

**Roberto A. Ortega, MS**  
MD-PhD student, Albert Einstein College of Medicine

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**Personal statement:** My overarching career goal is to develop as a physician-scientist, including as a practicing physician and as a grant-funded statistical geneticist and translational researcher.

## Education

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June 2023-current	<b>Albert Einstein College of Medicine</b> Medical Scientist Training Program (MSTP, MD-PhD)	<b>Bronx, NY</b>
August 2011	<b>The University of Texas at El Paso</b> Master of Science, Statistics (MS) Thesis: Computational Methods of Hidden Markov Models with Respect to CpG Island Prediction in DNA Sequences Advisor: Ming-Ying Leung, PhD El Paso, TX	<b>El Paso, TX</b>
December 2008	<b>The University of Texas at El Paso</b> Bachelor of Science, Psychology (BS, Statistics minor)	<b>El Paso, TX</b>
May 2005	<b>El Paso Community College</b> Associate of Arts, General studies	<b>El Paso, TX</b>

## Professional Work

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<b>Biostatistician II</b> Icahn School of Medicine at Mount Sinai / Mount Sinai Beth Israel Clinical and translational genetics of movement disorders Neurology - movement disorders New York, NY USA PI: Rachel Saunders-Pullman, MD, MPH, MS	August 2021-June 2023
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<b>Senior Associate Researcher</b> Icahn School of Medicine at Mount Sinai / Mount Sinai Beth Israel	December 2015-August 2021
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<b>Research Associate</b> Icahn School of Medicine at Mount Sinai / Mount Sinai Beth Israel	September 2012-December 2015
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**Co-director, Evidence-based medicine Journal Club.** Neurology Residents 2019-present

<b>Coordinator - Cardiac Catheterization Lab</b> New York Presbyterian Columbia University Medical Center New York, NY USA	November 2011 - September 2012
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## Authored Manuscripts

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**First author**

1. **Ortega, RA**, Bressman, SB, Raymond, D., et al. (2023), Response: Differences in Sex-Specific Frequency of Glucocerebrosidase Variant Carriers and Familial Parkinsonism. *Mov Disord*, 38: 714-715. <https://doi.org/10.1002/mds.29353>
2. **Ortega RA**, Bressman SBB, Raymond D, et al. Differences in Sex-Specific Frequency of Glucocerebrosidase Variant Carriers and Familial Parkinsonism. *Mov Disord*. 2022 Aug 27. DOI: 10.1002/mds.29197. PMID: 36054306
3. **Ortega RA**, Bodamer O, Peake RWA, et al. Assessment of Glucocerebrosidase Enzyme Activity in Parkinson Disease Using Multiple Approaches. *Mov Disord*. 2022 Feb 1. doi: 10.1002/mds.28951. PMID: 35106836.
4. **Ortega RA**, Wang C, Raymond D, et al. Association of Dual LRRK2 G2019S and GBA Variations With Parkinson Disease Progression. *JAMA Netw Open*. 2021;4(4):e215845.
5. **Ortega RA**, Groves M, Mirelman A, et al. Evidence for increased completed suicide in first-degree relatives of *LRRK2* G2019S mutation Parkinson's disease. *J Neurol Neurosurg Psychiatry*. 2018 Nov 8. PubMed PMID: 30409888.
6. **Ortega RA**, Torres PA, Swan M, et al. Glucocerebrosidase enzyme activity in *GBA* mutation Parkinson's disease. *J Clin Neurosci*. 2016 Jun; 28:185-6. Epub 2016 Feb 5. PubMed PMID: 26857292; PubMed Central PMCID: PMC4856562.

