Stanford

Louanne Hudgins

Professor of Pediatrics (Genetics) at the Lucile Salter Packard Children's Hospital and, by courtesy, of Obstetrics and Gynecology at the Stanford University Medical Center

Pediatrics - Medical Genetics

CLINICAL OFFICES

- **Medical Genetics**

  730 Welch Rd

  2nd Fl

  Palo Alto, CA 94304

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

Bio

CLINICAL FOCUS

- Perinatal Genetics
- Clinical Genetics and Genomics

ACADEMIC APPOINTMENTS

- Professor - Med Center Line, Pediatrics - Medical Genetics
- Professor - Med Center Line (By courtesy), Obstetrics & Gynecology
- Member, Maternal & Child Health Research Institute (MCHRI)

HONORS AND AWARDS

- Mosbacher Family Distinguished Packard Fellow, Stanford University School of Medicine, Department of Pediatrics (2008-present)

PROFESSIONAL EDUCATION

- Board Certification: Clinical Genetics and Genomics, American Board of Medical Genetics and Genomics (2020)
- Fellowship: University of Connecticut School of Medicine Registrar (1990) CT
- Residency: University of Connecticut School of Medicine Registrar (1987) CT
- Internship: University of Connecticut School of Medicine Registrar (1985) CT
- Medical Education: University of Kansas School of Medicine (1984) KS

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

I am interested in prenatal genetic screening and diagnosis.

Teaching

COURSES

2019-20

- A Case Based Approach to Clinical Genetics: CHPR 274B, GENE 274B (Spr)
- Introduction to Medical Genetics: CHPR 272, GENE 272 (Aut)

2018-19

- A Case Based Approach to Clinical Genetics: CHPR 274B, GENE 274B (Spr)
- Introduction to Medical Genetics: CHPR 272, GENE 272 (Aut)

2017-18

- A Case Based Approach to Clinical Genetics: CHPR 274B, GENE 274B (Spr)
- Introduction to Medical Genetics: CHPR 272, GENE 272 (Aut)

2016-17

- A Case Based Approach to Clinical Genetics: CHPR 274B, GENE 274B (Spr)

Publications

PUBLICATIONS

- **DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract.** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Blackburn, A. T., Bekheirnia, N., Uma, V. C., Corkins, M. E., Xu, Y., Rosenfeld, J. A., Bainbridge, M. N., Yang, Y., Liu, P., Madan-Khetarpal, S., Delgado, M. R., Hudgins, L., Krantz, et al

  2019

- **Prenatally diagnosed omphalocele: characteristics associated with adverse neonatal outcomes.** *Journal of perinatology : official journal of the California Perinatal Association*

  Chock, V. Y., Davis, A. S., Cho, S., Bax, C., Fluharty, E., Weigel, N., Homeyer, M., Hudgins, L., Jones, R., Rubesova, E., Sylvester, K. G., Blumenfeld, Y. J., Hintz, et al

  2019

- **Developing a conceptual, reproducible, rubric-based approach to consent and result disclosure for genetic testing by clinicians with minimal genetics background** *GENETICS IN MEDICINE*

  Ormond, K. E., Hallquist, M. G., Buchanan, A. H., Dondanville, D., Cho, M. K., Smith, M., Roche, M., Brothers, K. B., Coughlin, C. R., Hercher, L., Hudgins, L., Jamal, S., Levy, et al

  2019; 21 (3): 727–35

- **Cornelia de Lange syndrome in diverse populations.** *American journal of medical genetics. Part A*

  Dowsett, L., Porras, A. R., Kruszka, P., Davis, B., Hu, T., Honey, E., Badoe, E., Thong, M., Leon, E., Girisha, K. M., Shukla, A., Nayak, S. S., Shotelersuk, et al

  2019

- **Correction: DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract.** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Blackburn, A. T., Bekheirnia, N., Uma, V. C., Corkins, M. E., Xu, Y., Rosenfeld, J. A., Bainbridge, M. N., Yang, Y., Liu, P., Madan-Khetarpal, S., Delgado, M. R., Hudgins, L., Krantz, et al

  2019

- **Clinical spectrum of individuals with pathogenic NF1 missense variants affecting p.Met1149, p.Arg1276 and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1.** *Human mutation*

