Stanford

Ana Tesi-Rocha, MD

- Clinical Professor, Pediatric Neurology
- Clinical Professor (By courtesy), Pediatrics
- Curriculum Vitae available Online

CLINICAL OFFICE (PRIMARY)

- **Child Neurology**

  730 Welch Rd Ste 206

  Palo Alto, CA 94304

  Tel (650) 723-0993 **Fax** (650) 721-6350

Bio

CLINICAL FOCUS

- Neuromuscular Diseases
- Neuromuscular Medicine

ACADEMIC APPOINTMENTS

- Clinical Professor, Pediatric Neurology
- Clinical Professor (By courtesy), Pediatrics
- Member, Wu Tsai Human Performance Alliance
- Member, Maternal & Child Health Research Institute (MCHRI)

PROFESSIONAL EDUCATION

- Board Certification: Neurology with Special Qualifications in Child Neurology, American Board of Psychiatry and Neurology (2010)
- Fellowship: Children's National Medical Center Pediatric Infectious Diseases Fellowship (2010) DC
- 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

Research & Scholarship

CLINICAL TRIALS

- Observation Study in Patients Age 0-5 Years With LAMA2-related Congenital Muscular Dystrophy, Recruiting
- Efficacy and Safety of Apitegromab in Patients With Later-Onset Spinal Muscular Atrophy Treated With Nusinersen or Risdiplam, Not Recruiting

Publications

PUBLICATIONS

- **Identification of prognostic biomarkers in a large cohort of patients with LGMD R2.** *Journal of neurology*
  Bolano-Diaz, C. F., Verdu-Diaz, J., Hao, D., James, M. K., Rufibach, L., Blamire, A., Reyngoudt, H., Carlier, P. G., Gordish-Dressman, H., Hilsden, H., Spuler, S., Day, J., Jones, et al
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- **Safety and efficacy of apitegromab in nonambulatory type 2 or type 3 spinal muscular atrophy (SAPPHIRE): a phase 3, double-blind, randomised, placebo-controlled trial.** *The Lancet. Neurology*
  Crawford, T. O., Servais, L., Mercuri, E., Kölbel, H., Kuntz, N., Finkel, R. S., Krueger, J., Batley, K., Young, S. D., Marantz, J. L., Song, G., Yao, B., Zhao, et al
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- **Modeling of Dysferlinopathy (LGMDR2) Progression: A Longitudinal Fat Fraction Analysis.** *Neurology. Genetics*
  Bolano-Diaz, C. F., Reyngoudt, H., Wilson, I. J., James, M. K., Smith, F. E., Caldas de Almeida Araujo, E., Gordish-Dressman, H., Hilsden, H., Rufibach, L. E., Wallace, D., Ward, L., Stramare, R., Rampado, et al
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- **RGX-202, an Investigational Gene Therapy for the Treatment of Duchenne Muscular Dystrophy: Interim Clinical Data**
  Danos, O., Harper, A., Iannaccone, S., Kuntz, N., Tesi-Rocha, C., Veerapandiyan, A., Boulos, N., Curtiss, S., Fiscella, M., Gilmor, M., Pakola, S., Patel, H., Philips, et al
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- **Eteplirsen Treatment for Duchenne Muscular Dystrophy: A Qualitative Patient Experience Study.** *Advances in therapy*
  Iff, J., Carmichael, C., McKee, S., Sehinovych, I., McNeill, C., Tesi-Rocha, C., Henricson, E., Muntoni, F., Kitchen, H.
  2024

- **Assessing the Assisted Six-Minute Cycling Test as a Measure of Endurance in Non-Ambulatory Patients with Spinal Muscular Atrophy (SMA).** *Journal of clinical medicine*
  Tang, W. J., Gu, B., Montalvo, S., Dunaway Young, S., Parker, D. M., de Monts, C., Ataide, P., Ni Ghiollagain, N., Wheeler, M. T., Tesi Rocha, C., Christle, J. W., He, Z., Day, et al
  2023; 12 (24)

- **The emerging spectrum of neurodevelopmental comorbidities in early-onset Spinal Muscular Atrophy.** *European journal of paediatric neurology : EJPN : official journal of the European Paediatric Neurology Society*
  Baranello, G.
  2023; 48: 67-68

- **Cerebrospinal Fluid Proteomic Changes after Nusinersen in Patients with Spinal Muscular Atrophy.** *Journal of clinical medicine*
  Beaudin, M., Kamali, T., Tang, W., Hagerman, K. A., Dunaway Young, S., Ghiglieri, L., Parker, D. M., Lehallier, B., Tesi-Rocha, C., Sampson, J. B., Duong, T., Day, J. W.
  2023; 12 (20)

- **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.
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- **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
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- **Advancing Team-Based Care for Spinal Muscular Atrophy: A Multi-State Project ECHO Initiative and National Education Strategy**
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- **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.
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- **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*
  Klotz, J., Rocha, C. T., Young, S. D., Duong, T., Buu, M., Sampson, J., Day, J. W.
  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
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- **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.
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- **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
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- **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.
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- **Chronic Polyneuritis of Childhood** *JOURNAL OF PEDIATRICS*
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  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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- **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
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- **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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  2018

- **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*
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- **Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy.** *The New England journal of medicine*
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- **50 Years Ago in THE JOURNAL OF PEDIATRICS Myotonic Dystrophy: A Neglected Form of Mental Retardation** *JOURNAL OF PEDIATRICS*
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- **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.
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- **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.
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- **'Double trouble': diagnostic challenges in Duchenne muscular dystrophy in patients with an additional hereditary skeletal dysplasia.** *Neuromuscular disorders*
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- **Pentoxifylline as a rescue treatment for DMD A randomized double-blind clinical trial** *NEUROLOGY*
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- **LIQUID FORMULATION OF PENTOXIFYLLINE IS A POORLY TOLERATED TREATMENT FOR DUCHENNE DYSTROPHY** *MUSCLE & NERVE*
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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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  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
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- **Update on diagnosis and treatment of hereditary and acquired polyneuropathies in childhood.** *Supplements to Clinical neurophysiology*
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- **Drop episodes in Coffin-Lowry syndrome: an unusual type of startle response** *EPILEPTIC DISORDERS*
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