-phenotyped cohorts of children with autism for longitudinal and intensive inquiry into potential genetic, genomic, epigenetic, and metabolic contribution to this casually heterogeneous disability. I have directed the South Carolina Autism Project: A Search for Causes which has identified several specific genetic and epigenetic etiologies of autism and now is exploring global disturbances in metabolism which appear to be the most consistent laboratory findings.

a. 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. 1998 Apr 1;76(4):327-36. PubMed PMID: 9545097.
b. Jones JR, Skinner C, Friez MJ, Schwartz CE, Stevenson RE. Hypothesis: dysregulation of methylation of brain-expressed genes on the X chromosome and autism spectrum disorders. Am J Med Genet A. 2008 Sep 1;146A(17):2213-20. PubMed PMID: 18698615.
c. Boccuto L, Lauri M, Sarasua SM, Skinner CD, Buccella D, Dwivedi A, Orteschi D, Collins JS, Zollino M, Visconti P, Dupont B, Tiziano D, Schroer RJ, Neri G, Stevenson RE, Gurrieri F, Schwartz CE. Prevalence of SHANK3 variants in patients with different subtypes of autism spectrum disorders. Eur J Hum Genet. 2013 Mar;21(3):310-6. PubMed PMID: 22892527; PubMed Central PMCID: PMC3573207.
d. Boccuto L, Chen CF, Pittman AR, Skinner CD, McCartney HJ, Jones K, Bochner BR, Stevenson RE, Schwartz CE. Decreased tryptophan metabolism in patients with autism spectrum disorders. Mol Autism. 2013 Jun 3;4(1):16. PubMed PMID: 23731516; PubMed Central PMCID: PMC3680090.

Complete List of Published Work in My Bibliography:
http://1.usa.gov/1Yfoe6o

D. Additional Information: Research Support and/or Scholastic Performance

Ongoing Research Support

2017-43, SCDDSN
Stevenson, Roger (PI)
07/01/09-06/30/17
SC Neural Tube Defect Prevention Program

The goals of this project are to identify and characterize all NTDs that occur in South Carolina and to prevent occurrences and recurrences of Neural Tube Defects.
Role: PI

2017-30, SCDDSN
Stevenson, Roger (PI)
07/01/12-06/30/17
Autism: Clinical, Biochemical, Genetic, Genomic, and Epigenetic Testing
The goals are to identify various causes or predispositions to autism, to develop a blood-based screening/diagnostic test, and to develop a roadmap to treatment.
Role: PI