
THE ROLE OF ZIC1 IN CRANIAL SUTURE FORMATION

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The *zic1* gene plays an important role in embryonic development, in part by regulating the expression of many other genes including the engrailed gene. Previous investigators have reported that abnormal engrailed expression shifts the location of cranial suture formation and affects gene expression in the developing sutures (Deckelbaum et al. 2012). Such defects may cause a premature fusion of cranial sutures, leading to a serious birth defect known as craniosynostosis. Dr. Andrew Wilkie (Oxford University) has found that mutations in the human ZIC1 gene cause craniosynostosis. He hypothesizes that the engrailed gene is abnormally regulated in patients with these ZIC1 mutations. In collaboration with the Wilkie lab, we are testing this hypothesis by injecting RNA derived from the human ZIC1 mutants into *Xenopus* frog embryos. The goal of our experiments is to observe whether the mutated human ZIC1 genes affect the expression of the engrailed gene in frog embryos, which we were able to show by in situ hybridization. The degree of abnormality of engrailed expression caused by the various human ZIC1 mutations corresponds to the severity of the patients' phenotypes. These findings provide a better understanding of the molecular mechanisms underlying craniosynostosis and suggest possible gene regulatory pathways.