



Characterizing Cognition and Behavior Phenotype Across Genotype In Cornelia De Lange Syndrome (CdLS)

What: The purpose of this study is to better understand the cognitive and behavioral profiles of CdLS, and if these differ based on genotype (i.e., the specific variant causing the syndrome).

First, the study involves signing a consent form to agree to participate in the research. Once that is done, a caregiver will be sent a series of questionnaires that can be completed remotely on a computer. These remote inventories will have questions about the developmental history, cognition, and behavior functioning of the person with CdLS. The second part of the study involves virtual cognitive testing for about 90 minutes, which can be scheduled into 1-2 sessions based on the participant's preference. The individual with CdLS will complete an abbreviated test battery of cognitive measures that assess basic language and non-verbal skills. All individuals with CdLS, age 10 years and older, are eligible to enroll in the first part of the study (remote questionnaires). Those with a genetic variant (NIPBL, SMC1A, HDAC8) can enroll in the remote cognitive testing portion.

Next, a member of the study team may reach out to follow up via telephone/video conference.

Who: Any individual genetically diagnosed with CdLS based on clinical presentation or genetic testing (i.e. pathogenic variant in NIPBL or HDAC8; or a pathogenic missense variant in SMC1A)

Where: The study will take place remotely. All components can be completed on a laptop/computer without the need to visit our site in Baltimore. However, local families may opt to complete testing on site based on preference.

Why: Our group and our collaborators are extremely interested in treating patients with CdLS as well as studying ways to potentially treat this condition. The relationship between cognitive and behavior development across the different genes causing CdLS is still unknown. Studying the relationship between genotype and behaviors can help us consider ways to personalize treatment approaches across individuals with CdLS. Further developments in these areas will help guide future research trials and treatments.

When: The study is currently ongoing.

How: If interested in participating or if there are further questions, please reach out to

Dr. Rowena Ng: ngr@kennedykrieger.org

Phone: 443-923-4456