  Koczkowska, M., Callens, T., Chen, Y., Gomes, A., Hicks, A. D., Sharp, A., Johns, E., Uhas, K. A., Armstrong, L., Bosanko, K. A., Babovic-Vuksanovic, D., Baker, L., Basel, et al

  2019

- **Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy.** *American journal of medical genetics. Part A*
  Carter, L. B., Battaglia, A., Cherry, A., Manning, M. A., Ruzhnikov, M. R., Bird, L. M., Dowsett, L., Graham, J. M., Alkuraya, F. S., Hashem, M., Dinulos, M. B., Vallee, S., Adam, et al
  2019

- **Annemarie Sommer memorial.** *American journal of medical genetics. Part A*
  Hudgins, L.
  2019

- **Autosomal recessive Stickler syndrome resulting from a COL9A3 mutation.** *American journal of medical genetics. Part A*
  Hanson-Kahn, A., Li, B., Cohn, D. H., Nickerson, D. A., Bamshad, M. J., University of Washington Center for Mendelian Genomics, Hudgins, L.
  2018

- **Developing a conceptual, reproducible, rubric-based approach to consent and result disclosure for genetic testing by clinicians with minimal genetics background.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Ormond, K. E., Hallquist, M. L., Buchanan, A. H., Dondanville, D., Cho, M. K., Smith, M., Roche, M., Brothers, K. B., Coughlin, C. R., Hercher, L., Hudgins, L., Jamal, S., Levy, et al
  2018

- **ERCC4 variants identified in a cohort of patients with segmental progeroid syndromes** *HUMAN MUTATION*
  Mori, T., Yousefzadeh, M. J., Faridounnia, M., Chong, J. X., Hisama, F. M., Hudgins, L., Mercado, G., Wade, E. A., Barghouthy, A. S., Lee, L., Martin, G. M., Nickerson, D. A., Bamshad, et al
  2018; 39 (2): 255–65

- **Noninvasive Prenatal Diagnosis of Single-Gene Disorders by Use of Droplet Digital PCR** *CLINICAL CHEMISTRY*
  Camunas-Soler, J., Lee, H., Hudgins, L., Hintz, S. R., Blumenfeld, Y. J., El-Sayed, Y. Y., Quake, S. R.
  2018; 64 (2): 336–45

- **Current controversies in prenatal diagnosis 2: Cell-free DNA prenatal screening should be used to identify all chromosome abnormalities** *PRENATAL DIAGNOSIS*
  Chitty, L. S., Hudgins, L., Norton, M. E.
  2018; 38 (3): 160–65

- **Prenatal treatment of ornithine transcarbamylase deficiency.** *Molecular genetics and metabolism*
  Wilnai, Y., Blumenfeld, Y. J., Cusmano, K., Hintz, S. R., Alcorn, D., Benitz, W. E., Berquist, W. E., Bernstein, J. A., Castillo, R. O., Concepcion, W., Cowan, T. M., Cox, K. L., Lyell, et al
  2018

- **Noninvasive Prenatal Diagnosis of Single-Gene Disorders by Use of Droplet Digital PCR.** *Clinical chemistry*
  Camunas-Soler, J., Lee, H., Hudgins, L., Hintz, S. R., Blumenfeld, Y. J., El-Sayed, Y. Y., Quake, S. R.
  2018; 64 (2): 336–45

- **Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Yuan, B., Neira, J., Pehlivan, D., Santiago-Sim, T., Song, X., Rosenfeld, J., Posey, J. E., Patel, V., Jin, W., Adam, M. P., Baple, E. L., Dean, J., Fong, et al
  2018

- **KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Kennedy, J., Goudie, D., Blair, E., Chandler, K., Joss, S., McKay, V., Green, A., Armstrong, R., Lees, M., Kamien, B., Hopper, B., Tan, T. Y., Yap, et al
  2018

- **De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features.** *American journal of human genetics*
  Tokita, M. J., Chen, C. A., Chitayat, D., Macnamara, E., Rosenfeld, J. A., Hanchard, N., Lewis, A. M., Brown, C. W., Marom, R., Shao, Y., Novacic, D., Wolfe, L., Wahl, et al
  2018

