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Department of Human Genetics
Division of Medical Genetics
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Subtelomere Study Information Sheet

The aim of this study is to understand why the ends of chromosomes break. Rearrangements in the ends of chromosomes, also called subtelomeres, can cause birth defects, intellectual disability, and/or developmental delays. Between 3-6% of children with idiopathic intellectual disability have a subtelomeric rearrangement. Despite the prevalence of this type of chromosome rearrangement, the causes of subtelomeric rearrangements remain unknown. Our study uses microarrays to look very closely at the part of the subtelomere that rearranged to understand why that DNA broke.

We are informing parents about this study if they have a child with a previously identified subtelomeric rearrangement. We would like these parents to know about the study and offer them the opportunity to participate if they wish. Subtelomeric rearrangements are typically detected by tests performed in a Cytogenetics lab such as Fluorescence *In Situ* Hybridization (FISH) or Array Comparative Genome Hybridization (CGH). All chromosome ends are subject to breaks, and many children with rare chromosome disorders have a subtelomeric rearrangement.

The study is organized by Dr. Katie Rudd of Emory University School of Medicine in Atlanta, Georgia, USA. Dr. Rudd is a board-certified clinical Cytogeneticist who studies the mechanism of chromosome rearrangements. This study is funded by the March of Dimes and has been reviewed by the Emory University Institutional Review Board (IRB).

How to contact us to find out more about the study

If you would like to find out more about the study (even if you then decide not to take part) please contact Dr. Katie Rudd, at the address/phone number below:

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