Stanford

David Stevenson

Professor of Pediatrics (Genetics) at the Lucile Salter Packard Children's Hospital

Pediatrics - Medical Genetics

CLINICAL OFFICES

- **Pediatric Genetics**

  730 Welch Rd 2A

  Palo Alto, CA 94304

  Tel (650) 721-5804 **Fax** (650) 498-4555

Bio

BIO

David A. Stevenson, MD is a physician board certified in both pediatrics and medical genetics. He completed his pediatric residency at the University of New Mexico and completed his medical genetics residency at the University of Utah.

Dr. Stevenson is the program director for the Combined Pediatric-Medical Genetics Residency Program and the Medical Genetics Residency Program at Stanford. He is actively involved in graduate medical education and developing innovative ways of training the next generation of medical geneticists. In addition, as co-director of the Genetic Testing Optimization Service, he focuses on researching best practices for genetic testing utilization.

Dr. Stevenson sees all types of individuals with various genetic disorders in his clinical practice. However, he has particular interests in disorders of the Ras/MAPK pathway which includes neurofibromatosis type 1, Noonan syndrome, CFC syndrome, and Costello syndrome. He also has expertise in evaluating individuals with vascular anomalies including hereditary hemorrhagic telangiectasia, and skeletal dysplasias. Dr. Stevenson is on the scientific advisory board for Prader-Willi Syndrome Association (PWSA) and has a focus on treating individuals with Prader-Willi syndrome. He has research interests in identifying clinical trial endpoints and is actively involved in clinical trials.

CLINICAL FOCUS

- Clinical Genetics

ACADEMIC APPOINTMENTS

- Professor - Med Center Line, Pediatrics - Medical Genetics
- Member, Bio-X
- Member, Maternal & Child Health Research Institute (MCHRI)

ADMINISTRATIVE APPOINTMENTS

- Program Director, Combined Pediatric-Medical Genetics Residency, Stanford University, (2017- present)
- Program Director, Medical Genetics Residency, Stanford University, (2016- present)

- Co-Director, Genetic Testing Optimization Service, Stanford University, (2015- present)
- Service Chief, Medical Genetics, Stanford University, (2016- present)

HONORS AND AWARDS

- Alice L. Jee Memorial Young Investigator Award, Orthopaedic Research Society (2006)
- Junior Physician Investigator Award, Western Society for Pediatric Research (2009)
- AFMR Scholar Award, AFMR (2010)
- Young Investigator Research Award, Western Society for Pediatric Research (2009)
- Mid/Senior Career Clinical Award of Excellence, Stanford University Department of Pediatrics (2017)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- WSPR Council, Western Society of Pediatric Research (2011 - 2014)
- ACMGG Program Committee, American College of Medical Genetics and Genomics (2017 - present)

PROFESSIONAL EDUCATION

- Residency: University of New Mexico School of Medicine NM
- Internship: University of New Mexico School of Medicine (2000) NM
- Board Certification: Clinical Genetics, American Board of Medical Genetics and Genomics (2005)
- Fellowship: University of Utah (2005) UT
- Board Certification: Pediatrics, American Board of Pediatrics (2002)
- Medical Education: University of Utah (1995) UT
- Medical Genetics Fellowship, University of Utah , Medical Genetics (2005)
- Pediatric Residency, University of New Mexico , Pediatrics (2002)
- MD, University of Utah , Medicine (1999)
- BA, Utah State University , Biology (1995)

COMMUNITY AND INTERNATIONAL WORK

- CFC International Medical Advisory Board
- Co-chair, Costello Syndrome Professional Advisory Committee
- PWSA Scientific Advisory Board

Research & Scholarship

RESEARCH INTERESTS

- Research Methods
- Science Education

CURRENT RESEARCH AND SCHOLARLY INTERESTS

My research focuses on disorders of the RAS/MAPK pathway (e.g. NF1, Noonan, CFC, and Costello syndrome). I am working on understanding the impact of RAS signaling on the musculoskeletal system. Through multi-disciplinary collaborations I am utilizing genomic approaches to identify somatic events and modifiers in the RASopathies. I am also involved in identifying outcome measures for use in clinical trials for the associated orthopedic manifestations. Other areas of research involve vascular anomalies, Prader-Willi syndrome, and hypophosphatasia.

CLINICAL TRIALS

- A Study of Diazoxide Choline in Patients With Prader-Willi Syndrome, Not Recruiting

Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Medical Genetics (Fellowship Program)

Publications

PUBLICATIONS

- **Localization and age distribution of telangiectases in children and adolescents with hereditary hemorrhagic telangiectasia: A retrospective cohort study** *JOURNAL OF THE AMERICAN ACADEMY OF DERMATOLOGY*
  Gonzalez, C. D., Cipriano, S. D., Topham, C. A., Stevenson, D. A., Whitehead, K. J., Vanderhooft, S., Presson, A. P., McDonald, J.
  2019; 81 (4): 950–55

- **Phenotype of CM-AVM2 caused by variants in EPHB4: how much overlap with hereditary hemorrhagic telangiectasia (HHT)?** *GENETICS IN MEDICINE*
  Wooderchak-Donahue, W. L., Akay, G., Whitehead, K., Briggs, E., Stevenson, D. A., O'Fallon, B., Velinder, M., Farrell, A., Shen, W., Bedoukian, E., Skrabann, C. M., Antaya, R. J., Henderson, et al
  2019; 21 (9): 2007–14

- **Cardiac transplantation in children with Noonan syndrome.** *Pediatric transplantation*
  McCallen, L. M., Ameduri, R. K., Denfield, S. W., Dodd, D. A., Everitt, M. D., Johnson, J. N., Lee, T. M., Lin, A. E., Lohr, J. L., May, L. J., Pierpont, M. E., Stevenson, D. A., Chatfield, et al
  2019: e13535

- **PTPN11 Gain-of-Function Mutations Affect the Developing Human Brain, Memory, and Attention** *CEREBRAL CORTEX*
  Johnson, E. M., Ishak, A. D., Naylor, P. E., Stevenson, D. A., Reiss, A. L., Green, T.
  2019; 29 (7): 2915–23

- **Costello syndrome: Clinical phenotype, genotype, and management guidelines.** *American journal of medical genetics. Part A*
  Gripp, K. W., Morse, L. A., Axelrad, M., Chatfield, K. C., Chidekel, A., Dobyns, W., Doyle, D., Kerr, B., Lin, A. E., Schwartz, D. D., Sibbles, B. J., Siegel, D., Shankar, et al
  2019

- **Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Kumar, A., Zastrow, D. B., Kravets, E. J., Beleford, D., Ruzhnikov, M. Z., Grove, M. E., Dries, A. M., Kohler, J. N., Waggott, D. M., Yang, Y., Huang, Y., Mackenzie, K. M., Eng, et al
  2019; 179 (6): 966–77

- **NF1 Somatic Mutation in Dystrophic Scoliosis** *JOURNAL OF MOLECULAR NEUROSCIENCE*
  Margraf, R. L., VanSant-Webb, C., Mao, R., Viskochil, D. H., Carey, J., Hanson, H., D'Astous, J., Grossmann, A., Stevenson, D. A.
  2019; 68 (1): 11–18

- **Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review.** *American journal of medical genetics. Part A*
  Kumar, A., Zastrow, D. B., Kravets, E. J., Beleford, D., Ruzhnikov, M. R., Grove, M. E., Dries, A. M., Kohler, J. N., Waggott, D. M., Yang, Y., Huang, Y., Undiagnosed Diseases Network, Mackenzie, K. M., et al
  2019

- **Localization and age distribution of telangiectases in children and adolescents with hereditary hemorrhagic telangiectasia: A retrospective cohort study.** *Journal of the American Academy of Dermatology*
  Gonzalez, C. D., Cipriano, S. D., Topham, C. A., Stevenson, D. A., Whitehead, K. J., Vanderhooft, S., Presson, A. P., McDonald, J.
  2019

- **NF1 Somatic Mutation in Dystrophic Scoliosis.** *Journal of molecular neuroscience : MN*
  Margraf, R. L., VanSant-Webb, C., Mao, R., Viskochil, D. H., Carey, J., Hanson, H., D'Astous, J., Grossmann, A., Stevenson, D. A.

