Date of the CVA	

Section A. PERSONAL DATA

Name and Surname	José María Serratosa Fernández		
DNI		Age	
Researcher's identification number	Researcher ID		
	Scopus Author ID		
	ORCID		

A.1. Current professional situation

Institution	FUNDACION JIMENEZ DIAZ-UTE		
Dpt. / Centre	Neurología / Hospital Universitario Fundación Jiménez DíazNeurología		
Address	IIS-Fundación Jiménez Díaz, Avda Reyes Católicos, 2, 28040, Madrid		
Phone		Email	
Professional category	Jefe de Servicio	Start date	2008
UNESCO spec. code	249000 - Neurosciences		
Keywords	Medicine; Biomedicine; Molecular, cellular and genetic biology		

A.2. Academic education (Degrees, institutions, dates)

Bachelor/Master/PhD	University	Year
Medicina y Cirugía	Universidad Autónoma de Madrid	1993
Licenciado en Medicina y Cirugía	Universidad Autónoma de Madrid	1981

A.3. General quality indicators of scientific production

H Index 27

Main contribution: Discovered the locus of the first gene for Lafora Disease. Co-discoverer of the EPM2A gene, one of the two genes where mutations are found in Lafora Disease.

Section B. SUMMARY OF THE CURRICULUM

Dr José M Serratosa is a neurologist and epileptologist. He attended Medical School at the Autonomous University of Madrid and graduated in 1981 with Honors, after which he completed a residency training in Neurology at the Clínica Puerta de Hierro in Madrid. He specialized in epilepsy with the support of fellowships from the Epilepsy Foundation of America at the California Comprehensive Epilepsy Program of the University of California Los Angeles (UCLA) where he then worked as an assistant research neurologist. In 1993 he obtained his PhD degree from the Autonomous University of Madrid. Since 1995 he has been in charge of the Epilepsy Unit and the Genetic Epilepsies Laboratory at the Fundación Jiménez Díaz University Hospital in Madrid, Spain. He now is Chief of the Neurology Service and Director of the Eplilepsy Unit at the Fundación Jiménez Díaz University Hospital and Associate Professor at the School of Medicine of the Autonomous University of Madrid. His main research interests include the clinical and molecular genetics of the epilepsies (specially the idiopathic epilepsies, the epileptic encephalopathies and the progressive myoclonus epilepsies, mainly Lafora disease), the functional study of epilepsy gene products, and antiepileptic drug development. He heads a research lab funded with public funds and his group forms part of the Centre for Biomedical Network Research on Rare Diseases (CIBERER). He has published numerous papers and book chapters in the field of epilepsy, mainly on the genetics of the epilepsies. He has been a member of the editorial board of Epileptic Disorders and Seizure has been a member of the editorial board of Epilepsy Research and has served as Associate Editor for Genetics of

Epilepsia. He has also served as a member of the Genetics Commission of the International League Against Epilepsy (ILAE).

Research in my laboratory is aimed at the definition of epilepsy phenotypes with a genetic genetic base and to the identification of genes responsible for different types of epilepsy. We have studied the clinical characteristics and the molecular genetics since 1995. In 1995 working in the laboratory of Dr Antonio Delgado-Escueta I mapped the first gene for Lafora disease to chromosome 6q. In 1998 my group at the Fundación Jimenez Diaz in Madrid identified the 6q Lafora disease gene (EPM2A). We have worked in the characterization of the phenotype of the human phenotype as well as the mouse model of Lafora disease and have published extensively in Lafora disease and have several papers on the behavioral and epilepsy characteristics of the epm2a and epm2b mouse models. We have also contributed to the identification of numerous epilepsy genes responsible for familial epilepsies as well as epileptic encephalopathies through scientific collaborations. Our work has been recognized by the scientific community via invitations for review articles, lectures at national/international symposia, and requests to serve on grant review committees. I have directed 8 PhD thesis and have been invited speaker to numerous international congresses (World Congress of Neurology, European Academy of Neurology, International an European Epilepsy Congress) and Universities at public science forums (Europe, USA and Canada).

Section C. MOST RELEVANT MERITS (ordered by typology)

