





## Pepe Sara, Ph.D student

Address: [REDACTED]

Mobile phone: [REDACTED]

E-mail: [REDACTED]

### SUMMARY

Medical/farmaceutical biotechnologist with background in cellular and molecular neurobiology. Expertise in synaptic physiology and in preclinical research in mouse models of neurodevelopmental disorders.

### PROFESSIONAL EXPERIENCE

- **November 2019 – present (Ph.D student)**  
*Department of Experimental Medicine (DIMES)*, University of Genoa, Genoa, Italy.  
PhD course in Neuroscience – curriculum: Clinical and experimental neurosciences (DINOEMI, University of Genoa, Italy). Supervisor: Prof.ssa Anna Fassio.  
Project title: “Role of the epilepsy genes ATP6V1A and TBC1D24 in pH homeostasis and autophagy progression”.
- **April 2019 – October 2019 (Fellow)**  
*Center for Synaptic Neuroscience and Technology, Italian Institute of Technology (INSYN-IIT)*, Genoa, Italy.  
Fellowship in “Neuroscience and Brain Technologies”. Supervisors: Prof. Fabio Benfenati  
Project: “Measuring the functionality of the endocytic pathway in neuronal cells by using a fluorescent probe as a sensor of intraorganellar pH”.
- **March 2018 – March 2019 (M.Sc. Student)**  
*Department of Experimental Medicine (DIMES)*, University of Genoa, Genoa, Italy.  
Experimental thesis in Neurophysiology. Supervisor: Prof.ssa Anna Fassio.  
Project title: “The disease gene TBC1D24: novel interactors and functional implications”.
- **Sept 2015 – Dec 2016 (B.Sc. Student)**  
*Department of Experimental Medicine (DIMES)*, University of Genoa, Genoa, Italy.  
Experimental thesis in Neurophysiology. Supervisor: Prof.ssa Anna Fassio.  
Project title: “Role of TBC1D24 into the axonal and dendritic specification”.

### EDUCATION

- **2016 – 2019:** Master's degree in Medical-pharmaceutical biotechnology with the grades 110/110 *cum laude* at the University of Genoa, Genoa, Italy.

- **2013 – 2016:** Bachelor's degree in Biotechnology with the grades 110/110 *cum laude* at the University of Genoa, Genoa, Italy.
- **2008 – 2013:** High School Diploma with the grades 100/100 at Liceo Scientifico – indirizzo linguistico "A.Righi" of Cesena, Italy.

### TECHNICAL SKILLS

- **Cell cultures:** dissections for primary mouse and rat cultures (neurons and astrocytes), maintenance of mouse/rat cortical and hippocampal neurons and cell lines (HEK293T, fibroblasts and astrocytes), lentiviral production, lentiviral transduction of neurons and cell lines, transfection of neurons and cell lines by calcium phosphate, nucleofection and liposomes;
- **Molecular and cellular biology:** purification and analysis of protein samples by SDS PAGE and western blot, pulldown, co-immunoprecipitation, subcellular fractionation, DNA extraction, DNA amplification (maxi and miniprep, PCR) and purification, restriction enzymes digestion, agarose gel electrophoresis;
- **Microscopy and Live imaging experiments:** immunofluorescence stainings (estimation of protein expression, localization and colocalization), morphological analysis of neurons (sholl analysis, synaptic protein expression and synapse counting), lysosomal functional assays (LysoTracker and LysoSensor evaluation), synaptic vesicles cycling using synaptophysin-pHluorin tool. Expertise in light, epifluorescence and confocal microscopy;
- **Animal manipulation:** expertise in mouse handling and genotyping with murine models of synaptopathies.

### SOFT SKILLS

- **Mentorship skills:** supervising of 3 undergraduate students for the preparation of the experimental thesis in biotechnology and biology;
- **Collaboration skills:** collaboration with internal colleagues (DIMES and DIBRIS, University of Genoa, Genoa, Italy; Center for Synaptic Neuroscience and Technology, Italian Institute of Technology, Genoa, Italy), collaboration with external scientists nationally (Children's Hospital "A. Meyer", University of Florence, Florence, Italy) and internationally (MRC Harwell Institute, Harwell Campus, Oxfordshire, UK);
- **Project management skills:** following and planning more projects in the same time, ability to work under pressure and to complete tight deadlines.

### SOFTWARE SKILLS

- Data analysis & statistical software (MS Excel, GraphPad Prism)
- Graphic software (ImageJ, ImageLab, Adobe Photoshop)
- Molecular biology softwares (SnapGene, ApE)

## LANGUAGE SKILLS

Italian: Mother-tounge

English: Fluent (written and spoken)

French: Good (written and spoken)

Spanish: Fluent (written and spoken)

## CONFERENCES and POSTERS

1. **"TBC1D24 interacts with vATPase and regulates pH homeostasis and autophagy in neurons"** ORAL PRESENTATION at SIF YRP 2022. 13-15 July 2022. Bertinoro, Italy
2. **Pepe S.,** Aprile D., Castroflorio E., Parsons A., Soares T., Benfenati F., Oliver P. and Fassio A. "TBC1D24 interacts with v-ATPase and regulates pH homeostasis and autophagy in neurons". POSTER PRESENTATION at FENS FORUM 2022. 9-13 July 2022. Paris, France
3. **Pepe S.,** Aprile D., Castroflorio E., Parsons A., Soares T., Benfenati F., Oliver P. and Fassio A. "TBC1D24 interacts with v-ATPase and regulates pH homeostasis and autophagy in neurons". POSTER PRESENTATION at EMBO Workshop "Autophagy in brain health and disease". 11-14 May 2022. Sant Feliu de Guixols, Spain;
4. Esposito A., **Pepe S.,** Cerullo M.S., Cortese K., Maragliano L., Guerrini R., Benfenati F. and Fassio A. POSTER at IRCCS Ospedale Policlinico San Martino Scientific Retreat. 4-5 November 2021. Genoa, Italy;
5. **Pepe S.,** Maragliano L., Conti V., Mei D., Benfenati F., Guerrini R. and Fassio A. "Structure/function study on de novo mutations in ATP6V1A causing developmental encephalopathy with epilepsy." POSTER PRESENTATION at BraYn Conference. 25-26 November 2020 (web conference);
6. Esposito A., Falace A., Mei D., Conti V., **Pepe S.,** Giovedì S., Benfenati F., Guerrini R. and Fassio A. "ATP6V1A and DMXL2 loss of function impacts on lysosomal homeostasis and autophagy causing developmental and synaptic defects in neurons." POSTER at FENS 2020. 11-15 July 2020. Glasgow, Scotland (web conference).

## PUBLICATIONS

1. Guerrini R., Mei D., Kerti-Szigeti K., **Pepe S.** et al. "Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis." *Brain* 2022, 145(8):2687-2703. doi: 10.1093/brain/awac145. IF: 15.255
2. Di Lisa D., Muzzi L., **Pepe S.** et al., "On the way back from 3D to 2D: Chitosan promotes adhesion and development of neuronal networks onto culture supports." *Carbohydrate Polymers* 2022, Vol.297. doi: 10.1016/j.carbpol.2022.120049. IF: 10.723
3. Muzzi L., Di Lisa D., Falappa M., **Pepe S.** et al., "Human Excitatory Cortical Neurospheroids Coupled to High-Density MEAs: A Valid Platform for Functional Tests." (preprint on SSRN)