#### Co-author

7. Miltenberger-Miltenyi G, **Ortega RA**, Domingo A, et al. Genetic risk variants in New Yorkers of Puerto Rican and Dominican Republic heritage with Parkinson's disease. *NPJ Parkinsons Dis*. 2023 Dec 7;9(1):160. PMID: 38062033; PMCID: PMC10703927.
8. Saunders-Pullman R, **Ortega RA**, Wang C, et al. Association of olfactory performance with motor decline and age at onset in people with Parkinson's disease and the LRRK2 G2019S variant. *Neurology*. 2022 June 03. PMID: 35995594
9. Chitpin JG, Surendra A, Nguyen TT, Taylor GP, Xu H, Alecu I, **Ortega R**, et al. BATL: Bayesian annotations for targeted lipidomics. *Bioinformatics*. 2021 Dec 24. PMID: 34951624.
10. Leaver K, Viser A, Kopell BH, **Ortega RA**, et al. Clinical profiles and outcomes of deep brain stimulation in G2019S LRRK2 Parkinson disease. *J Neurosurg*. 2021 Nov 19:1-8. PMID: 34798606.
11. Radmard S, **Ortega RA**, et al. Using computerized spiral analysis to evaluate deep brain stimulation outcomes in Parkinson disease. *Clin Neurol Neurosurg*. 2021 Sep;208:106878. Epub 2021 Aug 8. PMID: 34418700.
12. Moran EE, Bressman SB, **Ortega RA**, et al. Cognitive Functioning of Glucocerebrosidase (GBA) Non-manifesting Carriers. *Front Neurol*. 2021 Feb 26. PMID: 33716938 DOI: 10.3389/fneur.2021.635958
13. Agalliu I, **Ortega RA**, Luciano MS, et al. Cancer outcomes among Parkinson's disease patients with leucine rich repeat kinase 2 mutations, idiopathic Parkinson's disease patients, and nonaffected controls. *Mov Disord*. 2019 Jul 26. PubMed PMID: 31348549.
14. Xu H, Boucher FR, Nguyen TT, Taylor GP, Tomlinson J, **Ortega RA**, et al. Differential mobility spectrometry as an orthogonal separation to LC-ESI-MS/MS for quantification of isomeric cerebrosides in plasma and cerebrospinal fluid. *J Lipid Res*. 2018 Nov 9. PubMed PMID: 30413651.
15. Pullman M, **Ortega R**, Glickman A, et al. Increased substantia nigra echogenicity in *LRRK2* family members without mutations. *Mov Disord*. 2018 Sep;33(9):1504-1505. PubMed PMID: 30145825.

