

Developmental and Behavioral Characteristics of Children with Dup15q Syndrome: Updates from a Large Cohort Study

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This talk will present data from a large cohort study of children with Dup15q Syndrome. Comprehensive phenotyping was conducted with 60 participants (ages 3-18), at the UCLA Dup15q Clinic as well as at two Dup15q Alliance Family Conferences. The majority of participants (73%) had isodicentric duplications, and 43% had epilepsy. Assessments domains included cognitive, language, social communication, autism characteristics, motor skills and adaptive behavior. For children with epilepsy, we also collected detailed information about seizure types, seizure severity and anti-epileptic medications.

Across all participants, there was a wide range of abilities. Both duplication type (interstitial or isodicentric) and epilepsy had a large impact on development across cognitive, language, motor and adaptive domains. Children with isodicentric duplications and epilepsy showed the greatest level of impairment, while children with interstitial duplications showed the least. Additionally, parents reported more elevated concerns regarding social withdrawal, stereotyped behavior and hyperactivity in children with isodicentric duplications compared with interstitial.

In addition to findings from the current study, we will also discuss best practices for conducting assessments with this population, potential avenues for intervention, and future research directions.