C.1. Publications

1 **Scientific paper**. Berthier, A.; et al. 2016. Pharmacological Interventions to Ameliorate Neuropathological Symptoms in a Mouse Model of Lafora Disease.Molecular neurobiology. 53-2, pp.1296-1309. ISSN 1559-1182.
2 **Scientific paper**. Giráldez, BG.; et al. 2015. Uniparental disomy as a cause of spinal muscular atrophy and progressive myoclonic epilepsy: phenotypic homogeneity due to the homozygous c.125C>T mutation in ASAH1.Neuromuscular disorders : NMD. 25-3, pp.222-226. ISSN 1873-2364.
3 **Scientific paper**. Larsen, J.; et al. 2015. The phenotypic spectrum of SCN8A encephalopathy.Neurology. 84-5, pp.480-489. ISSN 1526-632X.
4 **Scientific paper**. Ferlazzo, E.; et al. 2014. Mild Lafora disease: clinical, neurophysiologic, and genetic findings.Epilepsia. 55-12, pp.e129-e133. ISSN 1528-1167.
5 **Scientific paper**. uroEPINOMICS-RES Consortium; Epilepsy Phenome/Genome Project; Epi4K Consortium.2014. De novo mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies.American journal of human genetics. 95-4, pp.360-370. ISSN 1537-6605.
6 **Scientific paper**. Nava, C.; et al. 2014. De novo mutations in HCN1 cause early infantile epileptic encephalopathy.Nature genetics. 46-6, pp.640-645. ISSN 1546-1718.
7 **Scientific paper**. Lemke, JR.; et al. 2013. Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes.Nature genetics. 45-9, pp.1067-1072. ISSN 1546-1718.
8 **Scientific paper**. Dibbens, LM.; et al. 2013. Mutations in DEPDC5 cause familial focal epilepsy with variable foci.Nature genetics. 45-5, pp.546-551. ISSN 1546-1718.
9 **Scientific paper**. Criado, O.; et al. 2012. Lafora bodies and neurological defects in malin-deficient mice correlate with impaired autophagy.Human molecular genetics. 21-7, pp.1521-1533. ISSN 1460-2083.
10 **Scientific paper**. May. 2018. Rare coding variants in genes encoding GABA(A) receptors in genetic generalised epilepsies: an exome-based case-control study Lancet Neurology. Lancet. 17-8, pp.699-708-708.
11 **Scientific paper**. Bailey JN. 2018. Variant Intestinal-Cell Kinase in Juvenile Myoclonic Epilepsy New England Journal of Medicine. 378-11, pp.1018-1028.
12 **Scientific paper**. Ortega-Moreno L. 2017. Molecular diagnosis of patients with epilepsy and developmental delay using a customized panel of epilepsy genes.PLoS One. 12-11, pp.e0188978.
13 **Scientific paper**. 2017. Clinical spectrum and genotype – phenotype associations of KCNA2 -related encephalopathies Brain. pp.1-18.

14 **Scientific paper**. Villalba Orero, M.; et al. 2017. Lafora Disease Is an Inherited Metabolic Cardiomyopathy.Journal of the American College of Cardiology. 69-24, pp.3007-3009. ISSN 1558-3597.
15 **Scientific paper**. 2017. Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data.European journal of human genetics : EJHG. 25-7, pp.894-899. ISSN 1476-5438.
16 **Scientific paper**. Gómez Ibáñez, A.; et al. 2017. Efficacy and safety of eslicarbazepine-acetate in elderly patients with focal epilepsy: Case series.Seizure. 48, pp.53-56. ISSN 1532-2688.
17 **Scientific paper**. Sánchez Elexpuru, G.; et al. 2017. 4-Phenylbutyric acid and metformin decrease sensitivity to pentylenetetrazol-induced seizures in a malin knockout model of Lafora disease.Neuroreport. 28-5, pp.268-271. ISSN 1473-558X.
18 **Scientific paper**. Sánchez Elexpuru, G.; Serratosa, JM.; Sánchez, MP.2017. Sodium selenate treatment improves symptoms and seizure susceptibility in a malin-deficient mouse model of Lafora disease.Epilepsia. 58-3, pp.467-475. ISSN 1528-1167.
19 **Scientific paper**. 2017. De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies.American journal of human genetics. 100-1, pp.179. ISSN 1537-6605.
20 **Scientific paper**. Ortega Moreno, L.; et al. 2016. Novel mutation in STXBP1 gene in a patient with non-lesional Ohtahara syndrome.Neurologia (Barcelona, Spain). 31-8, pp.523-527. ISSN 1578-1968.
21 **Scientific paper**. Villanueva, V.; et al. 2016. Safety, efficacy and outcome-related factors of perampanel over 12 months in a real-world setting: The FYDATA study.Epilepsy research. 126, pp.201-210. ISSN 1872-6844.
22 **Scientific paper**. Hardies, K.; et al. 2016. Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline.Brain : a journal of neurology. 139-Pt 9, pp.2420-2430. ISSN 1460-2156.
23 **Scientific paper**. de Kovel, CG.; et al. 2016. Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients.Molecular genetics & genomic medicine. 4-5, pp.568-580. ISSN 2324-9269.
24 **Scientific paper**. Djémié, T.; et al. 2016. Pitfalls in genetic testing: the story of missed SCN1A mutations.Molecular genetics & genomic medicine. 4-4, pp.457-464. ISSN 2324-9269.
25 **Scientific paper**. Giráldez, BG.; Serratosa, JM.2015. Jeavons syndrome as an occipital cortex initiated generalized epilepsy: Further evidence from a patient with a photic-induced occipital seizure.Seizure. 32, pp.72-74. ISSN 1532-2688.
26 **Scientific paper**. Giráldez, BG.; et al. 2015. Long-term efficacy and safety of lacosamide monotherapy in the treatment of partial-onset seizures: A multicenter evaluation.Seizure. 29, pp.119-122. ISSN 1532-2688.
27 **Scientific paper**. Hardies, K.; et al. 2015. Recessive loss-of-function mutations in AP4S1 cause mild fever-sensitive seizures, developmental delay and spastic paraplegia through loss of AP-4 complex assembly.Human molecular genetics. 24-8, pp.2218-2227. ISSN 1460-2083.
28 **Scientific paper**. Syrbe, S.; et al. 2015. De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy.Nature genetics. 47-4, pp.393-399. ISSN 1546-1718.
29 **Scientific paper**. Striano, P.; et al. 2012. GLUT1 mutations are a rare cause of familial idiopathic generalized epilepsy.Neurology. 78-8, pp.557-619. ISSN 1526-632X.
30 **Review**. Gentry M. 2018. Lafora disease offers a unique window into neuronal glycogen metabolism J Biol Chem. 293-19, pp.7117-7125.
31 **Review**. 2018. Tau-Induced Pathology in Epilepsy and Dementia: Notions from Patients and Animal Models Int J Mol Sci. 19-4, pp.E1092..

