Stanford



David Stevenson

Professor of Pediatrics (Genetics)

Pediatrics - Medical Genetics

CLINICAL OFFICE (PRIMARY)

- **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 also 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-Univ Med 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, Not Recruiting

Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Medical Genetics (Fellowship Program)

Publications

PUBLICATIONS

- **Rapid Genomic Testing: A Study of Institutional Utilization and Outcomes.** *American journal of medical genetics. Part A*
  Johns, E. B., Yu, L., Rodriguez-Gil, J. L., López-Rivera, J. A., Stevenson, D. A., Suarez, C. J.
  2026

- **SHOC2 Is a Novel Cause of Central Conducting Lymphatic Anomaly.** *American journal of medical genetics. Part A*
  Ding, J. C., Sempowski, B. A., Zerbib, L., Adamson, G. T., Insinna, C., Marshall, C. M., Sinha, S. P., Breckenridge, A. J., Amedume, R. R., Johnson, J. T., Krikorian, G. L., Dykzeul, N., Floyd, et al
  2026

- **A qualitative interview study of medical genetics and genomics residents' perspectives on race, ethnicity, and racism in clinical settings and their related gaps in knowledge: "It's just taboo. Nobody talks about it.".** *Journal of genetic counseling*
  Gomez-Stafford, J. L., Vaunado, J., Campion, M. W., Clark, C., Murray, B. C., Niehaus, A. D., Stevenson, D. A., Tise, C. G., Martschenko, D. O.
  2026; 35 (3): e70216

- **CBL Syndrome With Granular Cell Tumor and café au lait macules: Expansion of the Phenotype.** *American journal of medical genetics. Part A*
  Harrington, C. N., Kohl, E., Gilitwala, Z., Teng, J., Siegel, D. H., Charville, G. W., Steffner, R. J., Stevenson, D. A.
  2026

- **The 9th International RASopathies Symposium.** *American journal of medical genetics. Part A*
  Castel, P., Schoyer, L., Stronach, B., Bogdanova, R., Bennett, A. M., Blakeley, J., Bornhorst, M., Bowers, T., Brachmann, S. M., Burkitt-Wright, E., Chatfield, K., De Luca, A., El-Chammas, et al
  2026

- **Molecular Characterization of Vascular Anomalies Tissues Guides Clinical Diagnosis.** *Lymphatic research and biology*
  Henslee, S. B., Wooderchak-Donahue, W. L., Grimmer, J. F., Szankasi, P., Bolia, A., Frigerio, A., Stevenson, D. A., Flores Daboub, J. A., Bayrak-Toydemir, P.
  2026: 15578585251391569

- **Diazoxide Choline Extended-Release Tablets in Prader-Willi Syndrome: A Randomized, Double-Blind, Withdrawal Period Study.** *The Journal of clinical endocrinology and metabolism*
  Miller, J. L., Bridges, N., Felner, E. I., Salehi, P., Yanovski, J. A., Stevenson, D. A., Mejia-Corletto, J., Shaikh, M. G., Abuzzahab, J., Fleischman, A., Kimonis, V., Shoemaker, A. H., Holland, et al
  2026

- **Medical Genetics and Genomics Residency Programs: Trends in Applications, Match Rates, and Matriculation from 2015 to 2024.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Niehaus, A. D., Stevenson, D. A., Blitzer, M. G.
  2025: 101635

- **Langerhans Cell Histiocytosis in Cardiofaciocutaneous Syndrome.** *American journal of medical genetics. Part A*
  Keehan, L., Sleightholm, R., Marks, L. J., Stevenson, D. A., Niehaus, A. D.
  2025: e64294

- **EXPRESS: The American Federation for Medical Research Maintains Commitment to Decades of Support of Vaccine and Vaccination Research to Improve Public Health.** *Journal of investigative medicine : the official publication of the American Federation for Clinical Research*
  Wiley, Z., Mahapatra, S., Agarwal, G., Baer, S. L., Bukhari, S., Cervantes, J. L., Correa, R. R., Crosby, S., Das, S., De Leon, J., Dickinson, J. D., Dutcher, J. P., Fuchs, et al
  2025: 10815589251327504

- **KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation.** *ArXiv*
  Cif, L., Demailly, D., Lin, J. P., Barwick, K. E., Sa, M., Abela, L., Malhotra, S., Chong, W. K., Steel, D., Sanchis-Juan, A., Ngoh, A., Trump, N., Meyer, et al
  2025

