Genetic causes of craniofacial anomalies

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About 40% of craniosynostosis is caused by a genetic defect, but in only 25% of the cases this change can be detected. The most commonly found genetic changes occur in the fibroblast growth factor receptor 2 and 3 genes and in TWIST1 gene. In collaboration with professor Wilkie from Oxford, we have identified a number of new causal genes in craniosynostosis, such as the EFNB1 and TCF12 genes. In addition, other genes involved in rare craniofacial malformations have been discovered; ALX3 and RAB23. Given the high number of unsolved cases, we continue our persuit to find the genetic causes by using new techniques such as whole genome sequencing.