Stanford

# Ana Tesi-Rocha, MD

Clinical Professor, Neurology & Neurological Sciences

Curriculum Vitae available Online

### CLINICAL OFFICES

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## Bio

### CLINICAL FOCUS

- Pediatric Neurology
- Neuromuscular Diseases

### ACADEMIC APPOINTMENTS

- Clinical Professor, Neurology & Neurological Sciences
- Member, Wu Tsai Human Performance Alliance
- Member, Maternal & Child Health Research Institute (MCHRI)

### PROFESSIONAL EDUCATION

- Fellowship: Children's National Medical Center Pediatric Infectious Diseases Fellowship (2010) DC
- Board Certification: Pediatric Neurology, American Board of Psychiatry and Neurology (2010)
- Internship: Georgetown University Hospital (2007) DC
- Residency: Hospital Nacional de Pediatria Juan P Garrahan (2001) Argentina
- Residency: Alejandro Posadas Hospital (1997) Argentina
- Medical Education: Universidad de Buenos Aires (1994)

### LINKS

- Get a Second Opinion: https://stanfordhealthcare.org/second-opinion/overview.html

## Publications

### PUBLICATIONS

- **Major Adverse Dystrophinopathy Events (MADE) score as marker of cumulative morbidity and risk for mortality in boys with Duchenne muscular dystrophy** *PROGRESS IN PEDIATRIC CARDIOLOGY*
  Kaufman, B. D., Garcia, A., He, Z., Tesi-Rocha, C., Buu, M., Rosenthal, D., Gordish-Dressman, H., Almond, C. S., Duong, T., CINRG DUCHENNE NAT HIST STUDY
  2023; 69

- **Spinal Muscular Atrophy Type 1: Fetal Diagnosis, Prenatal Coordination, and Postnatal Management in the Era of Novel Therapies.** *NeoReviews*
  Chitkara, R., Chock, V., Davis, A., Rocha, C. T., Day, J. W., Fluharty, B., Hintz, S.
  2022; 23 (7): e520-e526

- **Correction to: Reldesemtiv in Patients with Spinal Muscular Atrophy: a Phase 2 Hypothesis-Generating Study.** *Neurotherapeutics : the journal of the American Society for Experimental NeuroTherapeutics*
  Rudnicki, S. A., Andrews, J. A., Duong, T., Cockroft, B. M., Malik, F. I., Meng, L., Wei, J., Wolff, A. A., Genge, A., Johnson, N. E., Tesi-Rocha, C., Connolly, A. M., Darras, et al
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- **Advances in the therapy of Spinal Muscular Atrophy.** *The Journal of pediatrics*
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  2021

- **Reldesemtiv in Patients with Spinal Muscular Atrophy: a Phase 2 Hypothesis-Generating Study.** *Neurotherapeutics : the journal of the American Society for Experimental NeuroTherapeutics*
  Rudnicki, S. A., Andrews, J. A., Duong, T. n., Cockroft, B. M., Malik, F. I., Meng, L. n., Wei, J. n., Wolff, A. A., Genge, A. n., Johnson, N. E., Tesi-Rocha, C. n., Connolly, A. M., Darras, et al
  2021

- **Knee Strength and Ankle Range of Motion Impacts on Timed Function Tests in Duchenne Muscular Dystrophy: In the Era of Glucocorticoids.** *Journal of neuromuscular diseases*
  Duong, T., Canbek, J., Fernandez-Fernandez, A., Henricson, E., Birkmeier, M., Siener, C., Rocha, C. T., McDonald, C., Gordish-Dressman, H.
  2021

- **TCTEX1D1 is a genetic modifier of disease progression in Duchenne muscular dystrophy.** *European journal of human genetics : EJHG*
  Spitali, P., Zaharieva, I., Bohringer, S., Hiller, M., Chaouch, A., Roos, A., Scotton, C., Claustres, M., Bello, L., McDonald, C. M., Hoffman, E. P., Koeks, Z., Eka Suchiman, et al
  2020; 28 (6): 815-825

- **Genetic modifiers of respiratory function in Duchenne muscular dystrophy.** *Annals of clinical and translational neurology*
  Bello, L., D'Angelo, G., Villa, M., Fusto, A., Vianello, S., Merlo, B., Sabbatini, D., Barp, A., Gandossini, S., Magri, F., Comi, G. P., Pedemonte, M., Tacchetti, et al
  2020; 7 (5): 786-798

- **Combined Genome Sequencing and RNA Analysis Reveals and Characterizes a Deep Intronic Variant in IGHMBP2 in a Patient With Spinal Muscular Atrophy With Respiratory Distress Type 1.** *Pediatric neurology*
  Bodle, E. E., Zhu, W. n., Velez-Bartolomei, F. n., Tesi-Rocha, A. n., Liu, P. n., Bernstein, J. A.
  2020; 114: 16–20

