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Dena Matalon

Clinical Associate Professor, Pediatrics - Medical Genetics

CLINICAL OFFICE (PRIMARY)

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Bio

CLINICAL FOCUS

- Clinical Genetics

ACADEMIC APPOINTMENTS

- Clinical Associate Professor, Pediatrics - Medical Genetics
- Member, Maternal & Child Health Research Institute (MCHRI)

PROFESSIONAL EDUCATION

- Medical Education: Sidney Kimmel Medical College Thomas Jefferson University (2013) PA
- Board Certification: Pediatrics, American Board of Pediatrics (2018)
- Board Certification, American Board of Pediatrics , Pediatrics (2018)
- Board Certification: Clinical Genetics, American Board of Medical Genetics and Genomics (2017)
- Fellowship: Children's Hospital of Philadelphia (2017) PA
- Residency: Children's Hospital of Philadelphia (2016) PA
- Internship: Children's Hospital of Philadelphia (2014) PA

Publications

PUBLICATIONS

- **Characterization of the prenatal renal phenotype associated with 17q12, HNF1B, microdeletions.** *Prenatal diagnosis*

  Verscaj, C. P., Velez-Bartolomei, F., Bodle, E., Chan, K., Lyons, M. J., Thorson, W., Tan, W., Rodig, N., Graham, J. M., Peron, A., Quintero-Rivera, F., Zackai, E. H., Thomas, et al

  2023

- **Clinical, technical, and environmental biases influencing equitable access to clinical genetics/genomics testing: A points to consider statement of the American College of Medical Genetics and Genomics (ACMG).** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Matalon, D. R., Zepeda-Mendoza, C. J., Aarabi, M., Brown, K., Fullerton, S. M., Kaur, S., Quintero-Rivera, F., Vatta, M., ACMG Social, E. a.

  2023: 100812

- **Genomic sequencing in a cohort of individuals with fibular aplasia, tibial campomelia, and oligosyndactyly (FATCO) syndrome.** *American journal of medical genetics. Part A*
  Matalon, D. R., Bhoj, E. J., Li, D., McDougall, C., Schindewolf, E., Khalek, N., Wilkens, A., McManus, M., Deardorff, M. A., Zackai, E. H.
  2023

- **Creatine Transporter Deficiency Presenting as Failure to Thrive: A Case Report of a Novel SLC6A8 Variant Causing a Treatable but Likely Underdiagnosed Genetic Disorder.** *Journal of investigative medicine high impact case reports*
  Tise, C. G., Palma, M. J., Cusmano-Ozog, K. P., Matalon, D. R.
  2023; 11: 23247096231154438

- **DNA methylation episignature for Witteveen-Kolk syndrome due to SIN3A haploinsufficiency.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Coenen-van der Spek, J., Relator, R., Kerkhof, J., McConkey, H., Levy, M. A., Tedder, M. L., Louie, R. J., Fletcher, R. S., Moore, H. W., Childers, A., Farrelly, E. R., Champaigne, N. L., Lyons, et al
  2022

- **Short Bones, Renal Stones, and Diagnostic Moans: Hypercalcemia in a Girl Found to Have Coffin-Lowry Syndrome.** *Journal of investigative medicine high impact case reports*
  Tise, C. G., Matalon, D. R., Manning, M. A., Byers, H. M., Grover, M.
  2022; 10: 23247096221101844

- **Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Cuinat, S., Nizon, M., Isidor, B., Stegmann, A., van Jaarsveld, R. H., van Gassen, K. L., van der Smagt, J. J., Volker-Touw, C. M., Holwerda, S. J., Terhal, P. A., Schuhmann, S., Vasileiou, G., Khalifa, et al
  2022

- **Reuben Matalon, MD, PhD, FACMG (1935-2021).** *Human gene therapy*
  Matalon, D. R., Velagaleti, G., Ahmed, S. S., Gessler, D. J., Gao, G.
  2022

- **Leiomyomatosis in an Infant With a SUFU Splice Site Variant: Case Report.** *Journal of pediatric hematology/oncology*
  Rao, R. R., Dulken, B. W., Matalon, D. R., Borensztein, M., McGuinness, M., Cizek, S. M., Bruzoni, M., Tan, S. Y., Kreimer, S.
  2022

- **Points to consider to avoid unfair discrimination and the misuse of genetic information: A statement of the American College of Medical Genetics and Genomics (ACMG).** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Seaver, L. H., Khushf, G., King, N. M., Matalon, D. R., Sanghavi, K., Vatta, M., Wees, K., ACMG Social, E. a.
  2022; 24 (3): 512-520

- **Clinical and molecular characterization of five new individuals with WAC-related intellectual disability: Evidence of pathogenicity for a novel splicing variant.** *American journal of medical genetics. Part A*
  Morales, J. A., Valenzuela, I., Cusco, I., Cogne, B., Isidor, B., Matalon, D. R., Gomez-Ospina, N.
  1800

- **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

- **Diagnostic journey and impact of enzyme replacement therapy for mucopolysaccharidosis IVA: a sibling control study.** *Orphanet journal of rare diseases*
  Ficicioglu, C. n., Matalon, D. R., Luongo, N. n., Menello, C. n., Kornafel, T. n., Degnan, A. J.
  2020; 15 (1): 336

- **De novo variants in SUPT16H cause neurodevelopmental disorders associated with corpus callosum abnormalities.** *Journal of medical genetics*
  Bina, R. n., Matalon, D. n., Fregeau, B. n., Tarsitano, J. J., Aukrust, I. n., Houge, G. n., Bend, R. n., Warren, H. n., Stevenson, R. E., Stuurman, K. E., Barkovich, A. J., Sherr, E. H.
  2020

- **ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis** *GENETICS IN MEDICINE*
  Deisseroth, C. A., Birgmeier, J., Bodle, E. E., Kohler, J. N., Matalon, D. R., Nazarenko, Y., Genetti, C. A., Brownstein, C. A., Schmitz-Abe, K., Schoch, K., Cope, H., Signer, R., Network, et al
  2019; 21 (7): 1585–93

- **LOCALIZING NEUROLOGIC FEATURES AT PRESENTATION OF VLCAD DEFICIENCY**
  Leahy, P., Matalon, D., Ruzhnikov, M., Cowan, T., Enns, G.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2019: 282

- **Paraspinal Atrophy Suggesting Underlying Genetic Etiology**
  Schwartz, D., Fong, J., Matalon, D., Wong, J., Greene, M.
  LIPPINCOTT WILLIAMS & WILKINS.2019

- **LOCALIZING NEUROLOGIC FEATURES AT PRESENTATION OF VLCAD DEFICIENCY**
  Leahy, P., Matalon, D., Ruzhnikov, M., Cowan, T., Enns, G.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2019: 311

- **THE DIAGNOSIS AND NATURAL HISTORY OF THE MUCOPOLYSACCHARIDOSIS IVA IN ONE FAMILY**
  Matalon, D. R., Dougherty, P., Lulis, L., Medne, L., Krantz, I., Yum, S., Ficicioglu, C.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2018: 249–50

- **The diagnosis and natural history of mucopolysaccharidosis type IVA in one family**
  Matalon, D., Dougherty, P., Lulis, L., Medne, L., Krantz, I., Yum, S., Ficicioglu, C.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2018: S92

- **ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Deisseroth, C. A., Birgmeier, J. n., Bodle, E. E., Kohler, J. N., Matalon, D. R., Nazarenko, Y. n., Genetti, C. A., Brownstein, C. A., Schmitz-Abe, K. n., Schoch, K. n., Cope, H. n., Signer, R. n., Martinez-Agosto, et al
  2018