2019

- **Phenotype of CM-AVM2 caused by variants in EPHB4: how much overlap with hereditary hemorrhagic telangiectasia (HHT)?** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Wooderchak-Donahue, W. L., Akay, G., Whitehead, K., Briggs, E., Stevenson, D. A., O'Fallon, B., Velinder, M., Farrell, A., Shen, W., Bedoukian, E., Skrabann, C. M., Antaya, R. J., Henderson, et al
  2019
- **Contributing factors of mortality in Prader-Willi syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Proffitt, J., Osann, K., McManus, B., Kimonis, V. E., Heinemann, J., Butler, M. G., Stevenson, D. A., Gold, J.
  2019; 179 (2): 196–205
- **Contributing factors of mortality in Prader-Willi syndrome.** *American journal of medical genetics. Part A*
  Proffitt, J., Osann, K., McManus, B., Kimonis, V. E., Heinemann, J., Butler, M. G., Stevenson, D. A., Gold, J.
  2018
- **Proceedings of the fifth international RASopathies symposium: When development and cancer intersect** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Rauen, K. A., Schoyer, L., Schill, L., Stronach, B., Albeck, J., Andresen, B. S., Cave, H., Ellis, M., Fruchtman, S. M., Gelb, B. D., Gibson, C. C., Gripp, K., Hefner, et al
  2018; 176 (12): 2924–29
- **Proceedings of the fifth international RASopathies symposium: When development and cancer intersect.** *American journal of medical genetics. Part A*
  Rauen, K. A., Schoyer, L., Schill, L., Stronach, B., Albeck, J., Andresen, B. S., Cave, H., Ellis, M., Fruchtman, S. M., Gelb, B. D., Gibson, C. C., Gripp, K., Hefner, et al
  2018: e40632
- **Response to Hannah-Shmouni and Stratakis.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Stewart, D. R., Korf, B. R., Nathanson, K. L., Stevenson, D. A., Yohay, K.
  2018
- **Expanding the clinical and molecular findings in RASA1 capillary malformation-arteriovenous malformation** *EUROPEAN JOURNAL OF HUMAN GENETICS*
  Wooderchak-Donahue, W. L., Johnson, P., McDonald, J., Blei, F., Berenstein, A., Sorscher, M., Mayer, J., Scheuerle, A. E., Lewis, T., Grimmer, J., Richter, G. T., Steeves, M. A., Lin, et al
  2018; 26 (10): 1521–36
- **A case report of a suspected dual diagnosis: 22q11.2 deletion syndrome and X-linked chondrodysplasia punctata** *CLINICAL DYSMORPHOLOGY*
  Brimble, E., Pacione, M., Farrelly, E., Stevenson, D. A., Ruzhnikov, M. Z.
  2018; 27 (4): 151–53
- **Predictive Value and Interrater Reliability of Radiographic Factors in Neurofibromatosis Patients With Dystrophic Scoliosis.** *Spine deformity*
  Larson, A. N., Ledonio, C. G., Brearley, A. M., Sucato, D. J., Carreon, L. Y., Crawford, A. H., Stevenson, D. A., Vitale, M. G., Moertel, C. L., Polly, D. W.
  2018; 6 (5): 560–67
- **Racial/ethnic disparities and incidence of malignant peripheral nerve sheath tumors: results from the Surveillance, Epidemiology, and End Results Program, 2000-2014** *JOURNAL OF NEURO-ONCOLOGY*
  Peckham-Gregory, E. C., Montenegro, R. E., Stevenson, D. A., Viskochil, D. H., Scheurer, M. E., Lupo, P. J., Schiffman, J. D.
  2018; 139 (1): 69–75
- **Use of Flow Cytometry for Diagnosis of Epilepsy Associated With Homozygous PIGW Variants** *PEDIATRIC NEUROLOGY*
  Foskett, G., Engleman, E., Klotz, J., Choi, O., Tolentino, L., Kochhar, A., Yang, Q., Stevenson, D. A.
  2018; 85: 67–70
- **PTPN11 Gain-of-Function Mutations Affect the Developing Human Brain, Memory, and Attention.** *Cerebral cortex (New York, N.Y. : 1991)*
  Johnson, E. M., Ishak, A. D., Naylor, P. E., Stevenson, D. A., Reiss, A. L., Green, T.
  2018
- **Care of adults with neurofibromatosis type 1: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG)** *GENETICS IN MEDICINE*
  Stewart, D. R., Korf, B. R., Nathanson, K. L., Stevenson, D. A., Yohay, K.

2018; 20 (7): 671–82

- **Epistaxis in Children and Adolescents With Hereditary Hemorrhagic Telangiectasia**
  Gonzalez, C. D., Mcdonald, J., Stevenson, D. A., Whitehead, K. J., Petersen, M. G., Presson, A. P., Ding, Q., Wilson, K. F.
  WILEY.2018: 1714–19

- **Use of Flow Cytometry for Diagnosis of Epilepsy Associated With Homozygous PIGW Variants.** *Pediatric neurology*
  Foskett, G. K., Engleman, E., Klotz, J., Choi, O., Tolentino, L., Kochhar, A., Yang, Q. Z., Stevenson, D. A.
  2018

- **Evaluation of racial disparities in pediatric optic pathway glioma incidence: Results from the Surveillance, Epidemiology, and End Results Program, 2000-2014** *CANCER EPIDEMIOLOGY*
  Peckham-Gregory, E. C., Montenegro, R. E., Stevenson, D. A., Viskochil, D. H., Scheurer, M. E., Lupo, P. J., Schiffman, J. D.
  2018; 54: 90–94

- **Quantitative Ultrasound and Tibial Dysplasia in Neurofibromatosis Type 1** *JOURNAL OF CLINICAL DENSITOMETRY*
  Stevenson, D. A., Hanson, H., Stevens, A., Carey, J., Viskochil, D., Sheng, X., Wheeler, K., Slater, H.
  2018; 21 (2): 179–84

- **Dietary intervention rescues myopathy associated with neurofibromatosis type 1** *HUMAN MOLECULAR GENETICS*
  Summers, M. A., Rupasinghe, T., Vasiljevski, E. R., Evesson, F. J., Mikulec, K., Peacock, L., Quinlan, K. R., Cooper, S. T., Roessner, U., Stevenson, D. A., Little, D. G., Schindeler, A.
  2018; 27 (4): 577–88

