European Reference Networks
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The *FUZ* gene in craniosynostosis: from a mouse knock-out model to patient-specific *in vitro* disease modelling.

Duration	8 months (6 months funding by EJP-RD)
Short Bio	I am a developmental biologist from Porto, Portugal. After concluding my MSc in Neurosciences at King's College of London, I have now undertaken a PhD position at Erasmus Medical Center, in Rotterdam, The Netherlands. I am researching a rare congenital condition called craniosynostosis, characterised by the premature fusion of the cranial sutures. Looking ahead, I aspire to contribute significantly to the improvement of personalised therapies and the field of developmental biology, advancing our understanding of the fundamental processes that lead to craniosynostosis. I am driven by the belief that scientific curiosity has the power to change lives and push the boundaries of knowledge.
Home Institution	Erasmus Medical Center, Rotterdam, The Netherlands
Host Institution	King's College of London, London, United Kingdom
Project Description	*FUZ* is a gene still poorly understood in the context of cranial suture development. We know that mutations in FUZ can lead to craniosynostosis. Understanding how FUZ is regulated is essential to select the best therapeutic approach to these patients. In vivo and in vitro disease models often work complementarily to the understanding of a disease. The patient-derived hiPSCs model, that we have developed in Rotterdam, constitutes a powerful tool to model cellular functions and test for therapies. Yet, we still need to understand how the gene FUZ, the patient-specific mutation and the therapeutic strategies perform in vivo. Along with the Fuz knock-out mouse model developed by Professor Karen Liu's lab, we aim to bring clarity to the fundamental role of FUZ in human development and in the aetiology of craniosynostosis.
Personal Statement	The bonds I've created in London, with several members of the scientific community, have not only expanded my career opportunities but also my passion for the work that we do. I look forward to continue meeting great minds like I did in King's College of London and here, at the Erasmus Medical Center. Ultimately, we all work synergistically to contribute to the development of medical research and improve children with rare condition's lifestyle, so they may live longer and healthier lives.