







## Postdoctoral Position: A Drug Approach to Salla Disease

The laboratory is looking for a biochemist/cell biologist to explore pharmacological approaches to treat a rare disease severely affecting neurological development of children.

Salla disease is caused by mutations of the *SLC17A5* gene, leading to trafficking and/or activity defects of sialin, the lysosomal exporter for sialic acids. The predominant mutation, R39C, partially impairs both processes. The main project consists in identifying drugs that rescue the trafficking defect of mutant sialin and could also potentiate its transport activity.

The candidate is expected to have expertise in biochemistry, cell biology, fluorescence microscopy, and should be familiar with cell culture, molecular biology and molecular pharmacology techniques.

The laboratory, located at Université Paris Cité, Campus Saint-Germain-des-Prés, France, focuses on membrane trafficking and membrane transport processes, particularly in the lysosome, that shape the maintenance and adaptive homeostasis of mammalian cells. We use multidisciplinary approaches (https://www.sppin.fr/the-teams/team-4-membrane-dynamics/).

The postdoc position is funded by the Salla Treatment and Research Foundation (S.T.A.R.) Foundation, <a href="https://www.sallaresearch.org/">https://www.sallaresearch.org/</a>. The recruited postdoc will interact with an international research consortium, the FSASD consortium.

Motivated individuals interested in this work are encouraged to contact to Dr. Bruno Gasnier (<a href="mailto:bruno.gasnier@u-paris.fr">bruno.gasnier@u-paris.fr</a>) and Dr. Christine Anne-Longin (<a href="mailto:christine.anne-longin@u-paris.fr">christine.anne-longin@u-paris.fr</a>). Please include your CV (resume) and a brief research summary and have three letters of recommendation emailed to Drs. Gasnier and Anne-Longin.

## **Relevant References:**

- Huizing M, Hackbarth ME, Adams DR, Wasserstein M, Patterson MC, Walkley SU, Gahl WA, FSASD
  Consortium, Free sialic acid storage disorder: Progress and promise. Neurosci Lett, 2021.
   <a href="https://doi.org/10.1016/j.neulet.2021.135896">https://doi.org/10.1016/j.neulet.2021.135896</a>
- Dubois L, Pietrancosta N, Cabaye A, Fanget I, Debacker C, Gilormini PA, Dansette PM, Dairou J, Biot C, Froissart R, Goupil-Lamy A, Bertrand HO, Acher FC, McCort-Tranchepain I, Gasnier B, Anne C, Amino Acids Bearing Aromatic or Heteroaromatic Substituents as a New Class of Ligands for the Lysosomal Sialic Acid Transporter Sialin. J Med Chem, 2020.
   <a href="https://doi.org/10.1021/acs.jmedchem.9b02119">https://doi.org/10.1021/acs.jmedchem.9b02119</a>