

Expanding the Clinical Spectrum: Updated Data on Comorbidities in Dravet Syndrome

Joan V. Skluzacek, BA; Kathryn P. Watts, MSc; Susan E. Goodliffe, BA;
Olivier Parsy; Karen Glenn; Angela P. Black, MD

RATIONALE:

Dravet syndrome is a pernicious epilepsy syndrome with onset in infancy generally characterized by intractable seizures, developmental impairments, and movement abnormalities. Up to 80% of clinically identified cases test positive for mutations of the SCN1A gene that result in the dysfunction of voltage-gated sodium channels in neurons (Dravet, Bureau, and Oguni). In 2005, parents participating in an online Family Forum and support group for families of children with Dravet syndrome now affiliated with the IDEA League (International Dravet syndrome Epilepsy Action League), gathered and presented preliminary data which indicates that the problems of these children are more extensive than this (Skluzacek and Wical). In September of 2009, they followed up with collection of updated data from the group in order to compare it with the 2005 results and see what more can be learned.

METHODS:

The IDEA League is a partnership of parents and professionals united in the purpose of creating greater awareness and understanding of Dravet syndrome. In 2005, parents in the IDEA League Family Forum had the opportunity to participate in surveys about their child's condition. Questions were chosen by group leaders to investigate if other children had been affected by problems similar to their own. The surveys took place over a three month period of time. Participation was voluntary and 65 of the 103 families with memberships chose to participate. In 2009, the process was repeated. Most of the previous survey questions were asked again, as well as some new ones. In addition, a survey was included asking about the various medical specialist involved in their child's care. This time the surveys took place within a two week period of time. Participation was voluntary, and 86 of the 608 families with memberships chose to participate.

RESULTS:

The parents of 81 children with Dravet syndrome completed the survey regarding the types of medical specialists that have been involved in or consulted regarding their child's care. Collectively they identified 29 different types of medical specialists that have had a role in the care of these children. As expected, between 50% and 100% of these families have consulted or been in the care of a primary general pediatrician, a neurologist or epileptologist, a dietician (ketogenic diet), a geneticist; speech, physical and occupational therapists; and a specialist in complementary medicine. More than a third of these children have also seen an endocrinologist or metabolic specialist, an otolaryngologist, an orthopedist, a cardiologist and an intensivist. In addition, about a quarter of the families surveyed have also seen an immunologist, an allergist, a developmental or neuro- ophthalmologist, a developmental or special needs dentist, a sleep specialist, and a gastroenterologist.

As in 2005, concerns emerged again about autism spectrum characteristics, including developmental regression, behavior, language and communication disorder, and sensory integration disorders. Concerns about chronic infections and immunity also re-emerged, especially regarding infections of the respiratory tract, but also gastritis and urinary tract infections. Problems in the areas of orthopedic function; growth, metabolism and nutrition, and sleep were also repeating concerns. A newly emerging area of concern relates to dental health, with a number of families reporting delayed tooth eruption, malocclusions, and more. In addition, there are concerns beginning to emerge regarding cardiac function.

MEDICAL SPECIALISTS INVOLVED IN THE CARE OF CHILDREN WITH DRAVET SYNDROME ON THE IDEA LEAGUE FAMILY FORUM

1	General Pediatrician	100%
2	Developmental Pediatrician	2%
3	Neurologist/Epileptologist	100%
4	Endocrinologist/metabolic Specialist	38%
5	Immunologist	22%
6	Infectious Disease Specialist	12%
7	Allergist	23%
8	Otolaryngologist (ENT)/Audiologist	43%
9	Ophthalmologist	4%
10	Developmental or Neuro- Ophthalmologist	28%
11	Developmental/Special Needs Dentist	27%
12	Sleep Specialist	20%
13	Developmental or Neuro- Psychiatrist/Psychologist	49%
14	Physiatrist/Specialist in Physical medicine	7%
15	Physical Therapist	80%
16	Occupational Therapist	79%
17	Speech Pathologist	89%
18	Orthopaedist/Orthotist	41%
19	Podiatrist	2%
20	Dermatologist	3%
21	Cardiologist	35%
22	Pulmonologist	11%
23	Geneticist	63%
24	Emergency Medicine Specialist	32%
25	Intensivist (Critical Care Specialist)	38%
26	Gastroenterologist	32%
27	Urologist	14%
28	Dietician	54%
29	Specialist in Complementary Medicine (Osteopath, Chiropractor, Nutritionist, Massage Therapist, etc.)	46%

CONCLUSIONS:

Though this data has been gathered through an informal process, which was not scientifically rigorous or controlled, it does indicate the need for further study of co-morbid conditions in Dravet syndrome. It seems clear that Dravet syndrome is a systemic disorder requiring integrated care of the whole child. Coordinated comprehensive care is essential to improving the outcome of children with Dravet syndrome and should include regular assessments of development, with specific evaluation for sensory integration and autism spectrum disorders; growth, metabolic and nutritional assessment; assessment of immune function, orthopedic evaluation, and sleep assessments. In addition, it appears that the care plan for a patient with Dravet syndrome should include appropriate dental evaluations and care, and may need to include assessments of cardiac function.

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