Principal Investigator: Gianluigi Zanusso

Project Title: Multi-center assessment of olfactory swab procedure performed by non-otolaryngologists for improving and simplifying human prion disease diagnosis

Summary: This is a multicenter collaborative study involving four units with expertise in prion disease: the University of Verona, Verona, Italy; the University of Göttingen, Germany; the Paris Brain Institute, Paris, France; and the Istituto Superiore di Sanità, Rome, Italy. Olfactory mucosa had been collected from a large number of patients affected with prion diseases using a nasal swab (same as that of a COVID-19 test) by non-specialists health care professional without the use of an endoscope

Results: Olfactory swab (OS) testing provided a prion disease diagnosis in 90% of patients, comparable to cerebrospinal fluid (CSF) testing.
The diagnosis of prion disorder was provided in nearly 100% of patients in whom both OS and CSF were available. In contrast, all patients who were RT-QuIC negative in both CSF and OS received non-prion disease diagnoses.
The RT-QuIC assay detected prions in OS with high fidelity among laboratories

Conclusions: OS is the simplest procedure for diagnosing prion disease, and RT-QuIC is a highly reliable assay. The time lag for the diagnosis becomes short, and the number of investigations is reduced. Prompt communication of prion disease diagnosis alleviates family anguish and offers a potential therapeutic window in the early stages of the disease.