CURRICULUM-VITAE

PERSONAL INFORMATION

First Name: MACARENA
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QUALIFICATIONS OBTAINED

2002: Medical Doctor Degree. Universidad de Sevilla, Spain.

2007: Specialist in Neurology. Hospital Universitario de Valme, Sevilla, Spain.

2015: PhD. Universidad de Sevilla.

POSTGRADUATE TRAINING PROGRAMS

- Rio-Hortega program: 2007-2010: Training in research in Neuromuscular diseases
- Research fellowship at Mayo Clinic, Rochester, MN (USA): February 2009- April 2010
  Department of Neurology. Supervised by Dr. Andrew Engel

WORK HISTORY

- June 2003 –June 2007 : Resident in Neurology at Hospital de Valme, Sevilla
- June -September 2007: Consultant Neurologist at University Hospital Virgen de Valme, Sevilla. Department of Neurology. Full time
- September 2007 to present: Clinical collaborator. Dr. Fernandez-Chacón´s laboratory: "Physiology of Synapse" at IBiS (Biomedicine Institute of Seville), Hospital Virgen del Rocio, Seville, Spain.
- January 2008- January 2011: "Rio Hortega" Program. Supported by the Spanish Ministry of Health. Developed at Hospital Virgen del Rocio, Seville, Department of Neurology, Neuromuscular Unit, in collaboration with Dr. Fernandez-Chacón´s laboratory, IBIS. Included training in basic and clinical research plus part-time clinical work developed in general Neurology and Neuromuscular clinics.
  - Included a 14 months (February 2009-April 2010) fellowship at Mayo Clinic, (Rochester, MN, USA). Department of Neurology. Neuromuscular laboratory (Supervised by Dr. Andrew Engel)
- January 2011 to June 2011: Consultant Neurologist at University Hospital Virgen del Rocío, Sevilla. Department of Neurology. Full time
- January 2012 to January 2013: Consultant Neurologist at Clinica Universitaria de Navarra, Madrid. Full time
- January 2013 to September 2015: Research fellow at Harry Perkins Institute for Medical Research, under the supervision of Prof. Nigel Laing. University of Western Australia. Molecular Neurogenetics. Perth, WA, Australia.
- July 2013 to July 2014: Senior Registrar at Royal Perth Hospital. Department of Neurology. Neurogenetic unit.
- January 2015 to May 2015: Senior Registrar at Royal Perth Hospital. Department of Neurology.
- January 2016 to present: Consultant Neurologist and postdoctoral researcher (Juan Rodes position) at Hospital Virgen del Rocio and Instituto de Biomedicina de Sevilla, Sevilla, Spain.

Awards and Scholarships:

- Program "Rio-Hortega" (Spanish Instituto de Salud Carlos III): 2008-2011.

- Research scholarship "Alfonso Martin Escudero": 2013-2015
- Elsevier award to best poster presentation. World muscle society 2014
- James Hopley Award for the best primary research journal article: 2015
- Program "Juan Rodes" (Spanish Instituto de Salud Carlos III): 2016-2018

Research Stays Abroad

- Department of Neurology (Andrew Engel's lab), Mayo Clinic, Minnesota, USA. January 2009 to April 2010. Role: Research fellow
- University of Western Australia, Harry Perkins Institute of Medical Research. (Nigel Laing's lab). Perth, WA, Australia. January 2013 to September 2015. Role: PhD student
- University of Western Australia, Harry Perkins Institute of Medical Research. (Nigel Laing's lab). Perth, WA, Australia. January 2017 to May 2017. Role: Postdoctoral fellow

Clinical Trials:

- January to December 2007: "PERFORM" (Prevention of cerebrovascular and cardiovascular events of ischaemic origin with Terutroban in patients with a history of ischaemic stroke or transient ischaemic attack). Role: Collaborator.
- September 2016 to present: CLN-PXT3003-02. Role: Collaborator

Research Projects and Grants

- "Caracterización de variantes genéticas causantes de enfermedades neuromusculares en población gitana española"
  Date: 2017-2018
  Role: Principal Investigator
  Funding Agency: Fundacion Progreso y Salud. Junta de Andalucia
  Funding: 60.000 €
  Project ID: PI-0139-2016
  Resigned by the commencement date due to incompatibility with other awarded grant.

