The study will:

- Help us learn more about the genetic causes of limb malformations
- Lead to improved patient counseling
- Aid in the development of genetic tests
- Increase our understanding of limb development and limb malformations
- Help inform individuals about how limb malformations are passed on to future generations

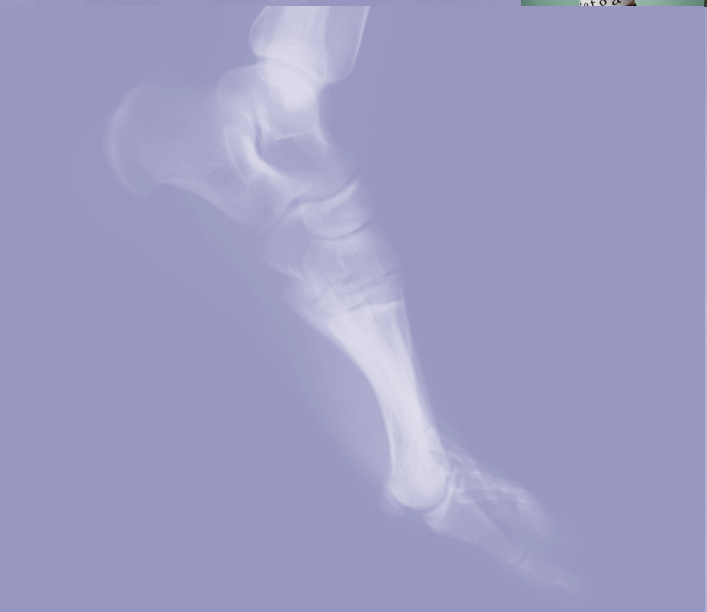
a study to identify

UCSF Limb Study

the genetic causes of limb malformations

Please help us learn more about the genetic causes of limb malformations!

If you are interested in participating in our study or have any questions, please contact:



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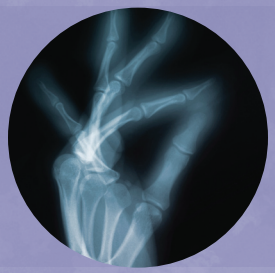
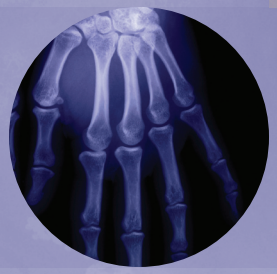
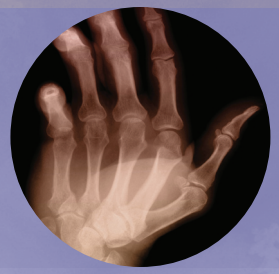
Institute for Human Genetics

We are looking for individuals who have limb malformations such as:

- Syndactyly (fused or webbed fingers/toes)
- Polydactyly (> 5 fingers/toes)
- Split hand and foot
- Reduction anomalies (< 5 fingers/toes)
- Brachydactlyly (short finger/toes)
- Clinodactyly (bent pinky fingers)
- Arachnodactyly (extra long fingers)
- Camptodactyly (bent little finger)

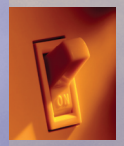






- Club feet
- Congenital radioulnar synostosis
- Congenital radial head dislocation

New advances in genetics have greatly increased our understanding of how mutations can cause isolated limb malformations – limb malformations that occur separately from any other clinical problems.



Many studies suggest that mutations responsible for isolated limb malformations can occur in regions that regulate genes that are important for limb development. These regions are called “enhancers” and act like light switches to activate genes at the right time and place. If there is a mutation in the enhancer, the gene is not turned “on” properly.

Our research is focused on identifying enhancers that are important for limb development and identifying mutations in these enhancers that may be responsible for isolated limb malformations.

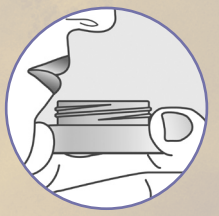
What is required of study participants?

1. Brief informed consent interview
2. Short questionnaire
3. DNA sample collected from blood or saliva
   - We will provide a kit for saliva collection that can be returned to us in a prepaid envelope

Patient privacy is very improtant to us and all information and samples are kept confidential

Collection of saliva for a DNA sample is easy, painless and can be done in the participant’s home. Participants do not need to travel to UCSF.

For adults



Collect saliva in the cup provided

For infants & young children

Use the sponges provided to collect saliva