PERSONAL INFORMATION

Sara Baldassari

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Sex Female | Date of birth 05/05/1988 | Nationality Italian

WORK EXPERIENCE

June 2017 – June 2019

Post-doc
ICM Institute, Paris, France
Genetics and physiopathology of focal epilepsies

May 2016 – May 2017

Co.Co.Co.

IRCCS - Istituto delle Scienze Neurologiche, Bologna, Italy

Targeted NGS in epilepsy: library preparation using Nextera enrichment (Illumina) and data analyses.

June 2014 – May 2016

Fellowship

Telethon Foundation

Analysis of WES data in patients with SHE, validation of the results and functional assays in the context of theTelethon Foundation project GGP13200, titled: “In-depth clinical and genetic study of familial and sporadic patients with nocturnal frontal lobe epilepsy (NFLE): identification of new genes by wes in 192 cases negative for mutations in the neuronal nicotinic acetylcholine receptor subunits genes”

June 2013 – June 2014

Fellowship

Sant’Orsola-Malpighi Hospital, Bologna, Italy

Analysis of WES data in patients with thrombocythemia and limb defects in the context of the Italian Ministry of Health project gr-2010-2318960.

EDUCATION AND TRAINING

January 2013 – January 2017

PhD in Biomedical Sciences and Oncology – curriculum in Human Genetics

University of Turin, Italy

Title of the final dissertation: Identification of novel candidate focal epilepsy genes by whole exome sequencing.

2010-2012

Master degree in Molecular and Cellular Biology (final degree mark: 110/110 cum laude)

University of Bologna, Italy

Title of the final dissertation: Study of the genetic basis of a form of focal epilepsy and of the familial cortical myoclonic tremor with epilepsy.

2007-2010

Bachelor degree in Biological Sciences (final degree mark: 110/110 cum laude)

University of Bologna, Italy

Title of the final dissertation: Breakpoint mapping of a translocation using FISH.

PERSONAL SKILLS

Mother tongue(s) Italian

Other language(s)

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken interaction	Spoken production	
English	B2	B2	B2	B2	B2

Communication skills Good communication skills gained during my PhD, through the presentation of my work to other PhD students and Professors, but also during journal clubs.

Organisational / managerial skills Good organizational skills acquired during my PhD: I have been involved in the training of various first and master degree students during their internship at the Medical Genetics Unit, Sant'Orsola-Malpighi Hospital, Bologna, Italy.

Job-related skills DNA extraction from various tissues, RNA extraction from whole blood or cell cultures, fluorescent in situ hybridization on metaphasic chromosomes, real-time PCR on both gDNA and cDNA using TaqMan method, Sanger sequencing, linkage analysis using microsatellite markers, use of optic and fluorescent microscope, DNA amplification using commercial kits, MLPA, cell cultures (mainly: fibroblasts, lymphoblasts and E.Coli), protein extraction from cell cultures, western blot, few experiences with cloning. NGS library preparation using the Nextera enrichment protocol (Illumina). Basic bioinformatic skills for the analysis of NGS data and linux operative system.

Digital competence

SELF-ASSESSMENT				
Information processing	Communication	Content creation	Safety	Problem solving
Basic user	Basic user	Basic user	Basic user	Basic user

good command of office suite (word processor, spread sheet, presentation software)