- **Medical management of muscle weakness in Duchenne muscular dystrophy.** *PloS one*
  Rivera, S. R., Jhamb, S. K., Abdel-Hamid, H. Z., Acsadi, G., Brandsema, J., Ciafaloni, E., Darras, B. T., Iannaccone, S. T., Konersman, C. G., Kuntz, N. L., McDonald, C. M., Parsons, J. A., Tesi Rocha, et al
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- **Towards regulatory endorsement of drug development tools to promote the application of model-informed drug development in Duchenne muscular dystrophy.** *Journal of pharmacokinetics and pharmacodynamics*

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2019

- **Chronic Polyneuritis of Childhood** *JOURNAL OF PEDIATRICS*

  Klotz, J., Rocha, C.

  2019; 208: 175

- **Assessment of disease progression in dysferlinopathy: A 1-year cohort study** *NEUROLOGY*

  Moore, U., Jacobs, M., James, M. K., Mayhew, A. G., Fernandez-Torron, R., Feng, J., Cnaan, A., Eagle, M., Bettinson, K., Rufibach, L. E., Lofra, R., Blamire, A. M., Carlier, et al

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- **Assessment of disease progression in dysferlinopathy: A 1-year cohort study.** *Neurology*

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  2019

- **50 Years Ago in The Journal of Pediatrics: Chronic Polyneuritis of Childhood.** *The Journal of pediatrics*

  Klotz, J. n., Tesi Rocha, C. n.

  2019; 208: 175

- **Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study** *JOURNAL OF NEUROLOGY NEUROSURGERY AND PSYCHIATRY*

  Moore, U. R., Jacobs, M., Fernandez-Torron, R., Jang, J., James, M. K., Mayhew, A., Rufibach, L., Mittal, P., Eagle, M., Cnaan, A., Carlier, P. G., Blamire, A., Hilsden, et al

  2018; 89 (11): 1224-+

- **Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy** *NEW ENGLAND JOURNAL OF MEDICINE*

  Mercuri, E., Darras, B. T., Chiriboga, C. A., Day, J. W., Campbell, C., Connolly, A. M., Iannaccone, S. T., Kirschner, J., Kuntz, N. L., Saito, K., Shieh, P. B., Tulinius, M., Mazzone, et al

  2018; 378 (7): 625–35

- **Long-term effects of glucocorticoids on function, quality of life, and survival in patients with Duchenne muscular dystrophy: a prospective cohort study** *LANCET*

  McDonald, C. M., Henricson, E. K., Abresch, R. T., Duong, T., Joyce, N. C., Hu, F., Clemens, P. R., Hoffman, E. P., Cnaan, A., Gordish-Dressman, H., CINRG Investigators

  2018; 391 (10119): 451–61

- **Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials.** *Journal of neurology, neurosurgery, and psychiatry*

  Diaz-Manera, J. n., Fernandez-Torron, R. n., LLauger, J. n., James, M. K., Mayhew, A. n., Smith, F. E., Moore, U. R., Blamire, A. M., Carlier, P. G., Rufibach, L. n., Mittal, P. n., Eagle, M. n., Jacobs, et al

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- **Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study.** *Journal of neurology, neurosurgery, and psychiatry*

  Moore, U. R., Jacobs, M. n., Fernandez-Torron, R. n., Jang, J. n., James, M. K., Mayhew, A. n., Rufibach, L. n., Mittal, P. n., Eagle, M. n., Cnaan, A. n., Carlier, P. G., Blamire, A. n., Hilsden, et al

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- **Loss-of-Function Mutations in LGI4, a Secreted Ligand Involved in Schwann Cell Myelination, Are Responsible for Arthrogryposis Multiplex Congenita** *AMERICAN JOURNAL OF HUMAN GENETICS*

  Xue, S., Maluenda, J., Marguet, F., Shboul, M., Quevarec, L., Bonnard, C., Ng, A. Y., Tohari, S., Thong Teck Tan, T. T., Kong, M. K., Monaghan, K. G., Cho, M. T., Siskind, et al

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- **Evidence for ACTN3 as a genetic modifier of Duchenne muscular dystrophy** *NATURE COMMUNICATIONS*

  Hogarth, M. W., Houweling, P. J., Thomas, K. C., Gordish-Dressman, H., Bello, L., Pegoraro, E., Hoffman, E. P., Head, S. I., North, K. N.

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- **Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy.** *The New England journal of medicine*

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- **Targeted Re-Sequencing Emulsion PCR Panel for Myopathies: Results in 94 Cases.** *Journal of neuromuscular diseases*

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- **50 Years Ago in THE JOURNAL OF PEDIATRICS Myotonic Dystrophy: A Neglected Form of Mental Retardation** *JOURNAL OF PEDIATRICS*

  Rocha, C. T.