16. Ratliff J, **Ortega RA**, Ooi HY, et al. Digitized spiral analysis may be a potential biomarker for brachial dystonia. *Parkinsonism Relat Disord*. 2018 Jul 20. PubMed PMID: 30037691.
17. Saunders-Pullman R, Mirelman A, Alcalay RN, Wang C, **Ortega RA**, et al. Progression in the *LRRK2*-Associated Parkinson Disease Population. *JAMA Neurol*. 2018 Mar 1;75(3):312-319. PubMed PMID: 29309488; PubMed Central PMCID: PMC5885854.
18. San Luciano M, Wang C, **Ortega RA**, et al. Sex differences in *LRRK2* G2019S and idiopathic Parkinson's Disease. *Ann Clin Transl Neurol*. 2017 Oct 19;4(11):801-810. PubMed PMID: 29159192; PubMed Central PMCID: PMC5682117.
19. Moran EE, Wang C, Katz M, Ozelius L, Schwartz A, Pavlovic J, **Ortega RA**, et al. Cognitive and motor functioning in elderly glucocerebrosidase mutation carriers. *Neurobiol Aging*. 2017 Oct. Epub 2017 Jun 24. PubMed PMID: 28728889; PubMed Central PMCID: PMC5647652.
20. Swan M, Doan N, **Ortega RA**, et al. Neuropsychiatric characteristics of *GBA*-associated Parkinson disease. *J Neurol Sci*. 2016 Nov 15; 370:63-69. Epub 2016 Aug 30. PubMed PMID: 27772789; PubMed Central PMCID: PMC5268078.
21. San Luciano M, Wang C, **Ortega RA**, et al. Digitized Spiral Drawing: A Possible Biomarker for Early Parkinson's Disease. *PLoS One*. 2016 Oct 12;11(10). eCollection 2016. PubMed PMID: 27732597; PubMed Central PMCID: PMC5061372.
22. Saunders-Pullman R, Alcalay RN, Mirelman A, Wang C, Luciano MS, **Ortega RA**, et al. REM sleep behavior disorder, as assessed by questionnaire, in G2019S *LRRK2* mutation PD and carriers. *Mov Disord*. 2015 Nov;30(13):1834-9. Epub 2015 Sep 14. PubMed PMID: 26366513; PubMed Central PMCID: PMC4715645.
23. Saunders-Pullman R, Mirelman A, Wang C, Alcalay RN, San Luciano M, **Ortega R**, et al. Olfactory identification in *LRRK2* G2019S mutation carriers: a relevant marker? *Ann Clin Transl Neurol*. 2014 Sep;1(9):670-8. Epub 2014 Sep 30. PubMed PMID: 25493281; PubMed Central PMCID: PMC4241794.
24. Hess CW, Hsu AW, Yu Q, **Ortega R**, Pullman SL. Increased variability in spiral drawing in patients with functional (psychogenic) tremor. *Hum Mov Sci*. 2014 Dec; 38:15-22. Epub 2014 Sep 18. PubMed PMID: 25240176.
25. Saunders-Pullman R, Fuchs T, San Luciano M, Raymond D, Brashear A, **Ortega R**, et al. Heterogeneity in primary dystonia: lessons from *THAP1*, *GNAL*, and *TOR1A* in Amish Mennonites. *Mov Disord*. 2014 May;29(6):812-8. Epub 2014 Feb 5. PubMed PMID: 24500857; PubMed Central PMCID: PMC4013240.
26. Alcalay RN, Mirelman A, Saunders-Pullman R, Tang MX, Mejia Santana H, Raymond D, Roos E, Orbe-Reilly M, Gurevich T, Bar Shira A, Gana Weisz M, Yasinovsky K, Zalis M, Thaler A, Deik A, Barrett MJ, Cabassa J, Groves M, Hunt AL, Lubarr N, San Luciano M, Miravite J, Palmese C, Sachdev R, Sarva H, Severt L, Shanker V, Swan MC, Soto-Valencia J, Johannes B, **Ortega R**, et al. Parkinson disease phenotype in Ashkenazi Jews with and without *LRRK2* G2019S mutations. *Mov Disord*. 2013 Dec;28(14):1966-71. Epub 2013 Oct 15. PubMed PMID: 24243757; PubMed Central PMCID: PMC3859844.

## Invited Presentations

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**PDBP NINDS Annual Meeting** (Virtual, October 2022). Presented two clinical papers from our

group describing (1) the relationship between baseline olfaction and motor and cognitive longitudinal progression in LRRK2 G2019S PD and idiopathic PD and separately (2) the sex-specific distribution in different genetic forms of PD, and sex-related differences in family history of parkinsonism

**PDBP NINDS Annual Meeting** (Bethesda, MD; August 2019). Presented an analysis update on grant U01NS094148, Evaluation of glucocerebrosidase pathway biomarkers in Parkinson Disease, including results from alpha-synuclein and glucocerebrosidase assays in plasma, serum, and CSF in GBA and non-GBA variant carriers with PD

**Movement Disorders Rounds** (Mount Sinai Beth Israel, NY; May 2018): Presented results of a clinical paper from our group describing the observation of increased frequency of suicide in first degree relatives of LRRK2 G2019S mutation parkinson disease

**PDBP NINDS Annual Meeting** (Bethesda, MD; August 2018). Presented an analysis update of grant U01NS094148, Evaluation of glucocerebrosidase pathway biomarkers in Parkinson Disease, including initial results of our study of peripheral glucocerebrosidase activity and sphingolipid levels

