Stanford

David Stevenson

Professor of Pediatrics (Genetics)

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.

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.

Dr. Stevenson is a section editor for "Genetics in Medicine", and a member of the ACMG Board of Directors.

CLINICAL FOCUS

- Clinical Genetics

ACADEMIC APPOINTMENTS

- Professor - University Medical 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

- Vice-Chair, APHMG Program Director SIG (2021 - present)
- ACMG Program Committee, American College of Medical Genetics and Genomics (2017 - 2021)
- ACMG Board of Directors, American College of Medical Genetics and Genomics (2021 - present)
- WSPR Council, Western Society of Pediatric Research (2011 - 2014)

PROFESSIONAL EDUCATION

- Residency: University of New Mexico School of Medicine (2002) 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, Recruiting

Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Medical Genetics (Fellowship Program)

Publications

PUBLICATIONS

- **Factors associated with the time to complete clinical exome sequencing in a pediatric patient population.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Lee, G., Yu, L., Suarez, C. J., Stevenson, D. A., Ling, A., Killer, L.
  2022

- **50 Years Ago in TheJournalofPediatrics: Advances in the Understanding of Prader-Willi syndrome.** *The Journal of pediatrics*
  Stafford, D. E., Stevenson, D. A.
  2022; 247: 154

- **Advances in the Understanding of Prader-Willi syndrome** *JOURNAL OF PEDIATRICS*
  Stafford, D. J., Stevenson, D. A.
  2022; 247: 154

- **MEK Inhibitors for Neurofibromatosis Type 1 Manifestations: Clinical Evidence and Consensus.** *Neuro-oncology*
  de Blank, P. M., Gross, A. M., Akshintala, S., Blakeley, J. O., Bollag, G., Cannon, A., Dombi, E., Fangusaro, J., Gelb, B. D., Hargrave, D., Kim, A., Klesse, L. J., Loh, et al
  2022

- **Evaluation of the impact of the 2021 revised Neurofibromatosis type 1 diagnostic criteria on time to diagnosis.** *American journal of medical genetics. Part A*
  Ho, W. Y., Farrelly, E., Stevenson, D. A.
  2022

- **Updated diagnostic criteria and nomenclature for neurofibromatosis type 2 and schwannomatosis: An international consensus recommendation.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Plotkin, S. R., Messiaen, L., Legius, E., Pancza, P., Avery, R. A., Blakeley, J. O., Babovic-Vuksanovic, D., Ferner, R., Fisher, M. J., Friedman, J. M., Giovannini, M., Gutmann, D. H., Hanemann, et al
  2022

- **A survey of program directors for combined pediatrics and medical genetics and genomics residency programs: Perspectives when evaluating applicants.** *American journal of medical genetics. Part A*
  Niehaus, A. D., Rassbach, C. E., Stevenson, D. A.
  2022

- **The seventh international RASopathies symposium: Pathways to a cure-expanding knowledge, enhancing research, and therapeutic discovery.** *American journal of medical genetics. Part A*
  Kontaridis, M. I., Roberts, A. E., Schill, L., Schoyer, L., Stronach, B., Andelfinger, G., Aoki, Y., Axelrad, M. E., Bakker, A., Bennett, A. M., Broniscer, A., Castel, P., Chang, et al
  2022

- **A survey of program directors for combined pediatrics-medical genetics and genomics residency programs: Perspectives when evaluating applicants**
  Niehaus, A., Rassbach, C., Stevenson, D.
  ELSEVIER SCIENCE INC.2022: S369-S370

- **Factors associated with the time to complete clinical exome sequencing in a pediatric patient population**
  Lee, G., Yu, L., Suarez, C., Stevenson, D., Ling, A., Derby, L.
  ELSEVIER SCIENCE INC.2022: S268-S269

- **Natural history of NF1 c.2970_2972del p.(Met992del): confirmation of a low risk of complications in a longitudinal study.** *European journal of human genetics : EJHG*
  Forde, C., Burkitt-Wright, E., Turnpenny, P. D., Haan, E., Ealing, J., Mansour, S., Holder, M., Lahiri, N., Dixit, A., Procter, A., Pacot, L., Vidaud, D., Capri, et al
  1800

- **Response to Hamosh etal.** *American journal of human genetics*
  Biesecker, L. G., Adam, M. P., Alkuraya, F. S., Amemiya, A. R., Bamshad, M. J., Beck, A. E., Bennett, J. T., Bird, L. M., Carey, J. C., Chung, B., Clark, R. D., Cox, T. C., Curry, et al
  2021; 108 (9): 1809-1810

