Stanford

David A. Stevenson, MD

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

Pediatrics - Medical Genetics

CLINICAL OFFICES

- **Pediatric Genetics**

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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. His clinical practice is in treating and diagnosing individuals with genetic conditions.

Dr. Stevenson sees all types of individuals with various genetic disorders. 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. 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.

CLINICAL FOCUS

- Clinical Genetics

ACADEMIC APPOINTMENTS

- Professor - Med Center Line, Pediatrics - Medical Genetics
- Member, Bio-X
- Member, Child Health Research Institute

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

- Board Certification: Clinical Genetics, American Board of Medical Genetics and Genomics (2005)
- Fellowship: University of Utah (2005) UT
- Residency: University of New Mexico NM
- Board Certification: Pediatrics, American Board of Pediatrics (2002)
- Internship: University of New Mexico (2000) NM
- 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

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.

Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Medical Genetics (Fellowship Program)

Publications

PUBLICATIONS

- **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

- **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

- **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

- **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

- **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

- **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.

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