



Abstract Presented At the 2006 Annual Meeting of the American Epilepsy Society
December 1-5, 2006

Title: Dravet's Syndrome: Expanding the Clinical Spectrum

Authors: ¹Joan V. Skluzacek, and ²Beverly S. Wical

Session: Sunday 12/03/06, 7:30am - 4:30pm, Poster Number: 2.090

Content: RATIONALE:

Dravet syndrome is a devastating epileptic syndrome with onset very early in childhood. Major progress has been made in understanding the genetic foundation of the disorder; yet much work remains to identify co-morbid conditions commonly associated with Dravet's. Appropriate care of affected children must include identification and management of these disorders to permit optimal health and enhance our understanding of the disorder.

METHODS:

Dravet syndrome is a rare disorder that makes systematic study of associated conditions difficult. The Dravet syndrome/SMEI Group, a support group for parents with affected children, is associated with the International Dravet syndrome Epilepsy Action League (IDEA League). At the time of data collection in 2005, 65 families participated in surveys. Not all families answered each question. Participation was voluntary. Questions were chosen by group leaders to investigate whether other children had been affected by problems similar to their own. Questions were asked regarding: immune dysregulation and susceptibility to infections; growth, metabolic or endocrine disorders; and autism spectrum disorder. Despite the uncontrolled nature of data collection, trends emerged that are suggestive of significant co-morbid conditions occurring in the majority of children with Dravet's.

RESULTS:

Of 61 respondents, 46 (75%) reported their children are prone to infection. Recurring/chronic otitis media, sinusitis, or bronchitis occurred in 50%. Fourteen of the 61 (23%) had been assessed by an immunologist, allergist, or infectious disease specialist. No child evaluated was found to have any specific immune deficiency. Thirty-eight of 61 (61%) reported their children had been assessed for endocrine, metabolic or absorption disorders. Mitochondrial disorders NOS had been diagnosed in 4/61 (6%). All 61 reported their children have difficulty regulating body temperature. The majority of children (38/61--61%) are underweight. Autism spectrum disorder was of concern to 35 (78%) of the 44 that responded to questions about behaviors. Autism has been diagnosed in 24% of these children. An additional 49% of children have autistic traits. 100% have communication disorders.

CONCLUSIONS:

Intractable epilepsy, significant cognitive impairment, and movement abnormalities are well recognized in Dravet's Syndrome. Our preliminary data suggest aspects of the syndrome go well beyond these features. Careful investigation of associated disorders in immune regulation and infection is needed. Documentation of pattern of infections is needed to separate out a true increased incidence from an apparent increase (due to increased awareness of infection as seizure trigger). Growth and nutritional assessment should be part of each child's coordinated care. Specific evaluation for autism spectrum disorders seems indicated. Associated symptoms must be sought in a prospective way to facilitate understanding of the manifestations of this complex disorder.