- **Bilirubin Production Is Increased in Newborn Mice Exposed to Isoflurane.** *Neonatology*
  Iwatani, S., Burgess, J., Kalish, F., Wong, R. J., Stevenson, D. K.
  2018; 115 (1): 21-27

- **Epistaxis in children and adolescents with hereditary hemorrhagic telangiectasia.** *The Laryngoscope*
  Gonzalez, C. D., Mcdonald, J., Stevenson, D. A., Whitehead, K. J., Petersen, M. G., Presson, A. P., Ding, Q., Wilson, K. F.
  2018; 128 (7): 1714–19

- **Variable clinical course of identical twin neonates with Alström syndrome presenting coincidentally with dilated cardiomyopathy.** *American journal of medical genetics. Part A*
  Hollander, S. A., Alsaleh, N., Ruzhnikov, M., Jensen, K., Rosenthal, D. N., Stevenson, D. A., Manning, M.
  2017; 173 (6): 1687-1689

- **The path forward: 2015 International Children's Tumor Foundation conference on neurofibromatosis type 1, type 2, and schwannomatosis.** *American journal of medical genetics. Part A*
  Blakeley, J. O., Bakker, A., Barker, A., Clapp, W., Ferner, R., Fisher, M. J., Giovannini, M., Gutmann, D. H., Karajannis, M. A., Kissil, J. L., Legius, E., Lloyd, A. C., Packer, et al
  2017; 173 (6): 1714-1721

- **Utilization of Whole-Exome Next-Generation Sequencing Variant Read Frequency for Detection of Lesion-Specific, Somatic Loss of Heterozygosity in a Neurofibromatosis Type 1 Cohort with Tibial Pseudarthrosis** *JOURNAL OF MOLECULAR DIAGNOSTICS*
  Margraf, R. L., VanSant-Webb, C., Sant, D., Carey, J., Hanson, H., D'Astous, J., Viskochil, D., Stevenson, D. A., Mao, R.
  2017; 19 (3): 468-474

- **Analysis of Copy Number Variants in 11 Pairs of Monozygotic Twins with Neurofibromatosis Type** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Sites, E. R., Smolarek, T. A., Martin, L. J., Viskochil, D. H., Stevenson, D. A., Ullrich, N. J., Messiaen, L. M., Schorry, E. K.
  2017; 173 (3): 647-653

- **Promoting appropriate genetic testing: the impact of a combined test review and consultative service.** *Genetics in medicine*
  Suarez, C. J., Yu, L., Downs, N., Costa, H. A., Stevenson, D. A.
  2017

- **Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders.** *Genome medicine*
  Bostwick, B. L., McLean, S., Posey, J. E., Streff, H. E., Gripp, K. W., Blesson, A., Powell-Hamilton, N., Tusi, J., Stevenson, D. A., Farrelly, E., Hudgins, L., Yang, Y., Xia, et al

2017; 9 (1): 73

- **Molecular and clinical spectra of FBXL4 deficiency.** *Human mutation*
  El-Hattab, A. W., Dai, H., Almannai, M., Wang, J., Faqeih, E. A., Al Asmari, A., Saleh, M. A., Elamin, M. A., Alfadhel, M., Alkuraya, F. S., Hashem, M., Aldosary, M. S., Almass, et al
  2017

- **Brief Report: The Prevalence of Neurofibromatosis Type 1 among Children with Autism Spectrum Disorder Identified by the Autism and Developmental Disabilities Monitoring Network** *JOURNAL OF AUTISM AND DEVELOPMENTAL DISORDERS*
  Bilder, D. A., Bakian, A. V., Stevenson, D. A., Carbone, P. S., Cunniff, C., Goodman, A. B., McMahon, W. M., Fisher, N. P., Viskochil, D.
  2016; 46 (10): 3369-3376

- **NALCN channelopathies: Distinguishing gain-of-function and loss-of-function mutations** *NEUROLOGY*
  Bend, E. G., Si, Y., Stevenson, D. A., Bayrak-Toydemir, P., Newcomb, T. M., Jorgensen, E. M., Swoboda, K. J.
  2016; 87 (11): 1131-1139

- **DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome.** *American journal of human genetics*
  Joshi, R. S., Garg, P., Zaitlen, N., Lappalainen, T., Watson, C. T., Azam, N., Ho, D., Li, X., Antonarakis, S. E., Brunner, H. G., Buiting, K., Cheung, S. W., Coffee, et al
  2016; 99 (3): 555-566

- **The Fourth International Symposium on Genetic Disorders of the Ras/MAPK pathway** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Schill, L., Schoyer, L., Andresen, B. S., Bakker, A., Bayrak-Toydemir, P., Burkitt-Wright, E., Chatfield, K., Elefteriou, F., Elgersma, Y., Fisher, M. J., Franz, D., Gelb, et al
  2016; 170 (8): 1959-1966

- **Respiratory System Involvement in Costello Syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Gomez-Ospina, N., Kuo, C., Ananth, A. L., Myers, A., Brennan, M., Stevenson, D. A., Bernstein, J. A., Hudgins, L.
  2016; 170 (7): 1849-1857

- **RASA1 somatic mutation and variable expressivity in capillary malformation/arteriovenous malformation (CM/AVM) syndrome.** *American journal of medical genetics. Part A*
  Macmurdo, C. F., Wooderchak-Donahue, W., Bayrak-Toydemir, P., Le, J., Wallenstein, M. B., Milla, C., Teng, J. M., Bernstein, J. A., Stevenson, D. A.
  2016; 170 (6): 1450-1454

- **The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer** *PLOS GENETICS*
  Hartung, A., Swensen, J., Uriz, I. E., Lapin, M., Kristjansdottir, K., Petersen, U. S., Bang, J. M., Guerra, B., Andersen, H. S., Dobrowolski, S. F., Carey, J. C., Yu, P., Vaughn, et al
  2016; 12 (5)

- **Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure** *GENETICS IN MEDICINE*
  Mulchandani, S., Bhoj, E. J., Luo, M., Powell-Hamilton, N., Jenny, K., Gripp, K. W., Elbracht, M., Eggermann, T., Turner, C. L., Temple, I. K., Mackay, D. J., Dubbs, H., Stevenson, et al
  2016; 18 (4): 309-315

- **The Occurrence of Occult Acetabular Dysplasia in Relatives of Individuals With Developmental Dysplasia of the Hip.** *Journal of pediatric orthopedics*
  Carroll, K. L., Schiffern, A. N., Murray, K. A., Stevenson, D. A., Viskochil, D. H., Toydemir, R., MacWilliams, B. A., Roach, J. W.
  2016; 36 (1): 96-100

- **Molecular diagnostics in the new era: clinical utility of a next generation sequencing panel in the diagnosis of HHT**
  Wooderchak-Donahue, W. L., McDonald, J., VanSant-Webb, C., Lewis, T., Stevenson, D. A., Bayrak-Toydemir, P.
  SPRINGER.2015: 533

- **An Attenuated Phenotype of Costello Syndrome in Three Unrelated Individuals with a HRAS c.179G>A (p.Gly60Asp) Mutation Correlates with Uncommon Functional Consequences** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Gripp, K. W., Sol-Church, K., Smpokou, P., Graham, G. E., Stevenson, D. A., Hanson, H., Viskochil, D. H., Baker, L. C., Russo, B., Gardner, N., Stabley, D. L., Kolbe, V., Rosenberger, et al
  2015; 167 (9): 2085-2097