- **Towards a new taxonomy of preterm birth** *Journal of Perinatology*
  Stevenson, D. K., Chang, A. L., Wong, R. J., Reiss, J. D., Gaudillière, B., Sylvester, K. G., Ling, X. B., Angst, M. S., Shaw, G. M., Katz, M., Aghaeepour, N., Marić , I.
  2024

- **Characterizing and Evaluating the Structures of Combined Pediatrics and Medical Genetics and Genomics Residency Programs.** *American journal of medical genetics. Part A*
  Niehaus, A. D., Stevenson, D. A.
  2024: e63916

- **Relaxation of Food Control Parameters Based on Improvements in the Food Safe Zone Questionnaire Occurs with Reduction of Hyperphagia in Clinical Trials of Diazoxide Choline Extended Release (DCCR) in Participants with Prader-Willi Syndrome**
  Bridges, N., Gevers, E., Yanovski, J., Salehi, P., Shoemaker, A., Abuzzahab, J., Obrynba, K., Fleischman, A., Stevenson, D., Wilding, J., Mathew, V., Viskochil, D., Felner, et al
  KARGER.2024: 54

- **Long-term Efficacy Results of Diazoxide Choline Extended-Release (DCCR) Tablets in Participants with Prader-Willi Syndrome from the Completed C601 (DESTINY PWS) and C602 Open Label Extension (OLE) Studies**
  Gevers, E., Bridges, N., Yanovski, J., Felner, E., Salehi, P., Shoemaker, A., Fleischman, A., Goldstone, A., Angulo, M., Stevenson, D., Obrynba, K., Shaikh, M., Holland, et al
  KARGER.2024: 72-73

- **LONG-TERM SAFETY OF DIAZOXIDE CHOLINE EXTENDED-RELEASE (DCCR) TABLETS IN PARTICIPANTS WITH PRADER-WILLI SYNDROME FROM THE COMPLETED C601 RANDOMIZED DOUBLE-BLIND PLACEBO-CONTROLLED STUDY (DESTINY PWS) AND C602 OPEN LABEL EXTENSION (OLE) STUDY**
  Gevers, E. F., Miller, J., Bird, L., Angulo, M., Yanovski, J., Salehi, P., Shoemaker, A., Butler, M. G., Obrynba, K., Stevenson, D. A., Kimonis, V., Wilding, J., Shaikh, et al
  KARGER.2024: 155-157

- **Risk of meningomyelocele mediated by the common 22q11.2 deletion.** *Science (New York, N.Y.)*
  Vong, K. I., Lee, S., Au, K. S., Crowley, T. B., Capra, V., Martino, J., Haller, M., Araújo, C., Machado, H. R., George, R., Gerding, B., James, K. N., Stanley, et al
  2024; 384 (6695): 584-590

- **RASopathies influences on neuroanatomical variation in children.** *Biological psychiatry. Cognitive neuroscience and neuroimaging*
  McGhee, C. A., Honari, H., Siqueiros-Sanchez, M., Serur, Y., van Staalduinen, E. K., Stevenson, D., Bruno, J. L., Raman, M. M., Green, T.
  2024

- **Immunological and hematological findings as major features in a patient with a new germline pathogenic CBL variant.** *American journal of medical genetics. Part A*
  Stellacci, E., Carter, J. N., Pannone, L., Stevenson, D., Moslehi, D., Venanzi, S., Bernstein, J. A., Tartaglia, M., Martinelli, S.
  2024: e63627

- **Osteogenesis imperfecta type XVII: expansion of the phenotype** *EGYPTIAN JOURNAL OF MEDICAL HUMAN GENETICS*
  Dunleavy, B. M., Schildt, A. J., Harrington, C., Stevenson, D. A.
  2024; 25 (1)

- **Time to diagnosis in rapid exome/genome sequencing in the clinical inpatient setting.** *American journal of medical genetics. Part A*
  Schildt, A., Stevenson, D. A., Yu, L., Anguiano, B., Suarez, C. J.
  2023

- **Diazoxide choline extended-release tablet in people with Prader-Willi syndrome: results from long-term open-label study.** *Obesity (Silver Spring, Md.)*
  Miller, J. L., Gevers, E., Bridges, N., Yanovski, J. A., Salehi, P., Obrynba, K. S., Felner, E. I., Bird, L. M., Shoemaker, A. H., Angulo, M., Butler, M. G., Stevenson, D., Goldstone, et al
  2023