Driving licence B

Publications

- **Baldassari S**, Licchetta L, Tinuper P, Bisulli F, Pippucci T. GATOR1 complex: the common genetic actor in focal epilepsies. J Med Genet. 2016 May 19. pii: jmedgenet-2016-103883.
- Licchetta L, Pippucci T, Bisulli F, Cantalupo G, Magini P, Alvisi L, **Baldassari S**, Martinelli P, Naldi I, Vanni N, Liguori R, Seri M, Tinuper P. A novel pedigree with familial cortical myoclonic tremor and epilepsy (FCMTE): clinical characterization, refinement of the FCMTE2 locus, and confirmation of a founder haplotype. Epilepsia. 2013 Jul;54(7):1298-306.
- Bisulli F, Naldi I, **Baldassari S**, Magini P, Licchetta L, Castegnaro G, Fabbri M, Stipa C, Ferrari, S, Seri M, Goncalves Silva GE, Tinuper P, Pippucci T. Autosomal dominant partial epilepsy with auditory features: Anew locus on chromosome 19q13.11-q13.31. Epilepsia. 2014 Mar 1.
- Magini P, Bisulli F, **Baldassari S**, Stipa C, Naldi I, Licchetta L, Menghi V, Tinuper P, Seri M, Pippucci T. LGI1 microdeletions are not a frequent cause of partial epilepsy with auditory features (PEAF). Epilepsy Res. 2014 Mar 26. pii:S0920-1211(14)00079-5.
- Pippucci T, Licchetta L, **Baldassari S**, Palombo F, Menghi V, D'Aurizio R, Leta C, Stipa C, Boero G, d'Orsi G, Magi A, Scheffer I, Seri M, Tinuper P and Bisulli F. Epilepsy with auditory features: a heterogenous clinico-molecular disease. Neurol Genet June 2015 vol. 1 no. 1 e5
- Ricos MG, Hodgson BL, Pippucci T, Saidin A, Sze Ong Y, Heron SE, Licchetta L, Bisulli F, Bayly MA, Hughes J, **Baldassari S**, Palombo F; Epilepsy Electroclinical Study Group, Santucci M, Meletti S, Berkovic SF, Rubboli G, Thomas PQ, Scheffer IE, Tinuper P, Geoghegan J, Schreiber AW, Dibbens LM. Mutations in the mTOR pathway regulators NPRL2 and NPRL3 cause focal epilepsy. Ann Neurol. 2015 Oct 27.
- Hildebrand MS, Tankard R, Gazina EV, Damiano JA, Lawrence KM, Dahl HH, Regan BM, Shearer AE, Smith RJ, Marini C, Guerrini R, Labate A, Gambardella A, Tinuper P, Lichetta L, **Baldassari S**, Bisulli F, Pippucci T, Scheffer IE, Reid CA, Petrou S, Bahlo M, Berkovic SF. PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. Ann Clin Transl Neurol. 2015 Aug;2(8):821-30.
- Bisulli F, Licchetta L, **Baldassari S**, Pippucci T, Tinuper P. DEPDC5 mutations in epilepsy with auditory features. Epilepsia. 2016 Feb;57(2):335.
- Neri I, Virdi A, Tortora G, **Baldassari S**, Seri M, Patrizi A. Novel p.Glu519Gln missense mutation in ST14 in a patient with ichthyosis, follicular atrophoderma and hypotrichosis and review of the literature. J Dermatol Sci. 2016 Jan;81(1):63-6.
- Henden L, Freytag S, Afawi Z, **Baldassari S**, Berkovic SF, Bisulli F, Canafoglia L, Casari G, Crompton DE, Depienne C, Gecz J, Guerrini R, Helbig I, Hirsch E, Keren B, Klein KM, Labauge P, LeGuern E, Licchetta L, Mei D, Nava C, Pippucci T, Rudolf G, Scheffer IE, Striano P, Tinuper P, Zara F, Corbett M, Bahlo M. Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2-2q11.2. Hum Genet. 2016 Jul 1.

Presentations

- SIGU conference, Rimini, 2015
  "GATOR1 complex mutations are implicated in focal epilepsy pathogenesis"
  Baldassari S, Licchetta L, Marconi C, Palombo F, Magini P, Seri M, LICE NFLE Study group, Tinuper P, Bisulli F, Pippucci T

Posters at congresses

- SIGU conference, Turin, 2016
  The contribution of de novo coding mutations to the pathogenesis of Focal Epilepsy
  Baldassari S, Licchetta L, Palombo F, NFLE LICE Study Group, Seri M, Bisulli F, Tinuper P, Pippucci T
- ESHG Congress, Barcelona 2016
  "Mutations of the mTORC1-regulating complex GATOR1 in focal epilepsies"
  Baldassari S, Licchetta L, Marconi C, Myers CT, Palombo F, Magini P, Mefford HC, Seri M, NFLE Lice Study Group*, Tinuper P, Bisulli F, Pippucci T
- ESHG Congress, Milan 2014
  "A novel missense mutation in ST14 in a patient with ichthyosis, follicular atrophoderma and hypotrichosis"
  Baldassari S, Tortora G, Balestri R, Neri I, Seri M
- SIGU conference, Rome, 2013
  "Autosomal Dominant Partial Epilepsy with Auditory Features: a novel locus maps to chromosome 19q13.11-q13.31"
  Baldassari S, Bisulli F, Naldi I, Magini P, Licchetta L, Castegnaro G, Fabbri M, Stipa C, Ferrari S, Seri M, Gonçalves Silva GE, Tinuper P, Pippucci T