- **An attenuated phenotype of Costello syndrome in three unrelated individuals with a HRAS c.179G>A (p.Gly60Asp) mutation correlates with uncommon functional consequences.** *American journal of medical genetics. Part A*

Gripp, K. W., Sol-Church, K., Smpokou, P., Graham, G. E., Stevenson, D. A., Hanson, H., Viskochil, D. H., Baker, L. C., Russo, B., Gardner, N., Stabley, D. L., Kolbe, V., Rosenberger, et al
2015; 167A (9): 2085-2097

- **Stress and Coping in Parents of Children with Prader-Willi Syndrome: Assessment of the Impact of a Structured Plan of Care** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Tvrdik, T., Mason, D., Dent, K. M., Thornton, L., Hornton, S. N., Viskochil, D. H., Stevenson, D. A.
  2015; 167A (5): 974-982

- **Corrigendum: Asfotase-a improves bone growth, mineralization and strength in mouse models of neurofibromatosis type-1.** *Nature medicine*
  de la Croix Ndong, J., Makowski, A. J., Uppuganti, S., Vignaux, G., Ono, K., Perrien, D. S., Joubert, S., Baglio, S. R., Granchi, D., Stevenson, D. A., Rios, J. J., Nyman, J. S., Elefteriou, et al
  2015; 21 (4): 414-?

- **Evaluation of somatic mutations in tibial pseudarthrosis samples in neurofibromatosis type 1** *JOURNAL OF MEDICAL GENETICS*
  Sant, D. W., Margraf, R. L., Stevenson, D. A., Grossmann, A. H., Viskochil, D. H., Hanson, H., Everitt, M. D., Rios, J. J., Elefteriou, F., Hennessey, T., Mao, R.
  2015; 52 (4): 256-261

- **Dystrophic Spinal Deformities in a Neurofibromatosis Type 1 Murine Model** *PLOS ONE*
  Rhodes, S. D., Zhang, W., Yang, D., Yang, H., Chen, S., Wu, X., Li, X., Yang, X., Mohammad, K. S., Guise, T. A., Bergner, A. L., Stevenson, D. A., Yang, et al
  2015; 10 (3)

- **Function and disability in children with Costello syndrome and Cardiofaciocutaneous syndrome.** *American journal of medical genetics. Part A*
  Johnson, B., Goldberg-Strassler, D., Gripp, K., Thacker, M., Leoni, C., Stevenson, D.
  2015; 167 (1): 40-44

- **Impaired PIEZO1 function in patients with a novel autosomal recessive congenital lymphatic dysplasia.** *Nature communications*
  Lukacs, V., Mathur, J., Mao, R., Bayrak-Toydemir, P., Procter, M., Cahalan, S. M., Kim, H. J., Bandell, M., Longo, N., Day, R. W., Stevenson, D. A., Patapoutian, A., Krock, et al
  2015; 6: 8329-?

- **Genetic Variants Associated with Port-Wine Stains.** *PloS one*
  Frigerio, A., Wright, K., Wooderchak-Donahue, W., Tan, O. T., Margraf, R., Stevenson, D. A., Grimmer, J. F., Bayrak-Toydemir, P.
  2015; 10 (7): e0133158

- **Impaired PIEZO1 function in patients with a novel autosomal recessive congenital lymphatic dysplasia.** *Nature communications*
  Lukacs, V., Mathur, J., Mao, R., Bayrak-Toydemir, P., Procter, M., Cahalan, S. M., Kim, H. J., Bandell, M., Longo, N., Day, R. W., Stevenson, D. A., Patapoutian, A., Krock, et al
  2015; 6: 8329-?

- **Genetic Variants Associated with Port-Wine Stains.** *PloS one*
  Frigerio, A., Wright, K., Wooderchak-Donahue, W., Tan, O. T., Margraf, R., Stevenson, D. A., Grimmer, J. F., Bayrak-Toydemir, P.
  2015; 10 (7)

- **Activity and participation in children with neurofibromatosis type 1** *RESEARCH IN DEVELOPMENTAL DISABILITIES*
  Johnson, B. A., Sheng, X., Perry, A. S., Stevenson, D. A.
  2015; 36: 213-221

- **Dystrophic spinal deformities in a neurofibromatosis type 1 murine model.** *PloS one*
  Rhodes, S. D., Zhang, W., Yang, D., Yang, H., Chen, S., Wu, X., Li, X., Yang, X., Mohammad, K. S., Guise, T. A., Bergner, A. L., Stevenson, D. A., Yang, et al
  2015; 10 (3)

- **The Cyclic AMP Pathway Is a Sex-Specific Modifier of Glioma Risk in Type I Neurofibromatosis Patients.** *Cancer research*
  Warrington, N. M., Sun, T., Luo, J., McKinstry, R. C., Parkin, P. C., Ganzhorn, S., Spoljaric, D., Albers, A. C., Merkelson, A., Stewart, D. R., Stevenson, D. A., Viskochil, D., Druley, et al
  2015; 75 (1): 16-21

- **Hereditary hemorrhagic telangiectasia: genetics and molecular diagnostics in a new era.** *Frontiers in genetics*
  McDonald, J., Wooderchak-Donahue, W., VanSant Webb, C., Whitehead, K., Stevenson, D. A., Bayrak-Toydemir, P.
  2015; 6: 1-?

- **Update from the 2013 International Neurofibromatosis Conference** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Plotkin, S. R., Albers, A. C., Babovic-Vuksanovic, D., Blakeley, J. O., Breakefield, X. O., Dunn, C. M., Evans, D. G., Fisher, M. J., Friedman, J. M., Giovannini, M., Gutmann, D. H., Kalamarides, M., McClatchey, et al
  2014; 164A (12): 2969-2978

- **Neurofibromin Deficiency-Associated Transcriptional Dysregulation Suggests a Novel Therapy for Tibial Pseudoarthrosis in NF1** *JOURNAL OF BONE AND MINERAL RESEARCH*
  Paria, N., Cho, T., Choi, I. H., Kamiya, N., Kayembe, K., Mao, R., Margraf, R. L., Obermosser, G., Oxendine, I., Sant, D. W., Song, M. H., Stevenson, D. A., Viskochil, et al
  2014; 29 (12): 2636-2642

- **Goltz syndrome and PORCN mosaicism** *INTERNATIONAL JOURNAL OF DERMATOLOGY*
  Stevenson, D. A., Chirpich, M., Contreras, Y., Hanson, H., Dent, K.
  2014; 53 (12): 1481-1484

- **Neural Tube Defects and Atypical Deletion on 22q11.2** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Leoni, C., Stevenson, D. A., Geiersbach, K. B., Paxton, C. N., Krock, B. L., Mao, R., Rope, A. F.
  2014; 164A (11): 2701-2706

- **Asfotase-alpha improves bone growth, mineralization and strength in mouse models of neurofibromatosis type-1** *NATURE MEDICINE*
  Ndong, J. d., Makowski, A. J., Uppuganti, S., Vignaux, G., Ono, K., Perrien, D. S., Joubert, S., Baglio, S. R., Granchi, D., Stevenson, D. A., Rios, J. J., Nyman, J. S., Elefteriou, et al
  2014; 20 (8): 904-910

