



## Ana Tesi-Rocha, MD

- Clinical Associate Professor, Neurology & Neurological Sciences
- Clinical Associate Professor, Pediatrics - Operations
- 📄 Curriculum Vitae available Online

### CLINICAL OFFICES

- **Child Neurology**

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### Bio

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### CLINICAL FOCUS

- Pediatric Neurology
- Neuromuscular Diseases

### ACADEMIC APPOINTMENTS

- Clinical Associate Professor, Neurology & Neurological Sciences
- Clinical Associate Professor, Pediatrics - Operations
- 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>

## Research & Scholarship

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### CLINICAL TRIALS

- A Study of CK-2127107 in Patients With Spinal Muscular Atrophy, Recruiting

## Publications

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### PUBLICATIONS

- **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): 815–25
- **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  
2020; 15 (10): e0240687
- **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., Velez-Bartolomei, F., Tesi-Rocha, A., Liu, P., Bernstein, J. A.  
2020; 114: 16–20
- **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): 786–98
- **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*  
Conrado, D. J., Larkindale, J., Berg, A., Hill, M., Burton, J., Abrams, K. R., Abresch, R. T., Bronson, A., Chapman, D., Crowther, M., Duong, T., Gordish-Dressman, H., Harnisch, et al  
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  
2019; 92 (5): E461–E474
- **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. M., Blamire, A. M., Carlier, et al  
2019
- **50 Years Ago in The Journal of Pediatrics: Chronic Polyneuritis of Childhood.** *The Journal of pediatrics*  
Klotz, J., Tesi Rocha, C.  
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., Fernandez-Torron, R., LLauger, J., James, M. K., Mayhew, A., Smith, F. E., Moore, U. R., Blamire, A. M., Carlier, P. G., Rufibach, L., Mittal, P., Eagle, M., Jacobs, et al  
2018
- **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
- **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  
2017; 100 (4): 659-665
- **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.  
2017; 8
- **Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy.** *The New England journal of medicine*  
Finkel, R. S., Mercuri, E., Darras, B. T., Connolly, A. M., Kuntz, N. L., Kirschner, J., Chiriboga, C. A., Saito, K., Servais, L., Tizzano, E., Topaloglu, H., Tulinius, M., Montes, et al  
2017; 377 (18): 1723-32
- **Targeted Re-Sequencing Emulsion PCR Panel for Myopathies: Results in 94 Cases.** *Journal of neuromuscular diseases*  
Punetha, J., Kesari, A., Uapinyoying, P., Giri, M., Clarke, N. F., Waddell, L. B., North, K. N., Ghaoui, R., O'Grady, G. L., Oates, E. C., Sandaradura, S. A., Bönnemann, C. G., Donkervoort, et al  
2016; 3 (2): 209-225
- **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.  
2011; 44 (2): 170-173
- **SPP1 genotype is a determinant of disease severity in Duchenne muscular dystrophy** *NEUROLOGY*  
Pegoraro, E., Hoffman, E. P., Piva, L., Gavassini, B. F., Cagnin, S., Ermani, M., Bello, L., Soraru, G., Pacchioni, B., Bonifati, M. D., Lanfranchi, G., Angelini, C., Kesari, et al  
2011; 76 (3): 219-226
- **Limb-Girdle and Congenital Muscular Dystrophies: Current Diagnostics, Management, and Emerging Technologies** *CURRENT NEUROLOGY AND NEUROSCIENCE REPORTS*  
Rocha, C. T., Hoffman, E. P.  
2010; 10 (4): 267-276
- **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