- "Creacion de base de datos de variantes geneticas en poblacion gitana para el diagnóstico y la prevención de enfermedades Neuromusculares" (Implementation of a database of genetic variants in gypsy population for the prevention of neuromuscular diseases)
  Date: 2017-2019
  Role: Principal Investigator
  Funding Agency: Instituto de Salud Carlos III (Spanish Ministry of Health)
  Funding: 86.515 €
  Project ID: PI16/00612

- "Mutaciones en protein O-glucosyltransferase 1 (POGLUT1) causan un nuevo tipo distrofia muscular asociada a hipoglicosilación de Notch y reducción en la expresión de alfa-distroglicano" (Mutations in POGLUT1 cause a new muscular dystrophy associated with hypoglycosilation of Notch and reduction of expression of alpha dystroglycan)
  Date: 2014-2015
  Role: Collaborator
  Funding Agency: Instituto de Salud Carlos III (Spanish Ministry of Health)
  Funding: 106.480 €
  Project ID: PI13/01739

- "Descripción de un nuevo fenotipo de distrofia muscular de cinturas autosómica recesiva e identificación del gen responsable mediante análisis genómico de última generación" (Description of a new phenotype of autosomic recessive limb girdle muscular dystrophy)
  Date: 2011-2013
  Role: Collaborator
  Funding Agency: Instituto de Salud Carlos III (Spanish Ministry of Health)
  Funding: 106.480 €
  Project ID: PI10/02410
- "Descripción de un nuevo fenotipo de síndrome miasténico congénito autosómico recesivo e identificación del gen responsable mediante análisis genómico de última generación." (Description of a Congenital Myasthenic Syndrome with a new phenotype and identification of causative mutations by NGS)
  Date: 2011-2012
  Role: Collaborator
  Funding Agency: Junta de Andalucía, Consejería de Salud. (Andalusian Govenment)
  Funding: 50.791,29 €
  Proyect ID: PI-0440-2010
- International Clinical Outcome Study of Dysferlinopathy
  Date: 2012-2015
  Role: Collaborator
  Funding Agency: Jain Foundation.
  Funding: per patient recruited
- Consorcio para generar una base de datos común para implementar la investigación básica y clínica en enfermedades neuromusculares. (Consortium to produce a common database to implement basic and clinic research in neuromuscular disorders)
  Date: 2011-2012.
  Funding Agency: CIBERNED.
  Role: Colaborador
  Funding: 286.000 €.

REGISTRATION HISTORY

2008- 2011**:** Colegio de médicos de Sevilla, Spain. (College of physicians of Seville, Spain)
2012- 2013: Colegio de médicos de Madrid, Spain. (College of physicians of Madrid, Spain)
July 2013 to July 2014 and January 2015 to May 2015: AHPRA/ Medical Board of Australia: Limited registration for post graduate training or supervised practice as a Medical Practitioner (Specialist pathway). Registration number MED0001833078.
January 2016 to present: Colegio de médicos de Sevilla, Spain. (College of physicians of Seville, Spain)

LANGUAGES

- Spanish: Mother tongue
- English:
  - Cambridge certificate: Advanced (CAE) June 2010
  - IELTS academic: June 2013 (Score 7.5)
- French: Basic level (A1)

PUBLICATIONS

1. Emilia Servián-Morilla, Hideyuki Takeuchi, Tom V Lee, Jordi Clarimon, Fabiola Mavillard, Estela Area-Gómez, Eloy Rivas, Jose L Nieto-González, Maria C Rivero, **Macarena Cabrera**, Leonardo Gómez-Sánchez, Jose A Martínez-López, Beatriz Estrada, Celedonio Márquez, Yolanda Morgado, Xavier Suárez-Calvet, Guillermo Pita, Anne Bigot, Eduard Gallardo, Rafael Fernández-Chacón, Michio Hirano, Robert S

Haltiwanger, Hamed Jafar-Nejad, Carmen Paradas. A POGLUT1 mutation causes a muscular dystrophy with reduced Notch signaling and satellite cell loss. Embo Molecular Medicine. In press.