**PDBP NINDS Annual Meeting** (Bethesda, MD; August 2017). Presented an analysis update of grant U01NS094148, Evaluation of glucocerebrosidase pathway biomarkers in Parkinson Disease, including a pilot study of peripheral glucocerebrosidase activity and leukocyte levels, as well as GBA variant distribution, and the noted sex differences in the cohort

**PDBP NINDS Consortium Call:** Presented methodology of algorithm developed for accessing, utilizing, and interpreting NeuroX Data (genotyping platform in PD) in PDBP

**American Academy of Neurology** (Platform presentation, San Diego 2013): Presented results of a clinical paper from our group describing the progression of motor dysfunction in elderly LRRK2 carriers, including prior to phenoconversion to PD

## Posters & Presentations

1. **ANA Annual Conference 2021** (virtual): Poster 396. Clusters of Olfactory Performance Are Associated with Motor Decline in LRRK2 G2019S Variant Parkinson Disease. **RA Ortega**, R Gerber, D Raymond, K Leaver, N Urval, V Katsnelson, RN Alcalay, A Mirelman, MC Brumm, CS Coffey, T Simuni, AD Siderowf, K Marek, N Giladi, KS Marder, SB Bressman, R Saunders-Pullman
2. **MDS Annual Conference 2021** (virtual): Poster 745. Investigating LRRK2 and GBA genetic variability in underrepresented populations. C. Paisan-Ruiz, V. Katsnelson, N. Urval, **R. Ortega**, A. Debebe, I. Peter, S. Bressman, R. Saunders-Pullman
3. **ANA Annual Conference 2020** (virtual; October 2020): Poster 474. Slower Progression in Double Mutation GBA-LRRK2 (G2019S) Associated Parkinson Disease. **RA Ortega**, Cuiling Wang, Deborah Raymond, Fion Chu, Roy N. Alcalay, Anat Mirelman, Yuliya Kuras, Clemens R. Scherzer, Karen S. Marder, MD, Nir Giladi, Susan B. Bressman, Rachel Saunders-Pullman
4. **ANA Annual Conference 2020** (virtual; October 2020): Poster 497. Association of Co-