- **Screening children with neurofibromatosis type 1 for autism spectrum disorder** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Tinker, J., Carbone, P. S., Viskochil, D., Mathiesen, A., Ma, K., Stevenson, D. A.
  2014; 164 (7): 1706-1712

- **L1CAM whole gene deletion in a child with L1 syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Chidsey, B. A., Baldwin, E. E., Toydemir, R., Ahles, L., Hanson, H., Stevenson, D. A.
  2014; 164 (6): 1555-1558

- **Postural control in children with and without neurofibromatosis type 1** *HUMAN MOVEMENT SCIENCE*
  Johnson, B. A., MacWilliams, B. A., Stevenson, D. A.
  2014; 34: 157-163

- **CTF Meeting 2012: Translation of the Basic Understanding of the Biology and Genetics of NF1, NF2, and Schwannomatosis Toward the Development of Effective Therapies** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Widemann, B. C., Acosta, M. T., Ammoun, S., Belzberg, A. J., Bernards, A., Blakeley, J., Bretscher, A., Cichowski, K., Clapp, D. W., Dombi, E., Evans, G. D., Ferner, R., Fernandez-Valle, et al
  2014; 164 (3): 563-578

- **Multiscale, Converging Defects of Macro-Porosity, Microstructure and Matrix Mineralization Impact Long Bone Fragility in NF1** *PLOS ONE*
  Kuehnisch, J., Seto, J., Lange, C., Schrof, S., Stumpp, S., Kobus, K., Grohmann, J., Kossler, N., Varga, P., Osswald, M., Emmerich, D., Tinschert, S., Thielemann, et al
  2014; 9 (1)

- **An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge** *GENOME BIOLOGY*
  Brownstein, C. A., Beggs, A. H., Homer, N., Merriman, B., Yu, T. W., Flannery, K. C., DeChene, E. T., Towne, M. C., Savage, S. K., Price, E. N., Holm, I. A., Luquette, L. J., Lyon, et al
  2014; 15 (3)

- **Biomarkers of bone remodeling in children with mucopolysaccharidosis types I, II, and VI.** *Journal of pediatric rehabilitation medicine*
  Stevenson, D. A., Rudser, K., Kunin-Batson, A., Fung, E. B., Viskochil, D., Shapiro, E., Orchard, P. J., Whitley, C. B., Polgreen, L. E.
  2014; 7 (2): 159-165

- **Activity and participation in children with neurofibromatosis type 1.** *Research in developmental disabilities*
  Johnson, B. A., Sheng, X., Perry, A. S., Stevenson, D. A.
  2014; 36C: 213–21

- **Low Bone Mineral Content and Challenges in Interpretation of Dual-Energy X-Ray Absorptiometry in Children With Mucopolysaccharidosis Types I, II, and VI** *JOURNAL OF CLINICAL DENSITOMETRY*
  Polgreen, L. E., Thomas, W., Fung, E., Viskochil, D., Stevenson, D. A., Steinberger, J., Orchard, P., Whitley, C. B., Ensrud, K. E.
  2014; 17 (1): 200-206

- **Decreased bone mineral density in Costello syndrome** *MOLECULAR GENETICS AND METABOLISM*
  Leoni, C., Stevenson, D. A., Martini, L., De Sanctis, R., Mascolo, G., Pantaleoni, F., De Santis, S., La Torraca, I., Persichilli, S., Caradonna, P., Tartaglia, M., Zampino, G.
  2014; 111 (1): 41-45

- **Hyperactive Ras/MAPK signaling is critical for tibial nonunion fracture in neurofibromin-deficient mice** *HUMAN MOLECULAR GENETICS*
  Sharma, R., Wu, X., Rhodes, S. D., Chen, S., He, Y., Yuan, J., Li, J., Yang, X., Li, X., Jiang, L., Kim, E. T., Stevenson, D. A., Viskochil, et al
  2013; 22 (23): 4818-4828

- **BMP9 Mutations Cause a Vascular-Anomaly Syndrome with Phenotypic Overlap with Hereditary Hemorrhagic Telangiectasia** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Wooderchak-Donahue, W. L., McDonald, J., O'Fallon, B., Upton, P. D., Li, W., Roman, B. L., Young, S., Plant, P., Fueloep, G. T., Langa, C., Morrell, N. W., Botella, L. M., Bernabeu, et al
  2013; 93 (3): 530-537

- **Association of Twinning and Maternal Age with Major Structural Birth Defects in Utah, 1999 to 2008** *BIRTH DEFECTS RESEARCH PART A-CLINICAL AND MOLECULAR TERATOLOGY*
  Rider, R. A., Stevenson, D. A., Rinsky, J. E., Feldkamp, M. L.
  2013; 97 (8): 554-563

- **The generalized bone phenotype in children with neurofibromatosis 1: A sibling matched case-control study** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Armstrong, L., Jett, K., Birch, P., Kendler, D. L., McKay, H., Tsang, E., Stevenson, D. A., Hanley, D. A., Egeli, D., Burrows, M., Friedman, J. M.
  2013; 161A (7): 1654-1661

- **Skeletal abnormalities in lysosomal storage diseases.** *Pediatric endocrinology reviews : PER*
  Stevenson, D. A., Steiner, R. D.
  2013; 10: 406-416

- **Fractures in Children With Neurofibromatosis Type 1 From Two NF Clinics** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  George-Abraham, J. K., Martin, L. J., Kalkwarf, H. J., Rieley, M. B., Stevenson, D. A., Viskochil, D. H., Hopkin, R. J., Stevens, A. M., Hanson, H., Schorry, E. K.
  2013; 161A (5): 921-926

- **Approaches to Treating NF1 Tibial Pseudarthrosis: Consensus From the Children's Tumor Foundation NF1 Bone Abnormalities Consortium** *JOURNAL OF PEDIATRIC ORTHOPAEDICS*
  Stevenson, D. A., Little, D., Armstrong, L., Crawford, A. H., Eastwood, D., Friedman, J. M., Greggi, T., Gutierrez, G., Hunter-Schaedle, K., Kendler, D. L., Kolanczyk, M., Monsell, F., Oetgen, et al
  2013; 33 (3): 269-275

- **Copy Number Variation Analysis in 98 Individuals with PHACE Syndrome** *JOURNAL OF INVESTIGATIVE DERMATOLOGY*
  Siegel, D. H., Shieh, J. T., Kwon, E., Baselga, E., Blei, F., Cordisco, M., Dobyns, W. B., Duffy, K. J., Garzon, M. C., Gibbs, D. L., Grimmer, J. F., Hayflick, S. J., Krol, et al
  2013; 133 (3): 677-684

- **A cost savings approach to SPRED1 mutational analysis in individuals at risk for neurofibromatosis type 1** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Muram, T. M., Stevenson, D. A., Watts-Justice, S., Viskochil, D. H., Carey, J. C., Mao, R., Jackson, B.
  2013; 161A (3): 467-472

- **The genetics of vascular anomalies** *CURRENT OPINION IN OTOLARYNGOLOGY & HEAD AND NECK SURGERY*
  Frigerio, A., Stevenson, D. A., Grimmer, J. F.
  2012; 20 (6): 527-532

