**Title:** The Diagnostic Journey: Results From a Standardized Interview of Parents of Children with Neurodevelopmental Disorders and Associated Genetic Syndromes

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**Introduction:** Established medical practice parameters for genetic testing in Neurodevelopmental Disorders (NDDs) has led to the identification of numerous copy number variations and single gene disorders associated with these conditions (Schaefer et al, 2013; Duffourd et al, 2016; Sawyer et al, 2016). However, the rapid and detailed identification of genetic etiologies of NDDs has far outpaced our ability to provide timely medical counseling, guidance, and care. Families continue to face barriers in access to informed diagnostic evaluations and clinical management (Basel & McCarrier, 2016). In response to these barriers, our team at UCLA, through the Center for Autism Research and Treatment, has established a Developmental Neurogenetics Clinic (DNG) to provide comprehensive multidisciplinary care to children and adults with a NDD and a known or suspected genetic etiology. As part of this clinical program, we developed a comprehensive diagnostic journey interview (DJI) to identify the major barriers in access to care and gaps in subsequent medical support after a genetic diagnosis is made. This study reviews emerging themes from our collected data.

**Method:** The medical team, consisting of pediatric neurology, pediatric genetics, and child psychiatry developed the DJI based on previously developed interviews and clinical experience. Families were interviewed prior to their first appointment at the DNG clinic. A total of 25 families from DNG clinic have completed the DJI. The DJI includes both multiple choice and open ended questions regarding the neurodevelopmental diagnosis and the genetic diagnosis, with themes including time from initial concern to first clinical diagnoses, medical counseling and access to services after clinical diagnoses, and the overall impressions of the diagnostic odyssey.

**Results:** Individuals included in the DJI ranged from 2-30 years of age. Genetic diagnoses included, but were not limited to, mutations in the KMT2D gene (Kabuki syndrome), Chromosome 15q11.2-13.1 duplication syndrome, 1p36 deletion syndrome, FOXG1 syndrome, and 16p11.2 duplication. 70% of families had developmental concerns for their child in the first year of life. Greater than 70% of families noted there was no available medical evaluation for their specific concerns in their geographic area. 57% of families noted a considerable wait time to see a geneticist once referred by a primary care provider. If genetic testing was obtained prior to referral to a specialist, 80% of families reported minimal genetic counseling when results were given. After referral to a specialist, families noted that the most useful resources and informative tools were provided by the specializing health care provider (86%) and patient advocacy groups (57%). Individual families also reported the following gaps in care: (1) genetic report provided with no further counseling, (2) primary care providers did not always address developmental concerns with appropriate referral to specialized practice provider, and (3) although there was initial “fear and anxiety” in awaiting the results of the genetic testing, ultimately having the results was “worth it”.

**Discussion:** The DJI highlights several key themes in the diagnostic and clinical care process for children with genetic syndromes and NDDs, with gaps in the timing of diagnoses, access to resources after diagnosis, and communication around the implications of the genetic diagnosis. Data from this semi-qualitative interview will facilitate the development of best practice guidelines in clinical care, and support of families of the many children with new genetic diagnoses in the context of NDDs.

**References/Citations:**