2. **Macarena Cabrera-Serrano**, Jennifer E. Fugate, Jay Mandrekar and Alejandro Rabinstein. Impact of hyperglycemia in the outcome of patients with primary neuromuscular respiratory failure. Neurocritical Care. 2015. Aug;23(1):103-7

3. **Macarena Cabrera-Serrano**, Roula Ghaoui, Gianina Ravenscroft, et al. Expanding the phenotype of *GMPPB* mutations. Brain. 2015 Apr;138(Pt 4):836-44. doi: 10.1093/brain/awv013.

4. **Macarena Cabrera-Serrano**, Victoria A Fabian, Jordan Boutilier, et al. Adult onset distal and proximal myopathy with complete ophthalmoplegia associated with a novel de novo p.(Leu1877Pro) mutation in *MYH2*. Clin Genet. 2015 Dec;88(6):573-8. doi: 10.1111/cge.12552.

5. **Macarena Cabrera**. Miopatías metabólicas y tóxicas. Distrofias musculares. (Toxic and metabolic myopathies. Muscular dystrophies) [Review]. Medicine. 2015; 11 (75) 4516-27.

6. **Macarena Cabrera**. Protocolo clinico en Mialgias (Clinical protocol for the management of patients with myalgia). Medicine. 2015; 11 (75) 4540-4

7. **Macarena Cabrera-Serrano** , Junckerstorff RC, Atkinson V, Sivadorai P, Allcock RJ, Lamont P, Laing NG. Novel CHKB mutation expands the megaconial muscular dystrophy phenotype. Muscle Nerve. 2015 Jan;51(1):140-143. doi: 10.1002/mus.24446. Epub 2014 Nov 22.

8. Ohkawara B, **Cabrera-Serrano M**, Nakata T, et al. LRP4 third β-propeller domain mutations cause novel congenital myasthenia by compromising agrin-mediated MuSK signaling in a position-specific manner. Hum Mol Genet. 2014 Apr 1;23(7):1856-68

9. **Cabrera M**, Rabinstein A. Usefulness of pulmonary function tests and blood gases in acute neuromuscular respiratory failure. Eur J Neurol 2012;19(3):452-6.

10. **Cabrera M**, Rabinstein A. Neuromuscular diseases. In: Josephson, Freeman and Likosky. Neurohospitalist Medicine. Cambridge University Press, Cambridge 2011. ISBN 9780521172547.

11. **Cabrera M**, Rabinstein A. Causes and outcome of acute neuromuscular respiratory failure. Arch Neurol 2010 ;67(9):1089-1094

12. **Cabrera M**, Márquez C. Urgencias en patología neuromuscular (*Neuromuscular Emergencies*). In: Jesus Porta Etessan, Jose Antonio Olivan Usieto, Gemma Mas Sese et al. Manual Neurológico para el Manejo integral del paciente. Grupo Luzán 5, Madrid, 2009. ISBN:978-84-96989-03-0

13. **Cabrera M**, Paradas C, González A, Márquez C. Acute paraparesis following intravenous steroid therapy in a case of dural arteriovenous fistula. J Neurol. 2008;255(9):1432-3.

14. Paradas C, **Cabrera M**. Enfermedades Neuromusculares en el Anciano (*Neuromuscular disorders in the elderly*). In: Castilla L, Fernández C, Jiménez MD. Manual de Neurogeriatría. Grupo 2 Comunicación Médica SL. Madrid 2008; 345-358. ISBN: 978-84-612-2411-1

15. Galan J, **Cabrera M**. Epilepsia en el Anciano (*Epilepsy in the elderly*). In: Castilla L, Fernández C, Jiménez MD. Manual de Neurogeriatría. Grupo 2 Comunicación Médica SL. Madrid 2008; 189-200. ISBN: 978-84-612-2411-1

16. **Cabrera M**, Romero B, Del Río C, Romera M. Parálisis del III PC incompleta con afectación pupilar. In: IV Concurso de casos clínicos para residentes en neurología. Luzán 5 S.A. Ediciones. Madrid 2007; 161-3. ISBN: 978-84-96989-61-0