- **Peripheral muscle weakness in RASopathies** *MUSCLE & NERVE*
  Stevenson, D. A., Allen, S., Tidyman, W. E., Carey, J. C., Viskochil, D. H., Stevens, A., Hanson, H., Sheng, X., Thompson, B. A., Okumura, M. J., Reinker, K., Johnson, B., Rauen, et al

2012; 46 (3): 394-399

- **Total Hip Arthroplasty, Hip Osteoarthritis, Total Knee Arthroplasty, and Knee Osteoarthritis in Patients With Developmental Dysplasia of the Hip and Their Family Members: A Kinship Analysis Report** *JOURNAL OF PEDIATRIC ORTHOPAEDICS*
  Schiffern, A. N., Stevenson, D. A., Carroll, K. L., Pimentel, R., Mineau, G., Viskochil, D. H., Roach, J. W.
  2012; 32 (6): 609-612

- **Effects of a Plyometric Training Program for 3 Children With Neurofibromatosis Type 1** *PEDIATRIC PHYSICAL THERAPY*
  Johnson, B. A., Salzberg, C. L., Stevenson, D. A.
  2012; 24 (2): 199-208

- **Mosaicism in Stickler syndrome** *EUROPEAN JOURNAL OF MEDICAL GENETICS*
  Stevenson, D. A., Vanzo, R., Damjanovich, K., Hanson, H., Muntz, H., Hoffman, R. O., Bayrak-Toydemir, P.
  2012; 55 (6-7): 418-422

- **Candidate locus analysis for PHACE syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Mitchell, S., Siegel, D. H., Shieh, J. T., Stevenson, D. A., Grimmer, J. F., Lewis, T., Metry, D., Frieden, I., Blei, F., Kayserili, H., Drolet, B. A., Bayrak-Toydemir, P.
  2012; 158A (6): 1363-1367

- **Spinal arteriovenous fistulas in children with hereditary hemorrhagic telangiectasia** *JOURNAL OF NEUROSURGERY-PEDIATRICS*
  Calhoun, A. R., Bollo, R. J., Garber, S. T., McDonald, J., Stevenson, D. A., Hung, I. H., Brockmeyer, D. L., Walker, M. L.
  2012; 9 (6): 654-659

- **Gastrointestinal Bleeding in Infantile Hemangioma: A Complication of Segmental, Rather than Multifocal, Infantile Hemangiomas** *JOURNAL OF PEDIATRICS*
  Drolet, B. A., Pope, E., Juern, A. M., Sato, T., Howell, B., Puttgen, K. B., Lara-Corrales, I., Gilliam, A., Mancini, A., Powell, J., Siegel, D., Metry, D., Stevenson, et al
  2012; 160 (6): 1021-?

- **Analysis of skeletal dysplasias in the Utah population** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Carey, J. C., Byrne, J. L., Srisukhumbowornchai, S., Feldkamp, M. L.
  2012; 158A (5): 1046-1054

- **Letter to the Editor: Long-Term Experience with Duodenal Switch in Adolescents** *OBESITY SURGERY*
  Scheimann, A. O., Butler, M. G., Miller, J. L., Lee, P. D., Stevenson, D. A., Heinemann, J., Driscoll, D. J.
  2012; 22 (3): 517-518

- **Microdeletion 9q22.3 syndrome includes metopic craniosynostosis, hydrocephalus, macrosomia, and developmental delay** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Muller, E. A., Aradhya, S., Atkin, J. F., Carmany, E. P., Elliott, A. M., Chudley, A. E., Clark, R. D., Everman, D. B., Garner, S., Hall, B. D., Herman, G. E., Kivuva, E., Ramanathan, et al
  2012; 158A (2): 391-399

- **Hemangioma Is Associated with Atopic Disease** *OTOLARYNGOLOGY-HEAD AND NECK SURGERY*
  Grimmer, J. F., Williams, M. S., Pimentel, R., Mineau, G., Wood, G. M., Bayrak-Toydemir, P., Stevenson, D. A.
  2012; 146 (2): 206-209

- **RASA1 analysis: Clinical and molecular findings in a series of consecutive cases** *EUROPEAN JOURNAL OF MEDICAL GENETICS*
  Wooderchak-Donahue, W., Stevenson, D. A., McDonald, J., Grimmer, J. F., Gedge, F., Bayrak-Toydemir, P.
  2012; 55 (2): 91-95

- **Lower extremity strength and hopping and jumping ground reaction forces in children with neurofibromatosis type 1** *HUMAN MOVEMENT SCIENCE*
  Johnson, B. A., MacWilliams, B., Carey, J. C., Viskochil, D. H., D'Astous, J. L., Stevenson, D. A.
  2012; 31 (1): 247-254

- **5 ' UTR mutations of ENG cause hereditary hemorrhagic telangiectasia** *ORPHANET JOURNAL OF RARE DISEASES*
  Damjanovich, K., Langa, C., Blanco, F. J., McDonald, J., Botella, L. M., Bernabeu, C., Wooderchak-Donahue, W., Stevenson, D. A., Bayrak-Toydemir, P.
  2011; 6

- **Bone resorption in syndromes of the Ras/MAPK pathway** *CLINICAL GENETICS*

Stevenson, D. A., Schwarz, E. L., Carey, J. C., Viskochil, D. H., Hanson, H., Bauer, S., Weng, H. C., Greene, T., Reinker, K., Swensen, J., Chan, R. J., Yang, F., Senbanjo, et al
2011; 80 (6): 566-573

- **Mice lacking Nf1 in osteochondroprogenitor cells display skeletal dysplasia similar to patients with neurofibromatosis type I** *HUMAN MOLECULAR GENETICS*
  Wang, W., Nyman, J. S., Ono, K., Stevenson, D. A., Yang, X., Elefteriou, F.
  2011; 20 (20): 3910-3924

- **Molecular Confirmation of HRAS p.G12S in Siblings With Costello Syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Gripp, K. W., Stabley, D. L., Geller, P. L., Hopkins, E., Stevenson, D. A., Carey, J. C., Sol-Church, K.
  2011; 155A (9): 2263-2268

- **A SYSTEMATIC REVIEW: PLYOMETRIC TRAINING PROGRAMS FOR YOUNG CHILDREN** *JOURNAL OF STRENGTH AND CONDITIONING RESEARCH*
  Johnson, B. A., Salzberg, C. L., Stevenson, D. A.
  2011; 25 (9): 2623-2633

- **Familial Clustering of Hemangiomas** *ARCHIVES OF OTOLARYNGOLOGY-HEAD & NECK SURGERY*
  Grimmer, J. F., Williams, M. S., Pimentel, R., Mineau, G., Wood, G. M., Bayrak-Toydemir, P., Stevenson, D. A.
  2011; 137 (8): 757-760

- **Neurofibromin (Nf1) is required for skeletal muscle development** *HUMAN MOLECULAR GENETICS*
  Kossler, N., Stricker, S., Roedelsperger, C., Robinson, P. N., Kim, J., Dietrich, C., Osswald, M., Kuehnisch, J., Stevenson, D. A., Braun, T., Mundlos, S., Kolanczyk, M.
  2011; 20 (14): 2697-2709

- **Orthopaedic Conditions in Ras/MAPK Related Disorders** *JOURNAL OF PEDIATRIC ORTHOPAEDICS*
  Reinker, K. A., Stevenson, D. A., Tsung, A.
  2011; 31 (5): 599-605

