

LETTER OF INVITATION TO PARTICIPATE IN RESEARCH

I would like to invite you to participate in a research study on the biological basis of 15q Duplication Syndrome. **For this study we are interested in individuals diagnosed with 15q Duplication Syndrome as well as one or both of their unaffected parents.** We are contacting you because you have been seen at Stanford Children's health related to your diagnosis of 15q Duplication Syndrome. *If you have received this letter in error and were not diagnosed with this condition, please accept our sincerest apologies and discard the letter.* If you feel it is appropriate, please feel free to pass this information on to other families you know.

Researchers at the Stanford University School of Medicine are conducting a research study to determine the core mechanisms of 15q Duplication Syndrome and identify therapeutic targets. We aim to do this by collecting skin and/or blood cells from individuals with 15q Duplication Syndrome and converting them to induced pluripotent progenitor (iPS) cells to study cellular phenotypes.

Participation in this study requires the following one-time assessments to be done at no charge to you or your insurance:

- Travel to Stanford University Medical Center in Palo Alto, California
- A blood draw (5-10ml) and/or skin biopsy will be collected.
- Medical records including basic demographic information (age, sex, ethnicity) will be collected.

These assessments can be completed in less than 1 hour. No follow up will be conducted. Sample collection will pose minimal risk. Private health information will be de-identified, and will remain private. Participants will not receive any direct benefit from partaking in the study.

In general, to be eligible to participate in this study, the individual with 15q Duplication Syndrome must be:

- Between the ages of 1 year – 18 years old.
- Have a confirmed genetic diagnosis of 15q duplication syndrome and associated disorders (including but not limited to isodicentric chromosome 15, interstitial duplication of chromosome 15 and 15q11.2 microduplications).

Parents who wish to participate in this study may be of any age.

If you are interested or have any questions, please don't hesitate to call or email:

Elijah Kravets
e: ekravets@stanford.edu; Tel: 650-724-1881

Sincerely,

Dr. Jonathan Bernstein
Assistant Professor of Pediatrics
Stanford University School of Medicine
300 Pasteur Drive
Stanford, CA 94305
Web: medicalgenetics.stanford.edu

For general information about participant rights in studies conducted at the Stanford University School of Medicine, contact 1-866-680-2906