17. Romero B, **Cabrera M**, Del Rio C, Romera M. Cefalea y pérdida de sensibilidad en el hemicuerpo izquierdo de inicio brusco. En: IV Concurso de casos clínicos para residentes en neurología 2007. Madrid: Luzán 5 S.A., 2007; 151-3. ISBN: 978-84-96989-61-0

18. Del Rio C, **Cabrera M**, Romero B, Romera M. Disfonía y dificultad para la movilización lingual en un varón de 76 años de edad. En: IV Concurso de casos clínicos para residentes en neurología 2007. Madrid: Luzan 5 S.A. 2007;154-7. ISBN: 978-84-96989-61-0

19. Moniche F, Fernández C, **Cabrera M**. Cefalea explosiva durante la actividad sexual. En: Concurso de casos clínicos para residentes en neurología. Madrid: Luzan 5 S.A., 2004. ISBN: 84-7989-285-4

20. Fernández C, Moniche F, **Cabrera M**. Cefalea y enrojecimiento facial. En: Concurso de casos clínicos para residentes en neurología. Madrid: Luzan 5 S.A., 2004. ISBN: 84-7989-285-4

PARTICIPATION IN ELABORATION OF GUIDELINES

- Proceso diagnostico de ELA. In: Guia asistencial de Esclerosis Lateral Amiotrofica de la Junta de Andalucia (Clinical guidelines for management of ALS by Andalusian Government). Sevilla: Junta de Andalucia, 2012. ISBN: 978-84-923-8524-9

INVITED PRESENTATIONS

- Prevención de enfermedades neuromusculares genéticas mediante programas de detección de portadores en poblaciones de riesgo. [Preventing genetic neuromuscular diseases through carrier screening in high risk populations]. II Jornadas de Investigación Traslacional en Enfermedades Raras: últimos avances en enfermedades neuromusculares. Cordoba, Spain. November 2016
- Workshop for clinician researchers on exome sequencing. Australasian Neuromuscular Network. Auckland, New Zealand. April 2015.
- Aspectos clínicos de las neuropatías periféricas. (Diagnosis and management of peripheral neuropathies). Spanish Society of Neurology Annual Meeting. Barcelona, November 2012
- Topografía lesional de la diplopía: músculos extraoculares y unión neuromuscular. (M*uscle and neuromuscular junction diseases causing diplopia*) Spanish Society of Neurology Annual Meeting. Barcelona, November 2011

ABSTRACTS (last five years)

- **Macarena Cabrera-Serrano**, Gianina Ravenscroft, Erik Andersen, Dimitar N Azmanov, Catriona A McLean, Zornitza Stark, Mark R Davis, Andrew J Kornberg, Monique M Ryan, Nigel G Laing. Severe autosomal recessive congenital hypomyelinating neuropathy causing death in the first 4 months of life. World Muscle Society annual meeting. Granada, Spain. October 2016.
- R. Avila-Polo, E. Rivas, **M. Cabrera-Serrano**, [...], C. Paradas. Utrophin immunohistochemical expression in neuromuscular disorders. Granada, Spain. October 2016.
- Montse Olivé, Xesc Miralles, Juanjo Baiges, **Macarena Cabrera**, Gina Ravescroft, Nigel Laing, Dolores Moreno, Noemí Vidal. Adult-onset myopathy with characteristic inclusions and autophagic vacuoles. Granada, Spain. October 2016.
- P. Carbonell Corvillo · E. Servián-Morilla · E. Tristán Clavijo · **M. Cabrera** [...] · C. Paradas López. A novel MYH7 mutation causing the Laing distal myopathy in Andalucia. Granada, Spain. October 2016.
- **Macarena Cabrera-Serrano**, Roula Ghaoui, Gianina Ravenscroft, et al. Expanding the phenotype of GMPPB mutations. Australasian Neuromuscular Network. Auckland, New Zealand. April 2015.
- **Macarena Cabrera,** Roula Ghaoui, Phillipa Lamont, Nigel Clarke, Nigel Laing. Genetic defects in patients with rhabdomyolysis. Australasian Neuromuscular Network. Auckland, New Zealand. April 2015.
- **Macarena Cabrera**, Reimar Juckerstorff, Phillipa Lamont, Nigel Laing. Adult onset myopathy with complete ophthalmoplegia, dysphagia and distal muscle involvement due to de novo mutation in MYH2. World Muscle Society annual meeting. Berlin, Germany. October 2014.
- **Macarena Cabrera**, Merrilee Nedham, Phillipa Lamont, Nigel Laing. Clinical and molecular characterization of distal myopathies. World Muscle Society annual meeting. Berlin, Germany. October 2014.
- **Macarena Cabrera**, Roula Ghaoui, Dylan Mourdant, Phillipa Lamont, Nigel Clarke, Nigel Laing. Clinical and molecular characterization of rhabdomyolysis. World Muscle Society annual meeting. Berlin, Germany. October 2014.
- **Macarena Cabrera**, Roula Ghaoui, Dylan Mourdant, Phillipa Lamont, Nigel Laing. Genetic and clinical characterization of patients with repeated rhabdomyolysis. Australasian Neuromuscular Network annual meeting. Melbourne, April 2014.