- **The Musculoskeletal Phenotype of the RASopathies** *AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS*
  Stevenson, D. A., Yang, F.
  2011; 157C (2): 90-103

- **Multiple Increased Osteoclast Functions in Individuals with Neurofibromatosis Type 1** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Yan, J., He, Y., Li, H., Liu, Y., Zhang, Q., Jing, Y., Guo, Z., Zhang, W., Yang, D., Wu, X., Hanson, H., Li, et al
  2011; 155A (5): 1050-1059

- **Molecular diagnosis in hereditary hemorrhagic telangiectasia: findings in a series tested simultaneously by sequencing and deletion/duplication analysis** *CLINICAL GENETICS*
  McDonald, J., Damjanovich, K., Millson, A., Wooderchak, W., Chibuk, J. M., Stevenson, D. A., Gedge, F., Bayrak-Toydemir, P.
  2011; 79 (4): 335-344

- **Pediatric 25-hydroxyvitamin D concentrations in neurofibromatosis type 1** *JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM*
  Stevenson, D. A., Viskochil, D. H., Carey, J. C., Sheng, X., Murray, M., Moyer-Mileur, L., Shelton, J., Roberts, W. L., Bunker, A. M., Hanson, H., Bauer, S., D'Astous, J. L.
  2011; 24 (3-4): 169-174

- **Variable Expression of Neurofibromatosis 1 in Monozygotic Twins** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Rieley, M. B., Stevenson, D. A., Viskochil, D. H., Tinkle, B. T., Martin, L. J., Schorry, E. K.
  2011; 155A (3): 478-485

- **The role of motor proficiency in bone health in genetic syndromes** *DEVELOPMENTAL MEDICINE AND CHILD NEUROLOGY*
  Stevenson, D. A.
  2011; 53 (2): 103-104

- **SPRED1 Mutations in a Neurofibromatosis Clinic** *JOURNAL OF CHILD NEUROLOGY*
  Muram-Zborovski, T. M., Stevenson, D. A., Viskochil, D. H., Dries, D. C., Wilson, A. R., Mao, R.
  2010; 25 (10): 1203-1209

- **NF1 Exon 22 Analysis of Individuals With the Clinical Diagnosis of Neurofibromatosis Type 1** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Muram-Zborovski, T. M., Vaughn, C. P., Viskochil, D. H., Hanson, H., Mao, R., Stevenson, D. A.
  2010; 152A (8): 1973-1978

- **Parental Attitudes, Beliefs, and Perceptions about Genetic Testing for FAP and Colorectal Cancer Surveillance in Minors** *JOURNAL OF GENETIC COUNSELING*
  Levine, F. R., Coxworth, J. E., Stevenson, D. A., Tuohy, T., Burt, R. W., Kinney, A. Y.
  2010; 19 (3): 269-279

- **Speech-Language Characteristics of Children With Neurofibromatosis Type 1** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Thompson, H. L., Viskochil, D. H., Stevenson, D. A., Chapman, K. L.
  2010; 152A (2): 284-290

- **What's new in neurofibromatosis? Proceedings from the 2009 NF Conference: new frontiers.** *American journal of medical genetics. Part A*
  Kissil, J. L., Blakeley, J. O., Ferner, R. E., Huson, S. M., Kalamarides, M., Mautner, V., McCormick, F., Morrison, H., Packer, R., Ramesh, V., Ratner, N., Rauen, K. A., Stevenson, et al
  2010; 152A (2): 269-283

- **Motor Proficiency in Children With Neurofibromatosis Type 1** *PEDIATRIC PHYSICAL THERAPY*
  Johnson, B. A., MacWilliams, B. A., Carey, J. C., Viskochil, D. H., D'Astous, J. L., Stevenson, D. A.
  2010; 22 (4): 344-348

- **Proceedings from the 2009 genetic syndromes of the Ras/MAPK pathway: From bedside to bench and back.** *American journal of medical genetics. Part A*
  Rauen, K. A., Schoyer, L., McCormick, F., Lin, A. E., Allanson, J. E., Stevenson, D. A., Gripp, K. W., Neri, G., Carey, J. C., Legius, E., Tartaglia, M., Schubbert, S., Roberts, et al
  2010; 152A (1): 4-24

- **Pigmentary Findings in Neurofibromatosis Type 1-like Syndrome (Legius Syndrome) Potential Diagnostic Dilemmas** *JAMA-JOURNAL OF THE AMERICAN MEDICAL ASSOCIATION*
  Stevenson, D., Viskochil, D.
  2009; 302 (19): 2150-2151

- **Skeletal Abnormalities in Neurofibromatosis Type 1: Approaches to Therapeutic Options** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Elefteriou, F., Kolanczyk, M., Schindeler, A., Viskochil, D. H., Hock, J. M., Schorry, E. K., Crawford, A. H., Friedman, J. M., Little, D., Peltonen, J., Carey, J. C., Feldman, D., Yu, et al
  2009; 149A (10): 2327-2338

- **Health-Related Quality of Life Measures in Genetic Disorders: An Outcome Variable for Consideration in Clinical Trials** *AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS*
  Stevenson, D. A., Carey, J. C.
  2009; 151C (3): 255-260

- **Familial Predisposition to Developmental Dysplasia of the Hip** *JOURNAL OF PEDIATRIC ORTHOPAEDICS*
  Stevenson, D. A., Mineau, G., Kerber, R. A., Viskochil, D. H., Schaefer, C., Roach, J. W.
  2009; 29 (5): 463-466

- **Analysis of Radiographic Characteristics of Anterolateral Bowing of the Leg Before Fracture in Neurofibromatosis Type 1** *JOURNAL OF PEDIATRIC ORTHOPAEDICS*
  Stevenson, D. A., Carey, J. C., Viskochil, D. H., Moyer-Mileur, L. J., Slater, H., Murray, M. A., D'Astous, J. L., Murray, K. A.
  2009; 29 (4): 385-392

- **Tibial geometry in individuals with neurofibromatosis type 1 without anterolateral bowing of the lower leg using peripheral quantitative computed tomography** *BONE*
  Stevenson, D. A., Viskochil, D. H., Carey, J. C., Slater, H., Murray, M., Sheng, X., D'Astous, J., Hanson, H., Schorry, E., Moyer-Mileur, L. J.
  2009; 44 (4): 585-589

- **Bone Mineral Density in Children With Neurofibromatosis Type 1** *JOURNAL OF PEDIATRIC ORTHOPAEDICS*
  Stevenson, D. A., Murray, M., Viskochil, D. H., Carey, J. C., Moyer-Mileur, L. J.
  2008; 28 (7): 791-791

- **Autosomal recessive hypophosphatasia manifesting in utero with long bone deformity but showing spontaneous postnatal improvement** *JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM*
  Stevenson, D. A., Carey, J. C., Coburn, S. P., Ericson, K. L., Byrne, J. L., Mumm, S., Whyte, M. P.
  2008; 93 (9): 3443-3448

- **Evidence of increased bone resorption in neurofibromatosis type 1 using urinary pyridinium crosslink analysis** *PEDIATRIC RESEARCH*
  Stevenson, D. A., Schwarz, E. L., Viskochil, D. H., Moyer-Mileur, L. J., Murray, M., Firth, S. D., D'Astous, J. L., Carey, J. C., Pasquali, M.
  2008; 63 (6): 697-701