- **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

- **37th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2016 Annual Meeting.** *American journal of medical genetics. Part A*
  Keppler-Noreuil, K. M., Martinez-Agosto, J. A., Hudgins, L., Carey, J. C.
  2017: 2007–73
- **A novel missense variant in the GLI3 zinc finger domain in a family with digital anomalies.** *American journal of medical genetics. Part A*
  Crapster, J. A., Hudgins, L., Chen, J. K., Gomez-Ospina, N.
  2017
- **FOXP1-related intellectual disability syndrome: a recognisable entity.** *Journal of medical genetics*
  Meerschaut, I., Rochefort, D., Revençu, N., Pètre, J., Corsello, C., Rouleau, G. A., Hamdan, F. F., Michaud, J. L., Morton, J., Radley, J., Ragge, N., García-Miñaúr, S., Lapunzina, et al
  2017; 54 (9): 613–23
- **Germline Loss-of-Function Mutations in EPHB4 Cause a Second Form of Capillary Malformation-Arteriovenous Malformation (CM-AVM2) Deregulating RAS-MAPK Signaling.** *Circulation*
  Amyere, M., Revencu, N., Helaers, R., Pairet, E., Baselga, E., Cordisco, M. R., Chung, W. K., Dubois, J., Lacour, J. P., Martorell, L., Mazereeuw-Hautier, J., Pyeritz, R. E., Amor, et al
  2017
- **Recommendations for the integration of genomics into clinical practice** *GENETICS IN MEDICINE*
  Bowdin, S., Gilbert, A., Bedoukian, E., Carew, C., Adam, M. P., Belmont, J., Bernhardt, B., Biesecker, L., Bjornsson, H. T., Blitzer, M., D'Alessandro, L. C., Deardorff, M. A., Demmer, et al
  2016; 18 (11): 1075-1084
- **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
- **36th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2015 Annual Meeting** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Gripp, K. W., Adam, M. P., Hudgins, L., Carey, J. C.
  2016; 170 (7): 1665-1726
- **Prenatally Diagnosed Cases of Binder Phenotype Complicated by Respiratory Distress in the Immediate Postnatal Period.** *Journal of ultrasound in medicine*
  Blumenfeld, Y. J., Davis, A. S., Hintz, S. R., Milan, K., Messner, A. H., Barth, R. A., Hudgins, L., Chueh, J., Homeyer, M., Bernstein, J. A., Enns, G., Atwal, P., Manning, et al
  2016; 35 (6): 1353-1358
- **Novel X-linked syndrome of cardiac valvulopathy, keloid scarring, and reduced joint mobility due to filamin A substitution G1576R.** *American journal of medical genetics. Part A*
  Atwal, P. S., Blease, S., Braxton, A., Graves, J., He, W., Person, R., Slattery, L., Bernstein, J. A., Hudgins, L.
  2016; 170 (4): 891-895
- **Patient preferences for prenatal testing of microdeletion and microduplication syndromes** *PRENATAL DIAGNOSIS*
  Calonico, E., Blumenfeld, Y. J., Hudgins, L., Taylor, J.
  2016; 36 (3): 244-251
- **A Multifaceted Mentoring Program for Junior Faculty in Academic Pediatrics** *TEACHING AND LEARNING IN MEDICINE*
  Chen, M. M., Sandborg, C. I., Hudgins, L., Sanford, R., Bachrach, L. K.
  2016; 28 (3): 320-328
- **DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies** *EUROPEAN JOURNAL OF HUMAN GENETICS*
  Ji, J., Lee, H., Argiropoulos, B., Dorrani, N., Mann, J., Martinez-Agosto, J. A., Gomez-Ospina, N., Gallant, N., Bernstein, J. A., Hudgins, L., Slattery, L., Isidor, B., Le Caignec, et al
  2015; 23 (11): 1473-1481

- **Detection Rates for Aneuploidy by First-Trimester and Sequential Screening** *OBSTETRICS AND GYNECOLOGY*
  Baer, R. J., Flessel, M. C., Jelliffe-Pawlowski, L. L., Goldman, S., Hudgins, L., Hull, A. D., Norton, M. E., Currier, R. J.
  2015; 126 (4): 752-758

- **Detection Rates for Aneuploidy by First-Trimester and Sequential Screening.** *Obstetrics and gynecology*
  Baer, R. J., Flessel, M. C., Jelliffe-Pawlowski, L. L., Goldman, S., Hudgins, L., Hull, A. D., Norton, M. E., Currier, R. J.
  2015; 126 (4): 753-759