- Jordi Díaz, Ricard Rojas, Carmen Paradas, **Macarena Cabrera** et al. Myasthenia Gravis database: A multicenter spanish Project. 12 International Conference on Myasthenia Gravis and Related Disorders. New York, USA. May 2012.
- **Cabrera M**, Morgado Y, Benitez S, Paradas C. Factors determining response to pyridostigmine or steroid treatment in myasthenia gravis. 63th American Academy of Neurology Annual Meeting. New Orleans, USA. April 2012
- Paradas C, **Cabrera M**, Rivas E. Neuromuscular involvement in choreoacanthocytosis. 63th American Academy of Neurology Annual Meeting. New Orleans, USA. April 2012
- C. Paradas, **M. Cabrera**, E. Rivas. Patología del sistema nervioso periférico en la neuroacantositosis. Spanish Society of Neurology Annual Meeting. Barcelona, November, 2011.
- Escudero, **M. Cabrera**, E. Rivas. Retracciones articulares como signo predominante en la dermatomiositis. Spanish Society of Neurology Annual Meeting. Barcelona, November, 2011
- L. Villarreal, **M. Cabrera**, E. Rivas. Hallazgos atípicos de la biopsia muscular en paciente con déficit de CPT II. Reunión Anual de la Sociedad Española de Neurología. Barcelona, Noviembre 2011.
- **Cabrera M**, Rabinstein A. Impact of Glycemic Control on Outcome of Critically Ill Neuromuscular Patients. 63th American Academy of Neurology Annual Meeting. Honolulu, Hawaii. 2011
- **Cabrera M**, Rabinstein A. Usefulness Of Pulmonary Function Tests And Blood Gases In Acute Neuromuscular Respiratory Failure. 63th American Academy of Neurology Annual Meeting. Honolulu, Hawaii. 2011
- Paradas C, **Cabrera M**, Márquez C. Natural History of Dysferlinopathies: Two Congenital patients Follow-up. 63th American Academy of Neurology Annual Meeting. Honolulu, Hawaii. 2011
- **Cabrera M,** Allan J, Young NP, Daube JR, Milone M. Cardiac Abnormalities in Myotonic Dystrophy Type 2. XII International Congress on Neuromuscular Diseases . Naples, Italy. 2010
- **Cabrera M**, Rabinstein AA. Causes and outcome of acute neuromuscular respiratory failure. XII International Congress on Neuromuscular Diseases . Naples, Italy. 2010
- Paradas C, Márquez C, **Cabrera M**, et al. 3q13.13-Q21.2: Nuevo Locus para una Distrofia Muscular de Cinturas Autosómica Recesiva con Rasgos Radiológicos Inusuales. Spanish Society of Neurology Annual Meeting. Barcelona, November, 2010
  Paradas C, **Cabrera M**, Márquez C, et al. Presentación Congénita de una Disferlinopatía en 2 Pacientes: Seguimiento Durante 8 Años. Andalusian Society of Neurology Annual Meeting. Melilla. November, 2010