- **Brachymesomelic dysplasia with Peters anomaly of the eye results from disruptions of the X chromosome near the SHOX and SOX3 genes** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Bleyl, S. B., Byrne, J. L., South, S. T., Dries, D. C., Stevenson, D. A., Rope, A. F., Vianna-Morgante, A. M., Schoenwolf, G. C., Kivlin, J. D., Brothman, A., Carey, J. C.
  2007; 143A (23): 2785-2795

- **A novel multiple congenital anomaly-mental retardation syndrome with Pierre Robin sequence and cerebellar hypoplasia in two sisters** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Carey, J. C.
  2007; 143A (19): 2221-2226

- **Neurofibromatosis type 1 is a genetic skeletal disorder** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Viskochil, D. H., Carey, J. C.
  2007; 143A (17): 2082-2083

- **Gastric rupture and necrosis in Prader-Willi syndrome** *JOURNAL OF PEDIATRIC GASTROENTEROLOGY AND NUTRITION*
  Stevenson, D. A., Heinemann, J., Angulo, M., Butler, M. G., Loker, J., Rupe, N., Kendell, P., Cassidy, S. B., Scheimann, A.
  2007; 45 (2): 272-274

- **The use of anterolateral bowing of the lower leg in the diagnostic criteria for neurofibromatosis type 1** *GENETICS IN MEDICINE*
  Stevenson, D. A., Viskochil, D. H., Schorry, E. K., Crawford, A. H., D'Astous, J., Murray, K. A., Friedman, J. M., Armstrong, L., Carey, J. C.
  2007; 9 (7): 409-412

- **Mandibulofacial dysostosis in a patient with a de novo 2;17 translocation that disrupts the HOXD gene cluster** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Bleyl, S. B., Maxwell, T., Brothman, A. R., South, S. T.
  2007; 143A (10): 1053-1059

- **Deaths due to choking in Prader-Willi syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Heinemann, J., Angulo, M., Butler, M. G., Loker, J., Rupe, N., Kendell, P., Clericuzio, C. L., Scheimann, A. O.
  2007; 143A (5): 484-487

- **Bone mineral density in children and adolescents with neurofibromatosis type 1** *JOURNAL OF PEDIATRICS*
  Stevenson, D. A., Moyer-Mileur, L. J., Murray, M., Slater, H., Sheng, X., Carey, J. C., Dube, B., Viskochil, D. H.
  2007; 150 (1): 83-88

- **An absence of cutaneous neurofibromas associated with a 3-bp inframe deletion in Exon 17 of the NF1 gene (c.2970-2972 delAAT): evidence of a clinically significant NF1 genotype-phenotype correlation** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Upadhyaya, M., Huson, S. M., Davies, M., Thomas, N., Chuzhanova, N., Giovannini, S., Evans, D. G., HOWARD, E., Kerr, B., Griffiths, S., Consoli, C., Side, L., Adams, et al
  2007; 80 (1): 140-151

- **A new distal arthrogryposis syndrome characterized by plantar flexion contractures** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Swoboda, K. J., Sanders, R. K., Bamshad, M.
  2006; 140A (24): 2797-2801

- **Double inactivation of NF1 in tibial pseudarthrosis** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Stevenson, D. A., Zhou, H., Ashrafi, S., Messiaen, L. M., Carey, J. C., D'Astous, J. L., Santora, S. D., Viskochil, D. H.
  2006; 79 (1): 143-148

- **Clinical characteristics and natural history of Freeman-Sheldon syndrome** *PEDIATRICS*
  Stevenson, D. A., Carey, J. C., Palumbos, J., Rutherford, A., Dolcourt, J., Bamshad, M. J.

2006; 117 (3): 754-762

- **Clinical and molecular aspects of an informative family with neurofibromatosis type 1 and Noonan phenotype** *CLINICAL GENETICS*
  Stevenson, D. A., Viskochil, D. H., Rope, A. F., Carey, J. C.
  2006; 69 (3): 246-253
- **Familial congenital non-immune hydrops, chylothorax, and pulmonary lymphangiectasia** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Pysher, T. J., Ward, R. M., Carey, J. C.
  2006; 140A (4): 368-372
- **Case-control study of the muscular compartments and osseous strength in neurofibromatosis type 1 using peripheral quantitative computed tomography.** *Journal of musculoskeletal & neuronal interactions*
  Stevenson, D. A., Moyer-Mileur, L. J., Carey, J. C., Quick, J. L., Hoff, C. J., Viskochil, D. H.
  2005; 5 (2): 145-149
- **Calibration of 6q subtelomere deletions to define genotype/pheno type correlations** *CLINICAL GENETICS*
  Eash, D., Waggoner, D., Chung, J., Stevenson, D., Martinc, C. L.
  2005; 67 (5): 396-403
- **4p Terminal deletion and 11p subtelomeric duplication detected by genomic microarray in a patient with Wolf-Hirschhorn syndrome and an atypical phenotype** *JOURNAL OF PEDIATRICS*
  Stevenson, D. A., Carey, J. C., Cowley, B. C., Bayrak-Toydemir, P., Mao, R., Brothman, A. R.
  2004; 145 (6): 840-842
- **Paternal uniparental disomy of chromosome 14: Confirmation of a clinically-recognizable phenotype** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Brothman, A. R., Chen, Z., Bayrak-Toydemir, P., Longo, N.
  2004; 130A (1): 88-91
- **Pseudoaminopterin syndrome and trisomy 9** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Low, J., KING, J., Opitz, J. M., Miller, M. E.
  2004; 128A (2): 217-218
- **Contribution of malformations and genetic disorders to mortality in a children's hospital** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Carey, J. C.
  2004; 126A (4): 393-397
- **6q subtelomeric deletion: is there a recognizable syndrome?** *CLINICAL DYSMORPHOLOGY*
  Stevenson, D. A., Brothman, A. R., Carey, J. C., Chen, Z., Dent, K. M., Bale, J. F., Longo, N.
  2004; 13 (2): 103-106
- **Unexpected death and critical illness in Prader-Willi syndrome: Report of ten individuals** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Stevenson, D. A., Anaya, T. M., Clayton-Smith, J., Hall, B. D., Van Allen, M., Zori, R. T., Zackai, E. H., Frank, G., Clericuzio, C. L.
  2004; 124A (2): 158-164
- **Advise or consent? Issues in genetic testing of adolescents.** *Adolescent medicine (Philadelphia, Pa.)*
  Stevenson, D. A., Strasburger, V. C.
  2002; 13 (2): 213-?
- **Hydranencephaly in an infant with vascular malformations** *AMERICAN JOURNAL OF MEDICAL GENETICS*
  Stevenson, D. A., Hart, B. L., Clericuzio, C. L.
  2001; 104 (4): 295-298
- **Descriptive analysis of tibial pseudarthrosis in patients with neurofibromatosis 1** *Western-Society-for-Pediatric-Research Meeting*
  Stevenson, D. A., Birch, P. H., Friedman, J. M., Viskochil, D. H., Balestrazzi, P., Boni, S., Buske, A., Korf, B. R., Niimura, M., Pivnick, E. K., Schorry, E. K., Short, M. P., Tenconi, et al
  WILEY-LISS.1999: 413–19