University of California, Irvine
Study Information Sheet

Clinical Genetics of Craniosynostosis

Lead Researcher

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Study Location(s):
UCI GCRC

- We are currently recruiting families with a child with craniosynostosis (premature closure of the cranial sutures)

  Craniosynostosis, the premature fusion of one or more skull sutures, is a variable condition occurring in 1 out of 2,000 live births. This research is being done to find out what causes defects of the face and the skull that occurred prior to birth. Studies of family members who do not have such abnormalities will aid in determining which factors may or may not be related to these problems. The researchers will perform genetic studies to identify genes associated with craniosynostosis.

- The research procedures involves:
  The research involves blood drawing, cheek (buccal) swabs or mouthwash for DNA sampling, physical examination and measurements, review of medical record and digital photographs,

- The only foreseeable discomfort associated with the study is the invasion of your privacy. There are no direct benefits from participation in the study. However, this study may explain the cause for your child's craniosynostosis in some cases which may allow for a better description of the syndrome and genetic counseling.

  There are no other direct benefits to participants from being in this study. However, we hope that the information obtained may help us in the development of accurate and efficient diagnostic

testing and better treatment strategies of people with these conditions may potentially benefit the subjects in the future. Benefits for society are potentially great if the scientific and health communities gain information about the syndrome genes, their function as they relate to normal craniofacial development and the pathogenesis of these conditions.

If interested please contact Dr Virginia Kimonis at vkimonis@uci.edu