- **Knowledge, understanding, and uptake of noninvasive prenatal testing among Latina women** *PRENATAL DIAGNOSIS*
  Farrell, R., Hawkins, A., Barragan, D., Hudgins, L., Taylor, J.
  2015; 35 (8): 748-753

- **ClinGen - The Clinical Genome Resource** *NEW ENGLAND JOURNAL OF MEDICINE*
  Rehm, H. L., Berg, J. S., Brooks, L. D., Bustamante, C. D., Evans, J. P., Landrum, M. J., Ledbetter, D. H., Maglott, D. R., Martin, C. L., Nussbaum, R. L., Plon, S. E., Ramos, E. M., Sherry, et al
  2015; 372 (23): 2235-2242

- **Neonatal Pulmonary Arterial Hypertension and Noonan Syndrome: Two Fatal Cases with a Specific RAF1 Mutation** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Hopper, R. K., Feinstein, J. A., Manning, M. A., Benitz, W., Hudgins, L.
  2015; 167A (4): 882-885

- **De Novo Nonsense Mutations in KAT6A, a Lysine Acetyl-Transferase Gene, Cause a Syndrome Including Microcephaly and Global Developmental Delay.** *American journal of human genetics*
  Arboleda, V. A., Lee, H., Dorrani, N., Zadeh, N., Willis, M., Macmurdo, C. F., Manning, M. A., Kwan, A., Hudgins, L., Barthelemy, F., Miceli, M. C., Quintero-Rivera, F., Kantarci, et al
  2015; 96 (3): 498-506

- **Exome Sequencing for the Diagnosis of 46,XY Disorders of Sex Development.** *journal of clinical endocrinology and metabolism*
  Baxter, R. M., Arboleda, V. A., Lee, H., Barseghyan, H., Adam, M. P., Fechner, P. Y., Bargman, R., Keegan, C., Travers, S., Schelley, S., Hudgins, L., Mathew, R. P., Stalker, et al
  2015; 100 (2): E333-44

- **Increased body mass in infancy and early toddlerhood in Angelman syndrome patients with uniparental disomy and imprinting center defects.** *American journal of medical genetics. Part A*
  Brennan, M., Adam, M. P., Seaver, L. H., Myers, A., Schelley, S., Zadeh, N., Hudgins, L., Bernstein, J. A.
  2015; 167A (1): 142-146

- **Genomics in the clinic: ethical and policy challenges in clinical next-generation sequencing programs at early adopter USA institutions.** *Personalized medicine*
  Milner, L. C., Garrison, N. A., Cho, M. K., Altman, R. B., Hudgins, L., Galli, S. J., Lowe, H. J., Schrijver, I., Magnus, D. C.
  2015; 12 (3): 269–82

- **Increased body mass in infancy and early toddlerhood in Angelman syndrome patients with uniparental disomy and imprinting center defects.** *American journal of medical genetics. Part A*
  Brennan, M., Adam, M. P., Seaver, L. H., Myers, A., Schelley, S., Zadeh, N., Hudgins, L., Bernstein, J. A.
  2015; 167 (1): 142-146

- **Genomics in the clinic: ethical and policy challenges in clinical next-generation sequencing programs at early adopter USA institutions** *PERSONALIZED MEDICINE*
  Milner, L. C., Garrison, N. A., Cho, M. K., Altman, R. B., Hudgins, L., Galli, S. J., Lowe, H. J., Schrijver, I., Magnus, D. C.
  2015; 12 (3): 269-282

- **Patient preferences for prenatal testing of microdeletion and microduplication syndromes.** *Prenatal diagnosis*
  Calonico, E., Blumenfeld, Y. J., Hudgins, L., Taylor, J.
  2015

- **Perinatal Features of the RASopathies: Noonan Syndrome, Cardiofaciocutaneous Syndrome and Costello Syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Myers, A., Bernstein, J. A., Brennan, M., Curry, C., Esplin, E. D., Fisher, J., Homeyer, M., Manning, M. A., Muller, E. A., Niemi, A., Seaver, L. H., Hintz, S. R., Hudgins, et al

2014; 164A (11): 2814-2821

- **Attitudes of mothers of children with down syndrome towards noninvasive prenatal testing.** *Journal of genetic counseling*
  Kellogg, G., Slattery, L., Hudgins, L., Ormond, K.
  2014; 23 (5): 805-813