- Morbid Hypertension, Diabetes Mellitus and Body Mass Index in Idiopathic Parkinson's Disease (PD), LRRK2 Mutation PD and GBA Mutation PD. T Lin, D Raymond, S Elango, K Leaver, V Katsnelson, N Urval, L Meytin, V Shanker, M Swan, N Lubarr, M Groves, S Bressman, R Saunders-Pullman, **Roberto Ortega** (senior author)
5. **MDS PAS Annual Conference 2018** (Miami, FL; June 2018): Poster 212. Increased Peripheral Sphingolipids in GBA Mutation and Idiopathic Parkinson Disease. **RA Ortega**, Izolda Mileva, Amanda Glickman, Mariel Pullman, Imali Perera, Anna Vaigast, Sonya Elango, Elimelech Wieder, Michael Pauciulo, Deborah Raymond, William Nichols, Laurie Ozelius, Susan Bressman, Dongming Cai, Yusuf Hannun, Lina Obeid, Rachel Saunders-Pullman (New York, NY, USA)
  6. **MDS PAS Annual Conference 2018** (Miami, FL; June 2018): Long Term Outcomes of Late Deep Brain Stimulation in DYT1 Dystonia. HY Ooi, D Raymond, L Neimand, **RA Ortega et al.** (New York, NY, USA)
  7. **AAN Annual Conference 2018** (Los Angeles, CA; May 2018): Increased Frequency of Suicide In First Degree Relatives of LRRK2 G2019S Mutation Parkinson Disease (P1.052). **RA Ortega**, Anat Mirelman, Roy Alcalay, Sonya Elango, Deborah Raymond, Mark Groves, Nir Giladi, Karen Marder, Susan Bressman, Rachel Saunders-Pullman. *Neurology* Apr 2018, 90 (15 Supplement) P1.052
  8. **ANA Annual Conference 2017**: (San Diego, CA; October 2017): Possible link between Crohn's disease and LRRK2 mutation Parkinson's disease (Abstract 350423). **RA Ortega**, MS, Anat Mirelman, PhD, Claustre Pont-Sunyer, MD, Dolores Vilas, MD, Kathrin Brockmann, MD, Bjorg Warø, MD, Jan Aasly, MD, Karen Marder, MD MPH, Daniela Berg, MD, Tatiana Foroud, PhD, Eduardo Tolosa, MD, Nir Giladi, MD, Ilir Agalliu, MD ScD, Susan B Bressman, MD and Rachel Saunders-Pullman, MD MPH
  9. **ANA Annual Conference 2017**: (San Diego, CA; October 2017): Deep Brain Stimulation Surgery in N370S Glucocerebrosidase (GBA) Mutation Parkinson's Disease. P Laquer, BH Kopell, VL Shanker, **R Ortega**, et al (Abstract M250)
  10. **MDS International Congress 2017** (Vancouver, British Columbia, Canada; June 2017): Poster 1027. Spiral Analysis is a Promising Biomarker in LRRK2 G2019S Carriers. **RA Ortega**, M Pullman, A Glickman, I Perera, M San Luciano, A Mirelman, C Pont Sunyer, N Giladi, E Tolosa, Q Yu, SB Bressman, R Saunders-Pullman. \* Presented by R Saunders-Pullman
  11. **NYCEF Annual Meeting 2017** (NYU School of Medicine; March 2017): Cancer outcomes among LRRK2 G2019S Parkinson's Disease, idiopathic PD and controls: A pooled analysis using a standardized questionnaire. **RA Ortega**, M San Luciano, M Pullman, A Glickman, A Mirelman, C Pont Sunyer, D Vilas, Brockmann, B Warø, J Aasly, D Berg, T Foroud, E Tolosa, K Marder, N Giladi, SB Bressman, R Saunders-Pullman.
  12. **ANA Annual Conference 2016** (Baltimore, MD; October 2016) Poster S300. Dried Blood Spot Glucocerebrosidase Measurement is Reliable in Controls When adjusting for White Blood Cell Count. **RA Ortega**, OA Bodamer, Holly Merglesberg, C Wang, A Glickman, N Doan, D Raymond, JB Ratliff, J Squires, I Perera, A Vaigast, B Johannes, SB Bressman, R Saunders-Pullman.
  13. **MDS International Congress 2016** (Berlin, DE; June 2016): Poster 649. Frequency of GBA1 and LRRK2 G2019S mutations, and Body Mass Index in Ashkenazi Jews. N Doan, D Raymond, **RA Ortega**, et al.