C.2. Participation in R&D and Innovation projects

1 Lafora Epilepsy - Basic mechanisms to therapy (National Institute of Health). 01/07/2016-30/06/2021. 8.000.000 €.

2 Genética de las epilepsias humanas: Identificación de nuevos genes, diagnóstico precoz y su utilidad clínica. Ministerio de Educación y Ciencia SAF2014-59594-R. Retos de la Sociedad. José M Serratosa. (Instituto de Investigaciones Sanitarias-Fundación Jiménez Díaz). 01/01/2015-31/12/2017. 145,2 €. Principal investigator.

3 Genética de las epilepsias humanas: Hacia el diagnóstico precoz y la terapia personalizada Ministerio de Educación y Ciencia. Retos de la Sociedad. Jose Serratosa Fernandez. (Instituto de Investigaciones Sanitarias-Fundación Jiménez Díaz). 01/01/2014-31/12/2015. 60 €. Principal investigator.

4 EUI-EURC-2011-4325, Genética de los síndromes epilépticos raros Ministerio de Educación y Ciencia. EUROCORES. Jose Serratosa Fernandez. (Instituto de Investigaciones Sanitarias-Fundación Jiménez Díaz). 01/01/2012-01/04/2014. 96.000 €. Principal investigator.

5 Proyecto Integrado: Functional genomics and neurobiology of epilepsy: a basis for new therapeutic strategies Unión Europea FP6 LSHM-CT-2006-037315. Jose Serratosa Fernandez. (Fundación Jiménez Díaz). 01/01/2007-31/12/2010. 159.000 €. Principal investigator.

6 SAF2007-61003, Genética molecular de las epilepsias humanas Ministerio de Educación y Ciencia. Plan Nacional. José M Serratosa. (FUNDACION JIMENEZ DIAZ-UTE). 01/10/2007-04/10/2010. 346.000 €. Principal investigator.

7 Identificación de genes moduladores o modificadores del fenotipo de la enfermedad de Lafora en familias con enfermedad de Lafora asociada a mutaciones en los genes EPM2A y EPM2B" Mutua Madrileña. Pilar Gómez-Garre.From 2007. 31.500 €.

8 Red CIEN Instituto de Salud Carlos III. From 2005.

9 SAF2004-07151, Caracterización de genes responsables de las epilepsias idiopáticas Ministerio de Educación y Ciencia. José M Serratosa. (Fundación Jiménez Díaz). From 2004. 138.000 €.

10 Identificación de otros genes implicados en la enfermedad de Lafora. FIS (PI020536).. José M Serratosa.From 2002. 12.000.000 €.

11 Localización e identificación del segundo gen implicado en la enfermedad de Lafora. José M Serratosa LINEA: Caracterización de un gen implicado en las epilepsias idiopáticas generaliz. From 2001. 10.000.000 €.

12 Caracterización del gen de la epilepsia mioclónica progresiva de Lafora [enfermedad de Lafora]. Comisión Interministerial de Ciencia y Tecnología (SAF99-0013-C02-02). José M Serratosa.From 1999.

13 Caracterización del gen de la epilepsia mioclónica progresiva de Lafora [enfermedad de Lafora]. José M Serratosa.From 1999.

14 Localización de genes implicados en la epilepsia con ausencias en la infancia. Comisión Interministerial de Ciencia y Tecnología (SAF97-0201). José M Serratosa.From 1997.

15 Identificación y caracterización del gen de la epilepsia mioclónica progresiva de Lafora: construcción de una cartografía física de alta resolución y refinamiento de la región del gen. Comisión Interministerial de Ciencia y Tecnología (SAF96-0318). José M Serratosa.From 1996.

C.3. Participation in R&D and Innovation contracts

C.4. Patents