- **Clinical whole-exome sequencing: are we there yet?** *GENETICS IN MEDICINE*
  Atwal, P. S., Brennan, M., Cox, R., Niaki, M., Platt, J., Homeyer, M., Kwan, A., Parkin, S., Schelley, S., Slattery, L., Wilnai, Y., Bernstein, J. A., Enns, et al
  2014; 16 (9): 717-719

- **Nine patients with Xp22.31 microduplication, cognitive deficits, seizures, and talipes anomalies.** *American journal of medical genetics. Part A*
  Esplin, E. D., Li, B., Slavotinek, A., Novelli, A., Battaglia, A., Clark, R., Curry, C., Hudgins, L.
  2014; 164A (8): 2097-2103

- **Nine patients with Xp22.31 microduplication, cognitive deficits, seizures, and talipes anomalies.** *American journal of medical genetics. Part A*
  Esplin, E. D., Li, B., Slavotinek, A., Novelli, A., Battaglia, A., Clark, R., Curry, C., Hudgins, L.
  2014; 164 (8): 2097-2103

- **Noninvasive prenatal diagnosis in a fetus at risk for methylmalonic acidemia.** *Genetics in medicine*
  Gu, W., Koh, W., Blumenfeld, Y. J., El-Sayed, Y. Y., Hudgins, L., Hintz, S. R., Quake, S. R.
  2014; 16 (7): 564-567

- **Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance** *HUMAN MOLECULAR GENETICS*
  Kaiser, F. J., Ansari, M., Braunholz, D., Gil-Rodriguez, M. C., Decroos, C., Wilde, J. J., Fincher, C. T., Kaur, M., Bando, M., Amor, D. J., Atwal, P. S., Bahlo, M., Bowman, et al
  2014; 23 (11): 2888-2900

- **NIPT in a Clinical Setting: An analysis of Uptake in the First Months of Clinical Availability.** *Journal of genetic counseling*
  Taylor, J. B., Chock, V. Y., Hudgins, L.
  2014; 23 (1): 72-78

- **Expansion of the TARP syndrome phenotype associated with de novo mutations and mosaicism.** *American journal of medical genetics. Part A*
  Johnston, J. J., Sapp, J. C., Curry, C., Horton, M., Leon, E., Cusmano-Ozog, K., Dobyns, W. B., Hudgins, L., Zackai, E., Biesecker, L. G.
  2014; 164A (1): 120-128

- **Whole-Exome/Genome Sequencing and Genomics** *PEDIATRICS*
  Grody, W. W., Thompson, B. H., Hudgins, L.
  2013; 132: S211-S215

- **The Decision to Continue a Pregnancy Affected by Down Syndrome: Timing of Decision and Satisfaction with Receiving a Prenatal Diagnosis** *JOURNAL OF GENETIC COUNSELING*
  Hurford, E., Hawkins, A., Hudgins, L., Taylor, J.
  2013; 22 (5): 587-593

- **Best ethical practices for clinicians and laboratories in the provision of noninvasive prenatal testing.** *Prenatal diagnosis*
  Allyse, M. A., Sayres, L. C., Havard, M., King, J. S., Greely, H. T., Hudgins, L., Taylor, J., Norton, M. E., Cho, M. K., Magnus, D., Ormond, K. E.
  2013; 33 (7): 656-661

- **Expanding the Phenotype of Cardiovascular Malformations in Adams-Oliver Syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Algaze, C., Esplin, E. D., Lowenthal, A., Hudgins, L., Tacy, T. A., Tierney, E. S.
  2013; 161A (6): 1386-1389

- **Variables Influencing Pregnancy Termination Following Prenatal Diagnosis of Fetal Chromosome Abnormalities** *JOURNAL OF GENETIC COUNSELING*
  Hawkins, A., Stenzel, A., Taylor, J., Chock, V. Y., Hudgins, L.
  2013; 22 (2): 238-248

- **Mutations in FKBP10, which result in Bruck syndrome and recessive forms of osteogenesis imperfecta, inhibit the hydroxylation of telopeptide lysines in bone collagen** *HUMAN MOLECULAR GENETICS*
  Schwarze, U., Cundy, T., Pyott, S. M., Christiansen, H. E., Hegde, M. R., Bank, R. A., Pals, G., Ankala, A., Conneely, K., Seaver, L., Yandow, S. M., Raney, E., Babovic-Vuksanovic, et al