- **Arteriovenous Malformations-Current Understanding of the Pathogenesis with Implications for Treatment.** *International journal of molecular sciences*
  Schimmel, K., Ali, M. K., Tan, S. Y., Teng, J., Do, H. M., Steinberg, G. K., Stevenson, D. A., Spiekerkoetter, E.
  2021; 22 (16)

- **REiNS: Reliability of Handheld Dynamometry to Measure Focal Muscle Weakness in Neurofibromatosis Types 1 and 2.** *Neurology*
  Akshintala, S., Khalil, N., Yohay, K., Muzikansky, A., Allen, J., Yaffe, A., Gross, A. M., Fisher, M. J., Blakeley, J. O., Oberlander, B., Pudel, M., Engelson, C., Obletz, et al
  2021

- **Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Legius, E., Messiaen, L., Wolkenstein, P., Pancza, P., Avery, R. A., Berman, Y., Blakeley, J., Babovic-Vuksanovic, D., Cunha, K. S., Ferner, R., Fisher, M. J., Friedman, J. M., Gutmann, et al
  2021

- **Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Evans, D. G., Messiaen, L. M., Foulkes, W. D., Irving, R. E., Murray, A. J., Perez-Becerril, C., Rivera, B., McDonald-McGinn, D. M., Stevenson, D. A., Smith, M. J.
  2021

- **Congenital polyvalvular disease expands the cardiac phenotype of the RASopathies.** *American journal of medical genetics. Part A*
  Matalon, D. R., Stevenson, D. A., Bhoj, E. J., Santani, A. B., Keena, B. n., Cohen, M. S., Lin, A. E., Sheppard, S. E., Zackai, E. H.
  2021

- **Missense substitutions at a conserved 14-3-3 binding site in HDAC4 cause a novel intellectual disability syndrome.** *HGG advances*
  Wakeling, E. n., McEntagart, M. n., Bruccoleri, M. n., Shaw-Smith, C. n., Stals, K. L., Wakeling, M. n., Barnicoat, A. n., Beesley, C. n., Hanson-Kahn, A. K., Kukolich, M. n., Stevenson, D. A., Campeau, P. M., Ellard, et al
  2021; 2 (1): 100015

- **A dyadic approach to the delineation of diagnostic entities in clinical genomics.** *American journal of human genetics*
  Biesecker, L. G., Adam, M. P., Alkuraya, F. S., Amemiya, A. R., Bamshad, M. J., Beck, A. E., Bennett, J. T., Bird, L. M., Carey, J. C., Chung, B. n., Clark, R. D., Cox, T. C., Curry, et al
  2021; 108 (1): 8–15

- **Missense variants in CTNNB1 can be associated with vitreoretinopathy-Seven new cases of CTNNB1-associated neurodevelopmental disorder including a previously unreported retinal phenotype.** *Molecular genetics & genomic medicine*
  Rossetti, L. Z., Bekheirnia, M. R., Lewis, A. M., Mefford, H. C., Golden-Grant, K., Tarczy-Hornoch, K., Briere, L. C., Sweetser, D. A., Walker, M. A., Kravets, E., Stevenson, D. A., Bruenner, G., Sebastian, et al
  2020: e1542

- **Are Some Randomized Clinical Trials Impossible?** *Journal of pediatric orthopedics*

Rios, J. J., Richards, B. S., Stevenson, D. A., Oberlander, B., Viskochil, D., Gross, A. M., Dombi, E., Widemann, B. C., Plotkin, S. R., May, C. J., Ullrich, N. J., Goldstein, R. Y., Jain, et al

2020

- **KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation.** *Brain : a journal of neurology*

  Cif, L. n., Demailly, D. n., Lin, J. P., Barwick, K. E., Sa, M. n., Abela, L. n., Malhotra, S. n., Chong, W. K., Steel, D. n., Sanchis-Juan, A. n., Ngoh, A. n., Trump, N. n., Meyer, et al

  2020

- **Stress and Coping in Caregivers of Children with RASopathies: Assessment of the Impact of Caregiver Conferences.** *Journal of pediatric genetics*

  Ganetsos, A. n., Farrelly, E. n., Magoulas, P. n., Stevenson, D. A.

  2020; 9 (4): 235–42

- **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

- **"Following Through": Addressing the Racial Inequality for Preterm Infants and Their Families** *Pediatric Research*
  Stevenson, D. K., Wong, R. J., Profit, J. J., Shaw, G. M., Wang, C. J., Lee, H. C.
  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.
  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.** *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): 1714-1719

- **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

- **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. n., Posey, J. E., Streff, H. E., Gripp, K. W., Blesson, A. n., Powell-Hamilton, N. n., Tusi, J. n., Stevenson, D. A., Farrelly, E. n., Hudgins, L. n., Yang, Y. n., Xia, et al
  2017; 9 (1): 73

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