- **Two epilepsy-associated variants in KCNA2 (KV1.2) at position H310 oppositely affect channel functional expression.** *The Journal of physiology*
  Minguez-Vinas, T., Prakash, V., Wang, K., Lindstrom, S. H., Pozzi, S., Scott, S. A., Spiteri, E., Stevenson, D. A., Ashley, E. A., Gunnarsson, C., Pantazis, A.
  2023

- **A core outcome domain set to assess cutaneous neurofibromas related to neurofibromatosis type 1 in clinical trials.** *The British journal of dermatology*
  Fertitta, L., Bergqvist, C., Sarin, K., Plotkin, S. R., Moertel, C., Petersen, A. J., Cannon, A., Berman, Y., Pichard, D. C., Röhl, C., Lessing, A., Brizion, B., Peiffer, et al
  2023

- **Potential endpoints for assessment of bone health in persons with neurofibromatosis type 1.** *Clinical trials (London, England)*
  Gross, A. M., Plotkin, S. R., Watts, N. B., Fisher, M. J., Klesse, L. J., Lessing, A. J., McManus, M. L., Larson, A. N., Oberlander, B., Rios, J. J., Sarnoff, H., Simpson, B. N., Ullrich, et al
  2023: 17407745231201338

- **Monozygotic twins discordant for a congenital cranial dysinnervation disorder with features of Moebius syndrome.** *American journal of medical genetics. Part A*
  Gates, R. W., Webb, B. D., Stevenson, D. A., Jabs, E. W., DeFilippo, C., Ruzhnikov, M. R., Tise, C. G.
  2023

- **Jansen-de Vries syndrome: Expansion of the PPM1D clinical and phenotypic spectrum in 34 families.** *American journal of medical genetics. Part A*
  Wojcik, M. H., Srivastava, S., Agrawal, P. B., Balci, T. B., Callewaert, B., Calvo, P. L., Carli, D., Caudle, M., Colaiacovo, S., Cross, L., Demetriou, K., Drazba, K., Dutra-Clarke, et al
  2023

- **Frequency of Epistaxis and Telangiectasia in patients with Hereditary Hemorrhagic Telangiectasia (HHT) in comparison with the General Population: Curaçao Diagnostic Criteria Revisited.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  McDonald, J., Kornish, J., Stevenson, D. A., Hanson-Kahn, A., Balch, H., James, J., Naik, H., Whitehead, K. J.
  2023: 100865

- **Papillomas of Costello syndrome are not associated with human papillomavirus (HPV) infection in a small case series.** *Journal of the American Academy of Dermatology*
  Olsen, G. M., Johnson, L., Castel, P., Stevenson, D. A., White, K., Chiu, Y. E., Krol, A., Siegel, D. H.
  2023

- **Single Versus Multigene Testing for Hereditary Hearing Loss: Use and Costs in a Commercially Insured Cohort.** *Otolaryngology--head and neck surgery : official journal of American Academy of Otolaryngology-Head and Neck Surgery*
  Moon, P. K., Qian, Z. J., Stevenson, D. A., Chang, K. W.
  2023

- **Single Versus Multigene Testing for Hereditary Hearing Loss: Use and Costs in a Commercially Insured Cohort** *OTOLARYNGOLOGY-HEAD AND NECK SURGERY*
  Moon, P. K. K., Qian, Z., Stevenson, D. A. A., Chang, K. W. W.
  2023

- **Diazoxide Choline Extended-Release Tablet in People with Prader-Willi Syndrome: A Double-Blind, Placebo-Controlled Trial.** *The Journal of clinical endocrinology and metabolism*
  Miller, J. L., Gevers, E., Bridges, N., Yanovski, J. A., Salehi, P., Obrynba, K. S., Felner, E. I., Bird, L. M., Shoemaker, A. H., Angulo, M., Butler, M. G., Stevenson, D., Abuzzahab, et al
  2023

- **Bone health in RASopathies.** *American journal of medical genetics. Part C, Seminars in medical genetics*
  Stevenson, D. A., Viscogliosi, G., Leoni, C.

2022

- **Real world external validation of metabolic gestational age assessment in Kenya** *Plos Global Health*
  Hawken, S., Ward, V., Bota, A. B., Lamoureux, M., Ducharme, R., Wilson, L., Otieno, N., Munga, S., Nyawanda, B., Atito, R., Stevenson, D. K., Chakraborty, P., Darmstadt, et al
  2022; 2 (11): e0000652

- **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. E. 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. R. 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*
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