2013; 22 (1): 1-17

- **Uptake of cell free fetal DNA testing in women with positive serum screening** *33rd Annual Pregnancy Meeting of the Society-for-Maternal-Fetal-Medicine (SMFM)*
  Chetty, S., Taylor, J., Hudgins, L., Norton, M.
  MOSBY-ELSEVIER.2013: S256–S256
- **ARTHROGRYPOSIS, RENAL DYSFUNCTION AND CHOLESTASIS (ARC) SYNDROME: A NEW PATIENT CASE REPORT** *Western Regional Meeting of the American-Federation-for-Medical-Research*
  Brennan, M., SLATTERY, L., Esplin, E., Enns, G. M., Hudgins, L., Manning, M.
  LIPPINCOTT WILLIAMS & WILKINS.2013: 188–88
- **Expanding the phenotype of cardiovascular malformations in Adams-Oliver syndrome.** *American journal of medical genetics. Part A*
  Algaze, C., Esplin, E. D., Lowenthal, A., Hudgins, L., Tacy, T. A., Selamet Tierney, E. S.
  2013; 161 (6): 1386–89
- **Conservatively Managed Fetal Goiter: An Alternative to in utero Therapy.** *Fetal diagnosis and therapy*
  Blumenfeld, Y. J., Davis, A., Milan, K., Chueh, J., Hudgins, L., Barth, R. A., Hintz, S. R.
  2013; 34 (3): 184-187
- **Evidence that personal genome testing enhances student learning in a course on genomics and personalized medicine.** *PloS one*
  Salari, K., Karczewski, K. J., Hudgins, L., Ormond, K. E.
  2013; 8 (7)
- **Utilization of available prenatal screening and diagnosis: effects of the California screen program** *JOURNAL OF PERINATOLOGY*
  Blumenfeld, Y. J., Taylor, J., Lee, H. C., Hudgins, L., Sung, J. F., El-Sayed, Y. Y.
  2012; 32 (12): 907-912
- **Report of Two Patients and Further Characterization of Interstitial 9p13 Deletion-A Rare But Recurrent Microdeletion Syndrome?** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Niemi, A., Kwan, A., Hudgins, L., Cherry, A. M., Manning, M. A.
  2012; 158A (9): 2328-2335
- **Mutation risk associated with paternal and maternal age in a cohort of retinoblastoma survivors** *HUMAN GENETICS*
  Mills, M. B., Hudgins, L., Balise, R. R., Abramson, D. H., Kleinerman, R. A.
  2012; 131 (7): 1115-1122
- **Consanguinity and the risk of congenital heart disease** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Shieh, J. T., Bittles, A. H., Hudgins, L.
  2012; 158A (5): 1236-1241
- **What Is Your Diagnosis? The Diagnosis: Trichorhinophalangeal Syndrome Type I** *CUTIS*
  Snyder, J. R., Berk, D. R., Kwan, A., Hudgins, L., Bruckner, A. L.
  2012; 89 (2): 56-?
- **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
- **Noninvasive prenatal diagnosis: pregnant women's interest and expected uptake** *PRENATAL DIAGNOSIS*
  Tischler, R., Hudgins, L., Blumenfeld, Y. J., Greely, H. T., Ormond, K. E.
  2011; 31 (13): 1292-1299
- **Ectopia Lentis as the Presenting and Primary Feature in Marfan Syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Zadeh, N., Bernstein, J. A., Niemi, A. K., Dugan, S., Kwan, A., Liang, D., Hyland, J. C., Hoyme, H. E., Hudgins, L., Manning, M. A.
  2011; 155A (11): 2661-2668
- **Horseshoe Kidney and a Rare TSC2 Variant in Two Unrelated Individuals With Tuberous Sclerosis Complex** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*

Niemi, A., Northrup, H., Hudgins, L., Bernstein, J. A.
2011; 155A (10): 2534-2537

- **Familial Cardiac Valvulopathy Due to Filamin A Mutation** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Bernstein, J. A., Bernstein, D., Hehr, U., Hudgins, L.
  2011; 155A (9): 2236-2241

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