  2016; 170: 112-112

- **Exome Sequencing Identifies DYNC1H1 Variant Associated With Vertebral Abnormality and Spinal Muscular Atrophy With Lower Extremity Predominance.** *Pediatric neurology*

  Punetha, J., Monges, S., Franchi, M. E., Hoffman, E. P., Cirak, S., Tesi-Rocha, C.

  2015; 52 (2): 239-244

- **COOPERATIVE INTERNATIONAL NEUROMUSCULAR RESEARCH GROUP DUCHENNE NATURAL HISTORY STUDY DEMONSTRATES INSUFFICIENT DIAGNOSIS AND TREATMENT OF CARDIOMYOPATHY IN DUCHENNE MUSCULAR DYSTROPHY** *MUSCLE & NERVE*

  Spurney, C., Shimizu, R., Morgenroth, L. P., Kolski, H., Gordish-Dressman, H., Clemens, P. R.

  2014; 50 (2): 250-256

- **Novel large deletion in the ACTA1 gene in a child with autosomal recessive nemaline myopathy** *NEUROMUSCULAR DISORDERS*

  Friedman, B., Simpson, K., Tesi-Rocha, C., Zhou, D., Palmer, C. A., Suchy, S. F.

  2014; 24 (4): 331-334

- **'Double trouble': diagnostic challenges in Duchenne muscular dystrophy in patients with an additional hereditary skeletal dysplasia.** *Neuromuscular disorders*

  Donkervoort, S., Schindler, A., Tesi-Rocha, C., Schreiber, A., Leach, M. E., Dastgir, J., Hu, Y., Mankodi, A., Wagner, K. R., Friedman, N. R., Bönnemann, C. G.

  2013; 23 (12): 955-961

- **The cooperative international neuromuscular research group Duchenne natural history study: Glucocorticoid treatment preserves clinically meaningful functional milestones and reduces rate of disease progression as measured by manual muscle testing and other commonly used clinical trial outcome measures** *MUSCLE & NERVE*

  Henricson, E. K., Abresch, R. T., Cnaan, A., Hu, F., Duong, T., Arrieta, A., Han, J., Escolar, D. M., Florence, J. M., Clemens, P. R., Hoffman, E. P., McDonald, C. M.

  2013; 48 (1): 55-67

- **Pentoxifylline as a rescue treatment for DMD A randomized double-blind clinical trial** *NEUROLOGY*

  Escolar, D. M., Zimmerman, A., Bertorini, T., Clemens, P. R., Connolly, A. M., Mesa, L., Gorni, K., Kornberg, A., Kolski, H., Kuntz, N., Nevo, Y., Tesi-Rocha, C., Nagaraju, et al

  2012; 78 (12): 904-913

- **LIQUID FORMULATION OF PENTOXIFYLLINE IS A POORLY TOLERATED TREATMENT FOR DUCHENNE DYSTROPHY** *MUSCLE & NERVE*

  Zimmerman, A., Clemens, P. R., Tesi-Rocha, C., Connolly, A., Iannaccone, S. T., Kuntz, N., Arrieta, A., Hache, L., Henricson, E., Hu, F., Mayhew, J., Escolar, D. M.

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- **SPP1 genotype is a determinant of disease severity in Duchenne muscular dystrophy** *NEUROLOGY*

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- **Limb-Girdle and Congenital Muscular Dystrophies: Current Diagnostics, Management, and Emerging Technologies** *CURRENT NEUROLOGY AND NEUROSCIENCE REPORTS*

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- **PPAR alpha L162V underlies variation in serum triglycerides and subcutaneous fat volume in young males** *BMC MEDICAL GENETICS*

Uthurralt, J., Gordish-Dressman, H., Bradbury, M., Tesi-Rocha, C., Devaney, J., Harmon, B., Reeves, E. K., Brandoli, C., Hansen, B. C., Seip, R. L., Thompson, P. D., Price, T. B., Angelopoulos, et al

2007; 8

- **CINRG randomized controlled trial of creatine and glutamine in Duchenne muscular dystrophy** *ANNALS OF NEUROLOGY*

  Escolar, D. M., Buyse, G., Henricson, E., Leshner, R., Florence, J., Mayhew, J., Tesi-Rocha, C., Gorni, K., Pasquali, L., Patel, K. M., McCarter, R., Huang, J., Mayhew, et al

  2005; 58 (1): 151-155

- **Update on diagnosis and treatment of hereditary and acquired polyneuropathies in childhood.** *Supplements to Clinical neurophysiology*

  Rocha, C. T., Escolar, D. M.

  2004; 57: 255-271

- **Drop episodes in Coffin-Lowry syndrome: an unusual type of startle response** *EPILEPTIC DISORDERS*

  Caraballo, R., Rocha, A. T., Medina, C., Fejerman, N.

  2000; 2 (3): 173-176