Isaac Maximiliano Bugueno

Project title: *Comparison of rare disease patients' clinical pathways within the ERN oro-dental expert centers*

Duration	1 month
Short Bio	After obtaining my Bachelor and a Doctor of Dental Surgery degree in 2014, throughout my first postgraduate years, I worked as clinician, but also as a researcher at the University of Strasbourg in France. I earned my master's degree in physiopathology and then my PhD in immunology in 2019. My research was first centered on a ground-breaking study that aimed to show the physiopathological mechanisms involved in the destruction of the periodontium and in their interaction with systemic pathologies, through various molecular immune pathways. My experience in research and background in dentistry allowed me to refocus on the areas of human genetics and oro-craniofacial development in 2020. Since then, I have been party to several projects at the Translational Medicine and Neurogenetics department of the IGBMC and the Reference Center for Oro-dental rare diseases in Strasbourg. We carry out translational research on rare diseases with isolated or syndromic oro-dental manifestations through multidisciplinary and transversal approaches in order to optimize the diagnosis and the care of patients with oro-dental rare disorders. I currently work as lecturer and researcher in oro-cranio-facial development and oro-dental associated genetic disorders.
Home Institution	Reference Center for Rare Oral and Dental Diseases (CRMR-O-Rares), University Hospitals of Strasbourg.
Host institution	RadboudUMC - Radboud Universitair Medisch Centrum - The Radboudumc Craniofacial Team at the Dental School
Project description	Odontogenesis genetic-driven alterations at different stages, as in mineralization of the dentin or the enamel lead to

	visualize and assess later in life, in childhood or even adulthood, the presence of developmental dental anomalies such as anomalies of tooth number, shape, size, structure, eruption or resorption. These abnormalities most often exist in syndromes but may appear isolated due to the difficulty to associate them to other sometimes-discrete clinical signs. The main objective of this collaborative project is to improve our diagnostic capacity, our expertise in the clinical management of our patients affected by rare oro-dental diseases and to be able to correctly perform the phenotype/genotype association of these conditions. Thanks to an exchange of clinical practices and research approaches around these rare conditions in patients at the national and international level, we will increase our expertise in the field and improve personalized approaches in the care of patients affected by rare oral and dental diseases.
Personal statement	With this experience, I acquired new knowledge about the multidisciplinary treatment of patients with craniofacial malformations such as Treacher Collins Syndrome, Craniosynostosis, and several cases of cleft lip-palate syndromes and severe oligodontia. I was able to compare through this experience, at the different stages of the patients' diagnosis and treatment pathway, the clinical guidelines used by the Dutch practitioners and compare them to our practices in France. I was able to learn in detail about the social security and some specific issues in another European country. Several collaborative research projects and ideas were born during my stay, in order to improve not only our clinical practices, but also to better understand the pathophysiology of these diseases and various genetic mutations at the research level. Finally, we have improved the collaboration in between two main reference centers for oro-dental rare diseases of the ERN CRANIO and we intend to work in new European clinical guidelines in collaboration of two active countries at the ERN.