14. **MDS International Congress 2016** (Berlin, DE; June 2016): Poster 1621. Digitally captured Archimedes spiral indices correlate with clinical assessment of dystonia severity J.B. Ratliff, A. Mirallave, **R. Ortega**, et al.
15. **MDS International Congress 2016** (Berlin, DE; June 2016): Poster 610. The distinguishing clinical features of rapid-onset parkinson-dystonia (RDP) syndrome due to ATP1A3 mutations I.A. Meijer, **R.A. Ortega**, D. Raymond, et al.
16. **MDS International Congress 2015** (San Diego, CA; June 2015): Poster 1052. Development of biomarker battery to discriminate LRRK2 mutation carriers. **RA Ortega**, R Alcalay, A Mirelman, C Pont Sunyer, C Wang, Q Yu, J Hagenah, D Raymond, A Glickman, M Pullman, B Johannes, N Doan, LJ Ozelius, S Pullman, K Marder, N Giladi, E Tolosa, SB Bressman, R Saunders-Pullman.
17. **MDS International Congress 2015** (San Diego, CA; June 2015): Poster 1031. Substantia nigra hyperechogenicity in LRRK2 Mutation carriers. A Drobnis, J Hagenah, A Glickman, **RA Ortega**, et al.
18. **MDS International Congress 2015** (San Diego, CA; June 2015): Poster 1297. Psychiatric features of GNAL and THAP1 mutation dystonia. E Deegan, A Glickman, H Sarva, **RA Ortega**, et al.
19. **MDS International Congress 2015** (San Diego, CA; June 2015): Poster 1306. Lack of an association between cancer and THAP1 mutation dystonia. A Glickman, RA Ortega, H Sarva, et al.
20. **AAN Annual Conference 2015** (Washington D.C.; April 2015): REM Sleep Behavior Disorder and G2019S PD – RBD, as Measured by RBD Sleep Questionnaires is not A Promising Early Marker. **RA Ortega**, R Alcalay, A Mirelman, C Wang, M San Luciano, D Raymond, A Glickman, B Johannes, H Mejia-Santana, K Marder, N Giladi, S Bressman, R Saunders-Pullman, LRRK2 AJ Consortium. \* Presented by R Saunders-Pullman.
21. **AAN Annual Conference 2015** (Washington D.C.; April 2015): Poster P2.149. Peripheral  $\beta$ -glucocerebrosidase enzyme activity and lipidomics in  $\beta$ -glucocerebrosidase (GBA1) mutation Parkinson disease.
22. **NYCEF Annual Meeting 2015** (Columbia School of Medicine; February 2015). Application of Item Response Theory to Olfaction in a Multinational Study of LRRK2 Mutations and Parkinson Disease. **RA Ortega**, A Glickman, A Mirelman, et al.
23. **ANA Annual Conference 2014** (Baltimore, MD; October 2014): Discriminative validity of digitized spiral analysis in LRRK2 Parkinson disease. **RA Ortega**, C Wang, M San Luciano, A Mirelman, C Pont Sunyer, Q Yu, M Pullman, Jeannie Soto-Valencia, K Marder, N Giladi, S Bressman, E Tolosa, S Pullman, R Saunders-Pullman, LRRK2 AJ Consortium
24. **MDS International Congress 2014** (Stockholm, Sweden; June 2014): Poster 1412. Olfactory Discrimination in GNAL mutation dystonia. **RA Ortega**, A Glickman, H Sarva, D Raymond, A Deik, T Fuchs, LJ Ozelius, SB Bressman, R Saunders-Pullman.
25. **AAN Annual Conference 2014** (Philadelphia, PA; April 2014): Poster P4.042. Glucocerebrosidase Enzyme Activity in *GBA1* Mutation Parkinson Disease. **Ortega R**, Torres P, Boschung S, Swan M, Soto-Valencia J, Barrett M, et al.
26. **AAN Annual Conference 2014** (Philadelphia, PA; April 2014): Poster P2.024. Neuropsychiatric features of GBA-associated PD. M Swan, **R Ortega**, M Barrett, et al.
27. **AAN Annual Conference 2013** (San Diego, CA; March 2013) Poster P05.058.

Progression of Motor Dysfunction in Elderly *LRRK2* Carriers, Including Prior to Phenocconversion to PD. **Ortega R**, Deik A, Battista J, Wang C, Ozelius L, Lipton R, et al.

### **Awarded Travel**

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Movement Disorder Society, Stockholm, Sweden, 2014. Selected for a travel award based on abstract merit

Movement Disorder Society, San Diego, CA, 2015. Travel award

American Neurological Association, Baltimore, Maryland, 2014. Travel award

### **Additional Experiences**

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**Academic:** Pathology Interest Group Committee Member, Echo Free Clinic Volunteer (Patient Labs), Project Kindness Volunteer

**Analytical:** Stata, R, Python, VBA, Excel. Experience with SAS, SPSS, Java, C++, Matlab

**Technical:** Phlebotomy, Transcranial sonography (ultrasound of the midbrain) - Trained by Dr. Johann Hagenah and Dr. Norbert Brüggemann (Westküstenklinikum Heide, Neurology, Heide, Germany and University Hospital Lübeck, Lübeck, Germany)